

# CITATION REPORT

List of articles citing

## Global variation in copy number in the human genome

DOI: 10.1038/nature05329  
Nature, 2006, 444, 444-54.

**Source:** <https://exaly.com/paper-pdf/40444663/citation-report.pdf>

**Version:** 2024-04-25

This report has been generated based on the citations recorded by exaly.com for the above article. For the latest version of this publication list, visit the link given above.

The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

#	Paper	IF	Citations
2255	β-THALASSEMIA. 239-240		5
2254	Ionization Produced in Gaseous Reactions. <b>1925</b> , 11, 512-4		1
2253	Changes in residual volume following oxygen breathing. <b>1983</b> , 55, 817-24		3
2252	Real-world driving speciated VOC hot start emissions of the 2001 French passenger car fleet. <b>2001</b> , 27, 140		1
2251	Human genomics: in search of normality. <i>Nature</i> , <b>2006</b> , 444, 428-9	50.4	23
2250	Genome assembly comparison identifies structural variants in the human genome. <b>2006</b> , 38, 1413-8		133
2249	Accurate and reliable high-throughput detection of copy number variation in the human genome. <b>2006</b> , 16, 1566-74		122
2248	Genome-wide detection of human copy number variations using high-density DNA oligonucleotide arrays. <b>2006</b> , 16, 1575-84		156
2247	Forensic DNA and bioinformatics. <b>2007</b> , 8, 117-28		14
2246	Genome-wide association studies of cancer. <b>2007</b> , 3, 419-27		10
2245	Characterization of a recurrent 15q24 microdeletion syndrome. <b>2007</b> , 16, 567-72		159
2244	Modeling the monosomy for the telomeric part of human chromosome 21 reveals haploinsufficient genes modulating the inflammatory and airway responses. <b>2007</b> , 16, 2040-52		29
2243	Molecular cytogenetics: making it safe for human embryonic stem cells to enter the clinic. <b>2007</b> , 7, 395-406		7
2242	Sensitive and specific real-time polymerase chain reaction assays to accurately determine copy number variations (CNVs) of human complement C4A, C4B, C4-long, C4-short, and RCCX modules: elucidation of C4 CNVs in 50 consanguineous subjects with defined HLA genotypes. <b>2007</b> , 179, 3012-25		64
2241	The drifting human genome. <b>2007</b> , 104, 20147-20148		12
2240	Systematic prediction and validation of breakpoints associated with copy-number variants in the human genome. <b>2007</b> , 104, 10110-5		70
2239	Whole-genome profiling in liposarcomas reveals genetic alterations common to specific telomere maintenance mechanisms. <b>2007</b> , 67, 9221-8		19

2238	HtrA2 regulates beta-amyloid precursor protein (APP) metabolism through endoplasmic reticulum-associated degradation. <b>2007</b> , 282, 28285-95	48
2237	A newly recognised microdeletion syndrome involving 2p15p16.1: narrowing down the critical region by adding another patient detected by genome wide tiling path array comparative genomic hybridisation analysis. <b>2008</b> , 45, 122-4	35
2236	Array CGH analysis of copy number variation identifies 1284 new genes variant in healthy white males: implications for association studies of complex diseases. <b>2007</b> , 16, 2783-94	191
2235	Copy number variation in the human genome and its implications for cardiovascular disease. <b>2007</b> , 115, 3130-8	61
2234	Autism: the quest for the genes. <b>2007</b> , 9, 1-15	34
2233	Genetics of the cardiometabolic syndrome: new insights and therapeutic implications. <b>2007</b> , 1, 37-47	12
2232	The new mutation theory of phenotypic evolution. <b>2007</b> , 104, 12235-42	161
2231	Molecular karyotyping in patients with mental retardation using 100K single-nucleotide polymorphism arrays. <b>2007</b> , 44, 629-36	66
2230	Extreme context specificity in differential allelic expression. <b>2007</b> , 16, 537-46	45
2229	Genetic determinants of plasma lipoproteins. <b>2007</b> , 4, 600-9	14
2228	Evolution of the vertebrate twist family and synfunctionalization: a mechanism for differential gene loss through merging of expression domains. <b>2007</b> , 24, 1912-25	34
2227	Oligonucleotide arrays for high-resolution analysis of copy number alteration in mental retardation/multiple congenital anomalies. <b>2007</b> , 9, 617-25	36
2226	Copy-number variation in control population cohorts. <b>2007</b> , 16 Spec No. 2, R168-73	186
2225	Flexible and accurate detection of genomic copy-number changes from aCGH. <b>2007</b> , 3, e122	39
2224	Functional effects of nonsynonymous polymorphisms in the human TRPV1 gene. <b>2007</b> , 293, F1865-76	46
2223	The diploid genome sequence of an individual human. <b>2007</b> , 5, e254	1249
2222	A high-resolution map of segmental DNA copy number variation in the mouse genome. <b>2007</b> , 3, e3	178
2221	Population stratification of a common APOBEC gene deletion polymorphism. <b>2007</b> , 3, e63	183

2220	Copy number variants and common disorders: filling the gaps and exploring complexity in genome-wide association studies. <b>2007</b> , 3, 1787-99	157
2219	Hybridize and personalize: the new age of syndromal mental retardation diagnostics. <b>2007</b> , 68, 721-2	4
2218	General aspects of the genetics of SLE. <b>2007</b> , 40, 550-9	28
2217	Identification of novel candidate genes associated with cleft lip and palate using array comparative genomic hybridisation. <b>2008</b> , 45, 81-6	76
2216	Genomic deletion size at the epsilon-sarcoglycan locus determines the clinical phenotype. <b>2007</b> , 130, 2736-45	68
2215	A genome-wide scan for genes involved in primary vesicoureteric reflux. <b>2007</b> , 44, 710-7	32
2214	Array-based comparative genomic hybridization: clinical contexts for targeted and whole-genome designs. <b>2007</b> , 9, 553-9	37
2213	Catch me if you can: tracking down the genetic origins of congenital heart disease. <b>2007</b> , 28, 2701-2	
2212	Copy-number variants in patients with a strong family history of pancreatic cancer. <b>2007</b> , 6, 1592-9	33
2211	Molecular windows into speech and language disorders. <b>2007</b> , 59, 130-40	11
2210	Copy-number variations add a new layer of complexity in the human genome. <b>2007</b> , 176, 441-2	5
2209	High resolution array-CGH analysis of single cells. <b>2007</b> , 35, e15	120
2208	Structural variation in the human genome. <b>2007</b> , 356, 1169-71	105
2207	A novel syndrome of cerebral cavernous malformation and Greig cephalopolysyndactyly. Laboratory investigation. <b>2007</b> , 107, 495-9	4
2206	Chapter 1: Introduction. <b>2007</b> , 14, S2-S113	
2205	ATCG nucleotide fluctuation of <i>Deinococcus radiodurans</i> radiation genes. <b>2007</b> , 6694, 402	9
2204	Structural variation in the human genome: the impact of copy number variants on clinical diagnosis. <b>2007</b> , 9, 600-6	63
2203	Genomic drift and copy number variation of sensory receptor genes in humans. <b>2007</b> , 104, 20421-6	122

2202	Genotype-specific weight loss treatment advice: how close are we?. <b>2007</b> , 32, 351-66	11
2201	The neutral coalescent process for recent gene duplications and copy-number variants. <b>2007</b> , 177, 987-1000	26
2200	MLGA--a rapid and cost-efficient assay for gene copy-number analysis. <b>2007</b> , 35, e115	21
2199	Genetic susceptibility to peripheral arterial disease: a dark corner in vascular biology. <b>2007</b> , 27, 2068-78	49
2198	Rapid evolution of an X-linked microRNA cluster in primates. <b>2007</b> , 17, 612-7	126
2197	Positive selection at the protein network periphery: evaluation in terms of structural constraints and cellular context. <b>2007</b> , 104, 20274-9	111
2196	Runs of homozygosity reveal highly penetrant recessive loci in schizophrenia. <b>2007</b> , 104, 19942-7	282
2195	The UCSC Genome Browser Database: 2008 update. <b>2008</b> , 36, D773-9	423
2194	Language and genes: a new perspective on the origins of human cultural diversity. <b>2007</b> , 104, 10755-6	12
2193	Tiling resolution array comparative genomic hybridization, expression and methylation analyses of dup(1q) in Burkitt lymphomas and pediatric high hyperdiploid acute lymphoblastic leukemias reveal clustered near-centromeric breakpoints and overexpression of genes in 1q22-32.3. <b>2007</b> , 16, 2215-25	41
2192	Significant gene content variation characterizes the genomes of inbred mouse strains. <b>2007</b> , 17, 1743-54	81
2191	A portrait of copy-number polymorphism in <i>Drosophila melanogaster</i> . <b>2007</b> , 104, 19920-5	118
2190	The C20orf133 gene is disrupted in a patient with Kabuki syndrome. <b>2007</b> , 44, 562-9	42
2189	Genetic association of CTNNA3 with late-onset Alzheimer's disease in females. <b>2007</b> , 16, 2854-69	48
2188	A patient with vertebral, cognitive and behavioural abnormalities and a de novo deletion of NRXN1alpha. <b>2008</b> , 45, 239-43	104
2187	Genetic Profiling: Ethical Constraints upon Criminal Investigation Procedures. <b>2007</b> , 3, 236-252	
2186	Recurrent DNA inversion rearrangements in the human genome. <b>2007</b> , 104, 6099-106	70
2185	High-resolution single nucleotide polymorphism array analysis of epithelial ovarian cancer reveals numerous microdeletions and amplifications. <b>2007</b> , 13, 4731-9	135

2184	Development of a focused oligonucleotide-array comparative genomic hybridization chip for clinical diagnosis of genomic imbalance. <b>2007</b> , 53, 2051-9	48
2183	Investigation of human protein variants and their frequency in the general population. <b>2007</b> , 6, 1183-7	46
2182	Integrated profiling of basal and luminal breast cancers. <b>2007</b> , 67, 11565-75	232
2181	Pyrosequencing for discovery and analysis of DNA sequence variations. <b>2007</b> , 8, 1437-41	29
2180	Whole-genome array-CGH for detection of submicroscopic chromosomal imbalances in children with mental retardation. <b>2007</b> , 118, 1-7	29
2179	Intragenic breakpoints localized by array CGH in a t(2;6) familial translocation. <b>2007</b> , 119, 185-90	4
2178	Processes of de novo duplication of human alpha-globin genes. <b>2007</b> , 104, 10950-5	55
2177	QuantiSNP: an Objective Bayes Hidden-Markov Model to detect and accurately map copy number variation using SNP genotyping data. <b>2007</b> , 35, 2013-25	462
2176	Modeling recurrent DNA copy number alterations in array CGH data. <b>2007</b> , 23, i450-8	50
2175	A periodic pattern of SNPs in the human genome. <b>2007</b> , 17, 1414-9	6
2174	Array comparative genomic hybridization and computational genome annotation in constitutional cytogenetics: suggesting candidate genes for novel submicroscopic chromosomal imbalance syndromes. <b>2007</b> , 9, 642-9	14
2173	Copy Number Variation in the Human Genome. <b>2007</b> ,	
2172	The evolution of molecular ruler analysis for characterizing telomere imbalances: from fluorescence in situ hybridization to array comparative genomic hybridization. <b>2007</b> , 9, 566-73	11
2171	Comprehensive validation of array comparative genomic hybridization platforms: how much is enough?. <b>2007</b> , 9, 632-41	10
2170	Identifying cis- and trans-acting single-nucleotide polymorphisms controlling lymphocyte gene expression in humans. <b>2007</b> , 1 Suppl 1, S7	6
2169	Derivation of Human Embryonic Stem Cells. 35-51	2
2168	High-resolution genomic microarrays for X-linked mental retardation. <b>2007</b> , 9, 560-5	16
2167	HIF-1 regulates heritable variation and allele expression phenotypes of the macrophage immune response gene SLC11A1 from a Z-DNA forming microsatellite. <b>2007</b> , 110, 3039-48	60

2166	The C-MYB locus is involved in chromosomal translocation and genomic duplications in human T-cell acute leukemia (T-ALL), the translocation defining a new T-ALL subtype in very young children. <b>2007</b> , 110, 1251-61	214
2165	Molekulare Ursachen des Autismus. <b>2007</b> , 55, 647-648	
2164	Emerging Blueprint of Genome Suggests Structural Rearrangements May Explain Many Sporadic Cases of Disease. <b>2007</b> , 7, 46-47	
2163	Auf dem Weg zum Erbgut des Neandertalers. <b>2007</b> , 55, 146-148	0
2162	Host genetic variation and susceptibility to primate lentiviruses. <b>2007</b> , 1, 399-413	2
2161	Genomic copy number variation and its potential role in lipoprotein and metabolic phenotypes. <b>2007</b> , 18, 174-80	16
2160	Bibliography. Current world literature. Genetics and molecular biology. <b>2007</b> , 18, 199-221	
2159	Current status of genome-wide scanning for hypertension. <b>2007</b> , 22, 292-7	16
2158	Genetic and environmental factors in complex neurodevelopmental disorders. <b>2007</b> , 8, 429-44	73
2157	Personalised nutrition: status and perspectives. <b>2007</b> , 98, 26-31	61
2156	[DNA microarrays: technology and new insights in oncology]. <b>2007</b> , 28, 662-6	
2155	Co-inheritance of a novel deletion of the entire SPINK1 gene with a CFTR missense mutation (L997F) in a family with chronic pancreatitis. <b>2007</b> , 92, 168-75	23
2154	Female reproductive ageing: current knowledge and future trends. <b>2007</b> , 18, 58-65	283
2153	Introduction. <b>2007</b> , 17, 71-73	2
2152	Analyzing Gene Relationships for Down Syndrome with Labeled Transition Graphs. <b>2007</b> ,	0
2151	Transmitted cytogenetic abnormalities in patients with mental retardation: pathogenic or normal variants?. <b>2007</b> , 50, 243-55	38
2150	Dysregulation of cardiogenesis, cardiac conduction, and cell cycle in mice lacking miRNA-1-2. <b>2007</b> , 129, 303-17	1186
2149	A DNA replication mechanism for generating nonrecurrent rearrangements associated with genomic disorders. <b>2007</b> , 131, 1235-47	652

2148	Genetics of type 2 diabetes. <b>2007</b> , 17, 239-44	64
2147	Polymorphic miRNA-mediated gene regulation: contribution to phenotypic variation and disease. <b>2007</b> , 17, 166-76	122
2146	Use of array CGH in the evaluation of dysmorphology, malformations, developmental delay, and idiopathic mental retardation. <b>2007</b> , 17, 182-92	256
2145	Side effects of genome structural changes. <b>2007</b> , 17, 381-6	72
2144	Implementation of genetics to personalize medicine. <b>2007</b> , 4, 248-65	20
2143	Opposing functions of the T cell receptor kinase ZAP-70 in immunity and tolerance differentially titrate in response to nucleotide substitutions. <b>2007</b> , 27, 912-26	121
2142	MAPT gene duplications are not a cause of frontotemporal lobar degeneration. <b>2007</b> , 424, 61-5	14
2141	European guidelines on cardiovascular disease prevention in clinical practice: executive summary. <b>2007</b> , 194, 1-45	190
2140	Frequency assessment of SNPs for forensic identification in different populations. <b>2007</b> , 1, e1-3	6
2139	Characterization of Potocki-Lupski syndrome (dup(17)(p11.2p11.2)) and delineation of a dosage-sensitive critical interval that can convey an autism phenotype. <b>2007</b> , 80, 633-49	307
2138	Gene copy-number variation and associated polymorphisms of complement component C4 in human systemic lupus erythematosus (SLE): low copy number is a risk factor for and high copy number is a protective factor against SLE susceptibility in European Americans. <b>2007</b> , 80, 1037-54	364
2137	Cellular and clinical impact of haploinsufficiency for genes involved in ATR signaling. <b>2007</b> , 81, 77-86	57
2136	Reply to Lynch et al.. <b>2007</b> , 81, 420	
2135	Classification of human chromosome 21 gene-expression variations in Down syndrome: impact on disease phenotypes. <b>2007</b> , 81, 475-91	187
2134	A Bayesian approach to copy-number-polymorphism analysis in nuclear pedigrees. <b>2007</b> , 81, 808-12	13
2133	Simultaneous discovery and testing of deletions for disease association in SNP genotyping studies. <b>2007</b> , 81, 684-99	17
2132	Copy-number variations measured by single-nucleotide-polymorphism oligonucleotide arrays in patients with mental retardation. <b>2007</b> , 81, 768-79	106
2131	Impact of array comparative genomic hybridization-derived information on genetic counseling demonstrated by prenatal diagnosis of the TAR (thrombocytopenia-absent-radius) syndrome-associated microdeletion 1q21.1. <b>2007</b> , 81, 866-8	15



2130	Contribution of SHANK3 mutations to autism spectrum disorder. <b>2007</b> , 81, 1289-97	492
2129	Recurrent reciprocal genomic rearrangements of 17q12 are associated with renal disease, diabetes, and epilepsy. <b>2007</b> , 81, 1057-69	193
2128	Mendelian Inheritance in Man and its online version, OMIM. <b>2007</b> , 80, 588-604	489
2127	Identifying genes for coronary artery disease: An idea whose time has come. <b>2007</b> , 23 Suppl A, 7A-15A	24
2126	Gene copy number variation spanning 60 million years of human and primate evolution. <b>2007</b> , 17, 1266-77	127
2125	MLPA screening reveals novel subtelomeric rearrangements in holoprosencephaly. <b>2007</b> , 28, 1189-97	24
2124	Gene duplication: a drive for phenotypic diversity and cause of human disease. <b>2007</b> , 8, 17-35	172
2123	Functional genomics and schizophrenia: endophenotypes and mutant models. <b>2007</b> , 30, 365-99	38
2122	Relevance of Copy Number Variation to Human Genetic Disease. <b>2007</b> ,	
2121	High-resolution aCGH and expression profiling identifies a novel genomic subtype of ER negative breast cancer. <b>2007</b> , 8, R215	230
2120	Breaking the waves: improved detection of copy number variation from microarray-based comparative genomic hybridization. <b>2007</b> , 8, R228	110
2119	Functional constraint and small insertions and deletions in the ENCODE regions of the human genome. <b>2007</b> , 8, R180	30
2118	Identification of structural aberrations in cancer by SNP array analysis. <b>2007</b> , 8, 219	22
2117	Detailed analysis of 15q11-q14 sequence corrects errors and gaps in the public access sequence to fully reveal large segmental duplications at breakpoints for Prader-Willi, Angelman, and inv dup(15) syndromes. <b>2007</b> , 8, R114	62
2116	Characterization of the drugged human genome. <b>2007</b> , 8, 1063-73	42
2115	Beyond HLA: the significance of genomic variation for allogeneic hematopoietic stem cell transplantation. <b>2007</b> , 109, 1355-62	85
2114	PennCNV: an integrated hidden Markov model designed for high-resolution copy number variation detection in whole-genome SNP genotyping data. <b>2007</b> , 17, 1665-74	1278
2113	Comparative genome hybridization suggests a role for NRXN1 and APBA2 in schizophrenia. <b>2008</b> , 17, 458-65	315

2112	European guidelines on cardiovascular disease prevention in clinical practice: executive summary. Fourth Joint Task Force of the European Society of Cardiology and other societies on cardiovascular disease prevention in clinical practice (constituted by representatives of nine societies and by invited experts). <b>2007</b> , 14 Suppl 2, E1-40	309
2111	Multidrug resistance in epilepsy: a pharmacogenomic update. <b>2007</b> , 8, 1441-9	38
2110	Characterization and evolution of the novel gene family FAM90A in primates originated by multiple duplication and rearrangement events. <b>2007</b> , 16, 2572-82	21
2109	The gene balance hypothesis: from classical genetics to modern genomics. <b>2007</b> , 19, 395-402	315
2108	European guidelines on cardiovascular disease prevention in clinical practice: executive summary: Fourth Joint Task Force of the European Society of Cardiology and Other Societies on Cardiovascular Disease Prevention in Clinical Practice (Constituted by representatives of nine societies and by invited experts). <b>2007</b> , 28, 2375-414	1029
2107	Relative impact of nucleotide and copy number variation on gene expression phenotypes. <b>2007</b> , 315, 848-53	1361
2106	Elimination of altered karyotypes by sexual reproduction preserves species identity. <b>2007</b> , 50, 517-24	52
2105	Research in Computational Molecular Biology. <b>2007</b> ,	1
2104	Submicroscopic chromosomal imbalances detected by array-CGH are a frequent cause of congenital heart defects in selected patients. <b>2007</b> , 28, 2778-84	155
2103	Genomic Comparisons of Humans and Chimpanzees. <b>2007</b> , 36, 191-209	18
2102	Gene copy number variation in schizophrenia. <b>2007</b> , 96, 93-9	37
2101	Avian genomics in the 21st century. <b>2007</b> , 117, 6-13	10
2100	Variation of CNV distribution in five different ethnic populations. <b>2007</b> , 118, 19-30	43
2099	Integrative analysis of a cancer somatic mutome. <b>2007</b> , 6, 13	20
2098	The clinical implementation of sperm chromosome aneuploidy testing: pitfalls and promises. <b>2008</b> , 29, 124-33	43
2097	Clinical translation of genotyping and haplotyping data: implementation of in vivo pharmacology experience leading drug prescription to pharmacotyping. <b>2007</b> , 46, 807-24	22
2096	Characterization of breast cancer by array comparative genomic hybridization. <b>2007</b> , 85, 497-508	33
2095	Development and implementation of an analysis tool for array-based comparative genomic hybridization. <b>2007</b> , 46, 608-13	13

2094	Clinical implementation of chromosomal microarray analysis: summary of 2513 postnatal cases. <b>2007</b> , 2, e327	172
2093	Mutations in the UBIAD1 gene, encoding a potential prenyltransferase, are causal for Schnyder crystalline corneal dystrophy. <b>2007</b> , 2, e685	86
2092	[DNA microarrays in the clinic?]. <b>2007</b> , 23, 210-4	0
2091	Impact of normalization and filtering on linkage analysis of gene expression data. <b>2007</b> , 1 Suppl 1, S150	2
2090	A same gene for altruism and selfishness in primates. <b>2007</b> , 23, 440-4	0
2089	Tumor Genome Wide DNA Alterations Assessed by Array CGH in Patients with Poor and Excellent Survival following Operation for Colorectal Cancer. <b>2007</b> , 3, 117693510700300	13
2088	Integrating molecular biology into the veterinary curriculum. <b>2007</b> , 34, 658-73	2
2087	A large deletion of the PROS1 gene in a deep vein thrombosis patient with protein S deficiency. <b>2007</b> , 98, 783-9	19
2086	Human stem cells, chromatin, and tissue engineering: boosting relevancy in developmental toxicity testing. <b>2007</b> , 81, 20-40	3
2085	Out of the veil of death rode the one million! Neandertals and their genes. <b>2007</b> , 29, 105-10	4
2084	What a difference copy number variation makes. <b>2007</b> , 29, 311-3	31
2083	DNA microarrays in the clinic: how soon, how extensively?. <b>2007</b> , 29, 699-705	5
2082	Cancer genome sequencing: the challenges ahead. <b>2007</b> , 29, 783-94	79
2081	An evolution revolution provides further revelation. <b>2007</b> , 29, 1182-4	15
2080	A novel microdeletion at 16p11.2 harbors candidate genes for aortic valve development, seizure disorder, and mild mental retardation. <b>2007</b> , 143A, 1462-71	85
2079	Increase in GSK3beta gene copy number variation in bipolar disorder. <b>2007</b> , 144B, 259-65	100
2078	Cryptic telomere imbalance: a 15-year update. <b>2007</b> , 145C, 327-34	35
2077	Novel microdeletion syndromes. <b>2007</b> , 145C, 323-6	4

2076	The identification of microdeletion syndromes and other chromosome abnormalities: cytogenetic methods of the past, new technologies for the future. <b>2007</b> , 145C, 335-45	134
2075	Subtelomeric imbalances in phenotypically normal individuals. <b>2007</b> , 28, 958-67	67
2074	Array CGH identifies reciprocal 16p13.1 duplications and deletions that predispose to autism and/or mental retardation. <b>2007</b> , 28, 674-82	237
2073	Detection of pathogenic gene copy number variations in patients with mental retardation by genomewide oligonucleotide array comparative genomic hybridization. <b>2007</b> , 28, 1124-32	106
2072	Visualization of uniparental inheritance, Mendelian inconsistencies, deletions, and parent of origin effects in single nucleotide polymorphism trio data with SNP trio. <b>2007</b> , 28, 1225-35	41
2071	Double complex mutations involving F8 and FUNDC2 caused by distinct break-induced replication. <b>2007</b> , 28, 1198-206	51
2070	Rapid identification of disease-causing mutations using copy number analysis within linkage intervals. <b>2007</b> , 28, 1236-40	12
2069	Finding hippocampus minor. <b>2007</b> , 16, 88-93	1
2068	Delineation of a 1Mb breakpoint region at 1p13 in Wilms tumors by fine-tiling oligonucleotide array CGH. <b>2007</b> , 46, 607-15	18
2067	Allelic imbalances and microdeletions affecting the PTPRD gene in cutaneous squamous cell carcinomas detected using single nucleotide polymorphism microarray analysis. <b>2007</b> , 46, 661-9	74
2066	Homozygous deletion scanning of the lung cancer genome at a 100-kb resolution. <b>2007</b> , 46, 1000-10	59
2065	Genomic profile of chronic myelogenous leukemia: Imbalances associated with disease progression. <b>2007</b> , 46, 1039-50	46
2064	Toward a fluorescent single-strand conformation polymorphism technique that detects all mutations: F-DOVAM-S. <b>2007</b> , 368, 250-7	2
2063	La CGH array : un bouleversement de la pratique hospitalière en cytogénétique. <b>2007</b> , 28, 245-251	1
2062	High-throughput genotyping of a common deletion polymorphism disrupting the TRY6 gene and its association with breast cancer risk. <b>2007</b> , 8, 41	3
2061	An accurate method for quantifying and analyzing copy number variation in porcine KIT by an oligonucleotide ligation assay. <b>2007</b> , 8, 81	13
2060	Analysis of meiotic recombination in 22q11.2, a region that frequently undergoes deletions and duplications. <b>2007</b> , 8, 14	22
2059	MRPS18CP2 alleles and DEFA3 absence as putative chromosome 8p23.1 modifiers of hearing loss due to mtDNA mutation A1555G in the 12S rRNA gene. <b>2007</b> , 8, 81	8

2058	MLPA as a screening method of aneuploidy and unbalanced chromosomal rearrangements in spontaneous miscarriages. <b>2007</b> , 27, 765-71	36
2057	The evolutionary genetics of personality. <b>2007</b> , 21, 549-587	450
2056	Completing the map of human genetic variation. <i>Nature</i> , <b>2007</b> , 447, 161-5	50.4 153
2055	So similar, yet so different. <i>Nature</i> , <b>2007</b> , 449, 762-3	50.4
2054	Recurrent DNA copy number variation in the laboratory mouse. <b>2007</b> , 39, 1384-9	121
2053	Touching base. <b>2007</b> , 39, 941-941	3
2052	Rescuing distal crossovers. <b>2007</b> , 39, 1187-8	6
2051	Adaptive drool in the gene pool. <b>2007</b> , 39, 1188-90	16
2050	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. <b>2007</b> , 39, 319-28	1083
2049	Duplication of the MYB oncogene in T cell acute lymphoblastic leukemia. <b>2007</b> , 39, 593-5	211
2048	Methods and strategies for analyzing copy number variation using DNA microarrays. <b>2007</b> , 39, S16-21	367
2047	The population genetics of structural variation. <b>2007</b> , 39, S30-6	138
2046	FCGR3B copy number variation is associated with susceptibility to systemic, but not organ-specific, autoimmunity. <b>2007</b> , 39, 721-3	378
2045	Mutational and selective effects on copy-number variants in the human genome. <b>2007</b> , 39, S22-9	193
2044	A genome-wide association study for celiac disease identifies risk variants in the region harboring IL2 and IL21. <b>2007</b> , 39, 827-9	518
2043	Copy-number variation and association studies of human disease. <b>2007</b> , 39, S37-42	464
2042	Genomic rearrangements and sporadic disease. <b>2007</b> , 39, S43-7	331
2041	A new multipoint method for genome-wide association studies by imputation of genotypes. <b>2007</b> , 39, 906-13	2040

2040	Copy number variations and clinical cytogenetic diagnosis of constitutional disorders. <b>2007</b> , 39, S48-54	293
2039	Challenges and standards in integrating surveys of structural variation. <b>2007</b> , 39, S7-15	279
2038	Major changes in our DNA lead to major changes in our thinking. <b>2007</b> , 39, S3-5	84
2037	Discovery of a previously unrecognized microdeletion syndrome of 16p11.2-p12.2. <b>2007</b> , 39, 1071-3	166
2036	Diet and the evolution of human amylase gene copy number variation. <b>2007</b> , 39, 1256-60	973
2035	Autism: highly heritable but not inherited. <b>2007</b> , 13, 534-6	72
2034	The bridge between dendritic cells and asthma. <b>2007</b> , 13, 536-8	9
2033	Construction and use of spotted large-insert clone DNA microarrays for the detection of genomic copy number changes. <b>2007</b> , 2, 577-87	15
2032	Copy number variation map. <b>2007</b> , 8, 2-2	2
2031	Copy number variation doesn't copy SNPs. <b>2007</b> , 8, 247-247	1
2030	From microscopes to microarrays: dissecting recurrent chromosomal rearrangements. <b>2007</b> , 8, 869-83	107
2029	Copy number variants and genetic traits: closer to the resolution of phenotypic to genotypic variability. <b>2007</b> , 8, 639-46	335
2028	Mammalian karyotype evolution. <b>2007</b> , 8, 950-62	216
2027	Distribution of the D15Z1 copy number polymorphism. <b>2007</b> , 15, 441-5	19
2026	Insights into modern disease from our distant evolutionary past. <b>2007</b> , 15, 603-6	1
2025	A comprehensive screen for SNP associations on chromosome region 5q31-33 in Swedish/Norwegian celiac disease families. <b>2007</b> , 15, 980-7	12
2024	Beyond susceptibility. Behavioural genetics can advance our understanding of psychiatric disorders, but might not meet the expectations for new cures. <b>2007</b> , 8 Spec No, S3-6	1
2023	CCL3L1 and CCL4L1: variable gene copy number in adolescents with and without human immunodeficiency virus type 1 (HIV-1) infection. <b>2007</b> , 8, 224-31	48

2022	High-resolution genomic profiling of childhood ALL reveals novel recurrent genetic lesions affecting pathways involved in lymphocyte differentiation and cell cycle progression. <b>2007</b> , 21, 1258-66	303
2021	Pharmacogenetics and pharmacogenomics of schizophrenia: a review of last decade of research. <b>2007</b> , 12, 707-47	288
2020	Getting it right: designing microarray (and not 'microawry') comparative genomic hybridization studies for cancer research. <b>2007</b> , 87, 737-54	62
2019	A second generation human haplotype map of over 3.1 million SNPs. <i>Nature</i> , <b>2007</b> , 449, 851-61	50.4 3647
2018	The impact of array genomic hybridization on mental retardation research: a review of current technologies and their clinical utility. <b>2007</b> , 72, 271-87	36
2017	Pure subtelomeric microduplications as a cause of mental retardation. <b>2007</b> , 72, 362-8	24
2016	Interstitial deletion of chromosome 4p associated with mild mental retardation, epilepsy and polymicrogyria of the left temporal lobe. <b>2007</b> , 72, 593-8	9
2015	Deletion of entire HLA-A gene accompanied by an insertion of a retrotransposon. <b>2007</b> , 70, 144-50	35
2014	Platelet genomics and the risk of atherothrombosis. <b>2007</b> , 5 Suppl 1, 188-95	46
2013	Characterization of 26 miniSTR loci for improved analysis of degraded DNA samples. <b>2008</b> , 53, 73-80	109
2012	Evolution of man in the light of molecular genetics: a review. Part I. Our evolutionary history and genomics. <b>2007</b> , 144, 80-95	6
2011	Using expression arrays for copy number detection: an example from <i>E. coli</i> . <b>2007</b> , 8, 203	8
2010	Assessment of algorithms for high throughput detection of genomic copy number variation in oligonucleotide microarray data. <b>2007</b> , 8, 368	42
2009	Local Renyi entropic profiles of DNA sequences. <b>2007</b> , 8, 393	27
2008	Extensive lineage-specific gene duplication and evolution of the spiggin multi-gene family in stickleback. <b>2007</b> , 7, 209	12
2007	Inter-population variability of DEFA3 gene absence: correlation with haplotype structure and population variability. <b>2007</b> , 8, 14	20
2006	Large-scale copy number variants (CNVs): distribution in normal subjects and FISH/real-time qPCR analysis. <b>2007</b> , 8, 167	15
2005	Toward accurate high-throughput SNP genotyping in the presence of inherited copy number variation. <b>2007</b> , 8, 211	8

2004	Cis sequence effects on gene expression. <b>2007</b> , 8, 296	6
2003	X-chromosome tiling path array detection of copy number variants in patients with chromosome X-linked mental retardation. <b>2007</b> , 8, 443	46
2002	In silico and in vitro comparative analysis to select, validate and test SNPs for human identification. <b>2007</b> , 8, 457	13
2001	Cryptic del(13q14.2) and physiological deletions of immunoglobulin genes detected by high-resolution array comparative genomic hybridization in a patient with indolent chronic lymphocytic leukemia. <b>2007</b> , 176, 89-91	5
2000	Rearrangements involving 12q in myeloproliferative disorders: possible role of HMGA2 and SOCS2 genes. <b>2007</b> , 176, 80-8	25
1999	Identification of cryptic microaberrations in osteosarcoma by high-definition oligonucleotide array comparative genomic hybridization. <b>2007</b> , 179, 52-61	38
1998	Promiscuous mutations activate the noncanonical NF-kappaB pathway in multiple myeloma. <b>2007</b> , 12, 131-44	832
1997	Insertion of an HERV(K) LTR in the intron of NBPF3 is not required for its transcriptional activity. <b>2007</b> , 362, 1-5	6
1996	Influence of cytochrome P450 polymorphisms on drug therapies: pharmacogenetic, pharmacoeigenetic and clinical aspects. <b>2007</b> , 116, 496-526	848
1995	Non-B DNA conformations, mutagenesis and disease. <b>2007</b> , 32, 271-8	233
1994	Genetic variation in human disease and a new role for copy number variants. <b>2007</b> , 622, 33-41	28
1993	Cell culture models in developing nutrigenomics foods for inflammatory bowel disease. <b>2007</b> , 622, 94-102	30
1992	Nutrigenomics and gut health. <b>2007</b> , 622, 1-6	14
1991	Functional MAPT haplotypes: bridging the gap between genotype and neuropathology. <b>2007</b> , 27, 1-10	52
1990	Calculation of some characteristics of large genomes. <b>2007</b> , 76, 934-939	2
1989	Paired-end mapping reveals extensive structural variation in the human genome. <b>2007</b> , 318, 420-6	895
1988	Mannose binding lectin (MBL) copy number polymorphism in Zebrafish ( <i>D. rerio</i> ) and identification of haplotypes resistant to <i>L. anguillarum</i> . <b>2007</b> , 59, 861-72	41
1987	The hyper-IgE syndrome is not caused by a microdeletion syndrome. <b>2007</b> , 59, 913-26	4



1986	Novel repeat polymorphisms of the dopaminergic neurotransmitter genes among dogs and wolves. <b>2007</b> , 18, 871-9	31
1985	Scanning the horizon: what is the future of genome-wide association studies in accelerating discoveries in cancer etiology and prevention?. <b>2007</b> , 18, 479-84	18
1984	Linkage study of 14 candidate genes and loci in four large Dutch families with vesico-ureteral reflux. <b>2007</b> , 22, 1129-33	20
1983	What's new in karyotyping? The move towards array comparative genomic hybridisation (CGH). <b>2007</b> , 166, 637-43	36
1982	Molecular characterization of a polymorphic 3-Mb deletion at chromosome Yp11.2 containing the AMELY locus in Singapore and Malaysia populations. <b>2007</b> , 122, 237-49	28
1981	Variation in the selenoprotein S gene locus is associated with coronary heart disease and ischemic stroke in two independent Finnish cohorts. <b>2007</b> , 122, 355-65	85
1980	Germ-line DNA copy number variation frequencies in a large North American population. <b>2007</b> , 122, 345-53	122
1979	High-density oligonucleotide array with sub-kilobase resolution reveals breakpoint information of submicroscopic deletions in nevoid basal cell carcinoma syndrome. <b>2007</b> , 122, 459-66	25
1978	RAS/RAF pathway activation in gliomas: the result of copy number gains rather than activating mutations. <b>2007</b> , 114, 121-33	87
1977	Construction of a high-density and high-resolution human chromosome X array for comparative genomic hybridization analysis. <b>2007</b> , 52, 397-405	19
1976	Genetics of bipolar disorder. <b>2007</b> , 9, 504-11	19
1975	Genome mirror-2007. <b>2007</b> , 1, 147-8	
1974	Molecular mechanisms of chromosomal rearrangement during primate evolution. <b>2008</b> , 16, 41-56	59
1973	Addressing chromosome evolution in the whole-genome sequence era. <b>2008</b> , 16, 5-16	12
1972	Pharmacogenetics: data, concepts and tools to improve drug discovery and drug treatment. <b>2008</b> , 64, 133-57	42
1971	Functional genetic variation of human miRNAs and phenotypic consequences. <b>2008</b> , 19, 503-9	70
1970	Molekulare Karyotypisierung in der klinischen Anwendung. <b>2008</b> , 20, 419-431	1
1969	Molekulare Karyotypisierung in der klinischen Diagnostik. <b>2008</b> , 20, 386-394	3

1968	Strukturelle Genomvarianten. <b>2008</b> , 20, 401-406	1
1967	Neue Verfahren für Einzelzellanalysen in Forschung und Diagnostik. <b>2008</b> , 20, 407-415	
1966	Common and rare alleles as causes of complex phenotypes. <b>2008</b> , 10, 194-200	13
1965	Clinical implications of the "personal" genome. <b>2008</b> , 10, 361-3	3
1964	Genomic structural variation and schizophrenia. <b>2008</b> , 10, 171-7	10
1963	SNPs in genes with copy number variation: a question of specificity. <b>2008</b> , 87, 95-7	2
1962	Identification of a significant association of a single nucleotide polymorphism in TNXB with systemic lupus erythematosus in a Japanese population. <b>2008</b> , 53, 64-73	22
1961	Structural genomic variation in ischemic stroke. <b>2008</b> , 9, 101-8	26
1960	Heterogeneous dysregulation of microRNAs across the autism spectrum. <b>2008</b> , 9, 153-61	208
1959	A comparative analysis of two tissue procurement approaches for the genomic profiling of clinical colorectal cancer samples. <b>2008</b> , 23, 1089-98	2
1958	Array-CGH in patients with Kabuki-like phenotype: identification of two patients with complex rearrangements including 2q37 deletions and no other recurrent aberration. <b>2008</b> , 9, 27	26
1957	Complex aetiology of an apparently Mendelian form of mental retardation. <b>2008</b> , 9, 6	4
1956	Submicroscopic chromosome imbalance in patients with developmental delay and/or dysmorphism referred specifically for Fragile X testing and karyotype analysis. <b>2008</b> , 1, 2	9
1955	Generation of a genomic tiling array of the human major histocompatibility complex (MHC) and its application for DNA methylation analysis. <b>2008</b> , 1, 19	23
1954	Improved detection of global copy number variation using high density, non-polymorphic oligonucleotide probes. <b>2008</b> , 9, 27	20
1953	A large-scale survey of genetic copy number variations among Han Chinese residing in Taiwan. <b>2008</b> , 9, 92	23
1952	Population proteomics: investigation of protein diversity in human populations. <b>2008</b> , 8, 779-86	38
1951	Aneuploid mosaicism in the developing and adult cerebellar cortex. <b>2008</b> , 507, 1944-51	70

1950	Multiplex PCR-based real-time invader assay (mPCR-RETINA): a novel SNP-based method for detecting allelic asymmetries within copy number variation regions. <b>2008</b> , 29, 182-9	25
1949	Array-MLPA: comprehensive detection of deletions and duplications and its application to DMD patients. <b>2008</b> , 29, 190-7	49
1948	Genomewide SNP assay reveals mutations underlying Parkinson disease. <b>2008</b> , 29, 315-22	44
1947	GPCR NaVa database: natural variants in human G protein-coupled receptors. <b>2008</b> , 29, 39-44	40
1946	Profiling of copy number variations (CNVs) in healthy individuals from three ethnic groups using a human genome 32 K BAC-clone-based array. <b>2008</b> , 29, 398-408	43
1945	Genome-wide copy number analysis using copy number inferring tool (CNIT) and DNA pooling. <b>2008</b> , 29, 1055-62	9
1944	Long interspersed nuclear element-1 (LINE1)-mediated deletion of EVC, EVC2, C4orf6, and STK32B in Ellis-van Creveld syndrome with borderline intelligence. <b>2008</b> , 29, 931-8	43
1943	Somatic mosaicism for copy number variation in differentiated human tissues. <b>2008</b> , 29, 1118-24	172
1942	Molecular diagnosis of Duchenne/Becker muscular dystrophy: enhanced detection of dystrophin gene rearrangements by oligonucleotide array-comparative genomic hybridization. <b>2008</b> , 29, 1100-7	82
1941	Lung cancer epigenetics and genetics. <b>2008</b> , 123, 1-7	173
1940	High-resolution genomic and expression analyses of copy number alterations in breast tumors. <b>2008</b> , 47, 530-42	123
1939	High-resolution copy number arrays in cancer and the problem of normal genome copy number variation. <b>2008</b> , 47, 933-8	6
1938	New applications and developments in the use of multiplex ligation-dependent probe amplification. <b>2008</b> , 29, 4627-36	79
1937	On the analysis of copy-number variations in genome-wide association studies: a translation of the family-based association test. <b>2008</b> , 32, 273-84	49
1936	Problem of using cases with genetic anomalies as a reference group in case-control studies on drug use and birth defects. <b>2008</b> , 82, 173-4; author reply 175	1
1935	Neuroimaging findings in children with rare or novel de novo chromosomal anomalies. <b>2008</b> , 82, 200-10	1
1934	Fine-mapping subtelomeric deletions and duplications by comparative genomic hybridization in 42 individuals. <b>2008</b> , 146A, 730-9	23
1933	Inverted duplication with terminal deletion of 5p and no cat-like cry. <b>2008</b> , 146A, 1173-9	15

1932	Array comparative genomic hybridization (aCGH) analysis in Prader-Willi syndrome. <b>2008</b> , 146A, 854-60	64
1931	Analytical and clinical validity of whole-genome oligonucleotide array comparative genomic hybridization for pediatric patients with mental retardation and developmental delay. <b>2008</b> , 146A, 1942-54	43
1930	Homozygous deletions of a copy number change detected by array CGH: a new cause for mental retardation?. <b>2008</b> , 146A, 1903-10	11
1929	Deletion (1)(p32.2-p32.3) detected by array-CGH in a patient with developmental delay/mental retardation, dysmorphic features and low cholesterol: A new microdeletion syndrome?. <b>2008</b> , 146A, 2284-90	10
1928	Application of metaphase HR-CGH and targeted Chromosomal Microarray Analyses to genomic characterization of 116 patients with mental retardation and dysmorphic features. <b>2008</b> , 146A, 2361-9	15
1927	Genetic association studies of the chromosome 15 GABA-A receptor cluster in migraine with aura. <b>2008</b> , 147B, 37-41	18
1926	Up-regulation of ADM and SEPX1 in the lymphoblastoid cells of patients in monozygotic twins discordant for schizophrenia. <b>2008</b> , 147B, 557-64	23
1925	Gene copy number variation in schizophrenia. <b>2008</b> , 147B, 606-11	13
1924	The XVth World Congress of Psychiatric Genetics, October 7-11, 2007: Rapporteur summaries of oral presentations. <b>2008</b> , 147B, 233-77	4
1923	FBXL21 association with schizophrenia in Irish family and case-control samples. <b>2008</b> , 147B, 1231-7	10
1922	Genomic investigation of alpha-synuclein multiplication and parkinsonism. <b>2008</b> , 63, 743-50	269
1921	Epidemiology and Genetic Epidemiology. <b>2008</b> , 1109-1140	2
1920	Array-based DNA diagnostics: let the revolution begin. <b>2008</b> , 59, 113-29	118
1919	From human genetics and genomics to pharmacogenetics and pharmacogenomics: past lessons, future directions. <b>2008</b> , 40, 187-224	129
1918	Natural selection shapes genome-wide patterns of copy-number polymorphism in <i>Drosophila melanogaster</i> . <b>2008</b> , 320, 1629-31	217
1917	Copy number variant analysis of human embryonic stem cells. <b>2008</b> , 26, 1484-9	42
1916	Genetic mechanisms controlling cardiovascular development. <b>2008</b> , 1123, 10-9	67
1915	Structural variation of chromosomes in autism spectrum disorder. <b>2008</b> , 82, 477-88	1413

1914	The fine-scale and complex architecture of human copy-number variation. <b>2008</b> , 82, 685-95	289
1913	Phenotypically concordant and discordant monozygotic twins display different DNA copy-number-variation profiles. <b>2008</b> , 82, 763-71	437
1912	Evaluation of genetic variation contributing to differences in gene expression between populations. <b>2008</b> , 82, 631-40	177
1911	Detection, imputation, and association analysis of small deletions and null alleles on oligonucleotide arrays. <b>2008</b> , 82, 1316-33	32
1910	An algorithm for inferring complex haplotypes in a region of copy-number variation. <b>2008</b> , 83, 157-69	19
1909	Extensive copy-number variation of the human olfactory receptor gene family. <b>2008</b> , 83, 228-42	110
1908	Adaptive evolution of UGT2B17 copy-number variation. <b>2008</b> , 83, 337-46	112
1907	Recurrent CNVs disrupt three candidate genes in schizophrenia patients. <b>2008</b> , 83, 504-10	220
1906	Genome-wide copy-number-variation study identified a susceptibility gene, UGT2B17, for osteoporosis. <b>2008</b> , 83, 663-74	168
1905	In silico investigations on functional and haplotype tag SNPs associated with congenital long QT syndromes (LQTSs). <b>2008</b> , 2, 55-67	4
1904	Disorders of the genome architecture: a review. <b>2008</b> , 2, 69-76	22
1903	Genome-wide quantitative trait locus association scan of general cognitive ability using pooled DNA and 500K single nucleotide polymorphism microarrays. <b>2008</b> , 7, 435-46	112
1902	Understanding incidental findings in the context of genetics and genomics. <b>2008</b> , 36, 280-5, 212	93
1901	Homozygosity mapping in a family presenting with schizophrenia, epilepsy and hearing impairment. <b>2008</b> , 16, 750-8	26
1900	The copy number variant involving part of the alpha7 nicotinic receptor gene contains a polymorphic inversion. <b>2008</b> , 16, 1364-71	31
1899	The success of the genome-wide association approach: a brief story of a long struggle. <b>2008</b> , 16, 554-64	85
1898	Detection of submicroscopic constitutional chromosome aberrations in clinical diagnostics: a validation of the practical performance of different array platforms. <b>2008</b> , 16, 786-92	28
1897	The genome: you gain some, you lose some. <b>2008</b> , 16, 663	

1896	The effect of pedigree structure on detection of deletions and other null alleles. <b>2008</b> , 16, 1225-34		2
1895	Array-CGH fine mapping of minor and cryptic HR-CGH detected genomic imbalances in 80 out of 590 patients with abnormal development. <b>2008</b> , 16, 1318-28		10
1894	Genomic analysis of the HER2/TOP2A amplicon in breast cancer and breast cancer cell lines. <b>2008</b> , 88, 491-503		116
1893	Understanding the development of human bladder cancer by using a whole-organ genomic mapping strategy. <b>2008</b> , 88, 694-721		55
1892	High-resolution analysis of chromosome copy number alterations in angioimmunoblastic T-cell lymphoma and peripheral T-cell lymphoma, unspecified, with single nucleotide polymorphism-typing microarrays. <b>2008</b> , 22, 1891-8		37
1891	Clinicopathological features and global genomic copy number alterations of pilomyxoid astrocytoma in the hypothalamus/optic pathway: comparative analysis with pilocytic astrocytoma using array-based comparative genomic hybridization. <b>2008</b> , 21, 1345-56		32
1890	A study of rare structural variants in schizophrenia patients and normal controls from Chinese Han population. <b>2008</b> , 13, 911-3		29
1889	Genotype, haplotype and copy-number variation in worldwide human populations. <i>Nature</i> , <b>2008</b> , 451, 998-1003	50.4	662
1888	Mapping and sequencing of structural variation from eight human genomes. <i>Nature</i> , <b>2008</b> , 453, 56-64	50.4	878
1887	The complete genome of an individual by massively parallel DNA sequencing. <i>Nature</i> , <b>2008</b> , 452, 872-6	50.4	1424
1886	A glimmer of light for neuropsychiatric disorders. <i>Nature</i> , <b>2008</b> , 455, 890-3	50.4	75
1885	Copy-number variations associated with neuropsychiatric conditions. <i>Nature</i> , <b>2008</b> , 455, 919-23	50.4	513
1884	Accurate whole human genome sequencing using reversible terminator chemistry. <i>Nature</i> , <b>2008</b> , 456, 53-9	50.4	2615
1883	Integrative analysis reveals 53BP1 copy loss and decreased expression in a subset of human diffuse large B-cell lymphomas. <b>2008</b> , 27, 318-22		28
1882	Genome-wide DNA copy number analysis in pancreatic cancer using high-density single nucleotide polymorphism arrays. <b>2008</b> , 27, 1951-60		110
1881	How can we realize the promise of personalized antidepressant medicines?. <b>2008</b> , 9, 638-46		140
1880	What would you do if you could sequence everything?. <b>2008</b> , 26, 1125-33		155
1879	Distribution and functional impact of DNA copy number variation in the rat. <b>2008</b> , 40, 538-45		170

1878	Strong association of de novo copy number mutations with sporadic schizophrenia. <b>2008</b> , 40, 880-5	667
1877	Mouse segmental duplication and copy number variation. <b>2008</b> , 40, 909-14	180
1876	Evolutionary toggling of the MAPT 17q21.31 inversion region. <b>2008</b> , 40, 1076-83	138
1875	Closing gaps in the human genome with fosmid resources generated from multiple individuals. <b>2008</b> , 40, 96-101	47
1874	Germline rates of de novo meiotic deletions and duplications causing several genomic disorders. <b>2008</b> , 40, 90-5	235
1873	A robust statistical method for case-control association testing with copy number variation. <b>2008</b> , 40, 1245-52	143
1872	Deletion polymorphism upstream of IRGM associated with altered IRGM expression and Crohn's disease. <b>2008</b> , 40, 1107-12	527
1871	Systematic assessment of copy number variant detection via genome-wide SNP genotyping. <b>2008</b> , 40, 1199-203	174
1870	Integrated detection and population-genetic analysis of SNPs and copy number variation. <b>2008</b> , 40, 1166-74	773
1869	Recurrent reciprocal 1q21.1 deletions and duplications associated with microcephaly or macrocephaly and developmental and behavioral abnormalities. <b>2008</b> , 40, 1466-71	457
1868	Genome-wide association scan in women with systemic lupus erythematosus identifies susceptibility variants in ITGAM, PXX, KIAA1542 and other loci. <b>2008</b> , 40, 204-10	1021
1867	Copy-number analysis goes more than skin deep. <b>2008</b> , 40, 5-6	28
1866	Genomic rearrangements in the spotlight. <b>2008</b> , 40, 6-7	3
1865	ESR1 gene amplification in breast cancer: a common phenomenon?. <b>2008</b> , 40, 806-7; author reply 810-2	55
1864	Phenotypic variations on the theme of CNVs. <b>2008</b> , 40, 1392-3	46
1863	Comparing whole genomes using DNA microarrays. <b>2008</b> , 9, 291-302	159
1862	Leader of the pack: gene mapping in dogs and other model organisms. <b>2008</b> , 9, 713-25	194
1861	The functional repertoires of metazoan genomes. <b>2008</b> , 9, 689-98	91

1860	Explaining human uniqueness: genome interactions with environment, behaviour and culture. <b>2008</b> , 9, 749-63	131
1859	From diversity to delivery: the case of the Indian Genome Variation initiative. <b>2008</b> , 9 Suppl 1, S9-14	13
1858	Genomics, public health and developing countries: the case of the Mexican National Institute of Genomic Medicine (INMEGEN). <b>2008</b> , 9 Suppl 1, S5-9	30
1857	The evolution of animal chemosensory receptor gene repertoires: roles of chance and necessity. <b>2008</b> , 9, 951-63	443
1856	Genome-wide association studies: a new window into immune-mediated diseases. <b>2008</b> , 8, 631-43	103
1855	Identification of non-recurrent submicroscopic genome imbalances: the advantage of genome-wide microarrays over targeted approaches. <b>2008</b> , 16, 395-400	12
1854	A 3.2 Mb deletion on 18q12 in a patient with childhood autism and high-grade myopia. <b>2008</b> , 16, 312-9	14
1853	Copy number variations in the NF1 gene region are infrequent and do not predispose to recurrent type-1 deletions. <b>2008</b> , 16, 572-80	20
1852	Evaluation of coverage variation of SNP chips for genome-wide association studies. <b>2008</b> , 16, 635-43	88
1851	Development of therapeutic siRNAs for pachyonychia congenita. <b>2008</b> , 128, 50-8	55
1850	Monoallelic or biallelic LMO2 expression in relation to the LMO2 rearrangement status in pediatric T-cell acute lymphoblastic leukemia. <b>2008</b> , 22, 1434-7	14
1849	Cooperative genetic defects in TLX3 rearranged pediatric T-ALL. <b>2008</b> , 22, 762-70	27
1848	A whole genome association study of neuroticism using DNA pooling. <b>2008</b> , 13, 302-12	127
1847	CNTNAP2 gene dosage variation is associated with schizophrenia and epilepsy. <b>2008</b> , 13, 261-6	272
1846	Quantitative trait locus association scan of early reading disability and ability using pooled DNA and 100K SNP microarrays in a sample of 5760 children. <b>2008</b> , 13, 729-40	79
1845	Testing for equality of standardized composite measures of linkage disequilibrium. <b>2008</b> , 72, 292-6	2
1844	Segmental copy-number variation observed in Japanese by array-CGH. <b>2008</b> , 72, 193-204	12
1843	Large-scale molecular analysis of a 34 Mb interval on chromosome 6q: major refinement of the RP25 interval. <b>2008</b> , 72, 463-77	4



1842	Genotype-phenotype correlation in five Pelizaeus-Merzbacher disease patients with PLP1 gene duplications. <b>2008</b> , 73, 279-87	17
1841	Genetic factors in congenital heart malformation. <b>2008</b> , 73, 516-27	31
1840	Chromosome copy number analysis in screening for prognosis-related genomic regions in colorectal carcinoma. <b>2008</b> , 99, 1835-40	56
1839	Genetic developments in autoimmune thyroid disease: an evolutionary process. <b>2008</b> , 68, 671-82	37
1838	An evolutionary perspective on Y-chromosomal variation and male infertility. <b>2008</b> , 31, 376-82	39
1837	Identifying the susceptibility genes for coronary artery disease: from hyperbole through doubt to cautious optimism. <b>2008</b> , 263, 538-52	37
1836	A chromosomal duplication that includes the canine microsatellite INRA21 in Labrador Retrievers. <b>2008</b> , 39, 241-8	2
1835	Characterization of a novel microdeletion polymorphism on BTA5 in cattle. <b>2008</b> , 39, 655-8	3
1834	MPDA: microarray pooled DNA analyzer. <b>2008</b> , 9, 196	10
1833	Major copy proportion analysis of tumor samples using SNP arrays. <b>2008</b> , 9, 204	34
1832	Probe-specific mixed-model approach to detect copy number differences using multiplex ligation-dependent probe amplification (MLPA). <b>2008</b> , 9, 261	11
1831	The evolution of genome size in ants. <b>2008</b> , 8, 64	52
1830	Whole genome comparative studies between chicken and turkey and their implications for avian genome evolution. <b>2008</b> , 9, 168	98
1829	Strong position-dependent effects of sequence mismatches on signal ratios measured using long oligonucleotide microarrays. <b>2008</b> , 9, 317	27
1828	Short tandem repeats in human exons: a target for disease mutations. <b>2008</b> , 9, 410	39
1827	Copy number variations (CNVs) identified in Korean individuals. <b>2008</b> , 9, 492	17
1826	Experimental analysis of oligonucleotide microarray design criteria to detect deletions by comparative genomic hybridization. <b>2008</b> , 9, 497	13
1825	Analysis of the largest tandemly repeated DNA families in the human genome. <b>2008</b> , 9, 533	106

1824	Genetic profiling of myeloproliferative disorders by single-nucleotide polymorphism oligonucleotide microarray. <b>2008</b> , 36, 1471-9	40
1823	Role of DNA polymerases eta, iota and zeta in UV resistance and UV-induced mutagenesis in a human cell line. <b>2008</b> , 7, 1551-62	86
1822	Addressing informatics challenges in Translational Research with workflow technology. <b>2008</b> , 13, 771-7	7
1821	The array CGH and its clinical applications. <b>2008</b> , 13, 760-70	145
1820	Navigating the channels and beyond: unravelling the genetics of the epilepsies. <b>2008</b> , 7, 231-45	219
1819	Copy-number variation in sporadic amyotrophic lateral sclerosis: a genome-wide screen. <b>2008</b> , 7, 319-26	78
1818	Human MHC architecture and evolution: implications for disease association studies. <b>2008</b> , 35, 179-92	150
1817	A Genetics Primer for Social Health Research. <b>2008</b> , 2, 785-816	9
1816	The current excitement about copy-number variation: how it relates to gene duplications and protein families. <b>2008</b> , 18, 366-74	78
1815	The functional impact of structural variation in humans. <b>2008</b> , 24, 238-45	128
1814	Structural genomic variation and personalized medicine. <b>2008</b> , 358, 740-1	40
1813	Strategies and Resources for Marker Selection and Genotyping in Genetic Association Studies. <b>2008</b> , 149-183	
1812	Single Strand Conformation Polymorphism (SSCP) Analysis. <b>2008</b> , 117-131	
1811	Microduplications of 22q11.2 are frequently inherited and are associated with variable phenotypes. <b>2008</b> , 10, 267-77	153
1810	Multiple sclerosis genetics. <b>2008</b> , 318, 45-72	32
1809	Integrating genomic and clinical medicine: searching for susceptibility genes in complex lung diseases. <b>2008</b> , 151, 181-93	11
1808	Genetic analysis of the gene coding for DARPP-32 (PPP1R1B) in Japanese patients with schizophrenia or bipolar disorder. <b>2008</b> , 100, 334-41	15
1807	Clinical utility of contemporary molecular cytogenetics. <b>2008</b> , 9, 71-86	68

1806	Copy number alterations and copy number variation in cancer: close encounters of the bad kind. <b>2008</b> , 123, 176-82	29
1805	The human genome and sport, including epigenetics and athleticogenomics: a brief look at a rapidly changing field. <b>2008</b> , 26, 1127-33	10
1804	Copy number variation of beta-defensins and relevance to disease. <b>2008</b> , 123, 148-55	57
1803	Title Page / Table of Contents. <b>2008</b> , 123, 1-4	1
1802	Guías de práctica clínica sobre prevención de la enfermedad cardiovascular: versión resumida. <b>2008</b> , 61, 82.e1-82.e49	14
1801	Human telomere structure and biology. <b>2008</b> , 9, 1-19	94
1800	Genotyping platforms for mass-throughput genotyping with SNPs, including human genome-wide scans. <b>2008</b> , 60, 107-39	28
1799	MAQ, A NOVEL METHOD FOR PCR BASED, HIGH-THROUGHPUT COPY NUMBER VARIATION ANALYSIS APPLIED TO THE 22q11 DELETION SYNDROME. <b>2008</b> , 102, 185	
1798	AGE AT ONSET OF PSYCHOSIS IS RELATED TO GENOMIC GAINS IN CHROMOSOME 17q. <b>2008</b> , 102, 185	
1797	miRNA DYSFUNCTION IN SCHIZOPHRENIA: GENETIC AND FUNCTIONAL EVIDENCE. <b>2008</b> , 102, 185-186	
1796	Contributions of spermatozoa to embryogenesis: assays to evaluate their genetic and epigenetic fitness. <b>2008</b> , 16, 474-84	61
1795	Genomics: the next step to elucidate the etiology of calcific aortic valve stenosis. <b>2008</b> , 51, 1327-36	56
1794	Advances in multiple Sclerosis and Experimental Demyelinating Diseases. <b>2008</b> ,	3
1793	Essentials of Clinical Research. <b>2008</b> ,	9
1792	Model-based analysis of ChIP-Seq (MACS). <b>2008</b> , 9, R137	8406
1791	The signature of long-standing balancing selection at the human defensin beta-1 promoter. <b>2008</b> , 9, R143	53
1790	Finishing the finished human chromosome 22 sequence. <b>2008</b> , 9, R78	17
1789	Widespread duplications in the genomes of laboratory stocks of Dictyostelium discoideum. <b>2008</b> , 9, R75	56

1788	Computational methods for identification of recurrent copy number alteration patterns by array CGH. <b>2008</b> , 123, 343-51	19
1787	Classification of genetic profiles of Crohn's disease: a focus on the ATG16L1 gene. <b>2008</b> , 8, 199-207	10
1786	A highly conserved segmental duplication in the subtelomeres of Plasmodium falciparum chromosomes varies in copy number. <b>2008</b> , 7, 46	14
1785	Performance of whole-genome amplified DNA isolated from serum and plasma on high-density single nucleotide polymorphism arrays. <b>2008</b> , 10, 249-57	16
1784	Copy Number Variation Detection via High-Density SNP Genotyping. <b>2008</b> , 2008, pdb.top46	19
1783	Noninvasive prenatal diagnosis of fetal chromosomal aneuploidy by massively parallel genomic sequencing of DNA in maternal plasma. <b>2008</b> , 105, 20458-63	681
1782	Recurrent rearrangements of chromosome 1q21.1 and variable pediatric phenotypes. <b>2008</b> , 359, 1685-99	587
1781	Handbook of Obesity. <b>2008</b> ,	6
1780	Molecular Biomethods Handbook. <b>2008</b> ,	10
1779	Detection and validation of copy number variation in X-linked mental retardation. <b>2008</b> , 123, 44-53	10
1778	Positive selection in the human genome: from genome scans to biological significance. <b>2008</b> , 9, 143-60	63
1777	Targeting minor histocompatibility antigens in graft versus tumor or graft versus leukemia responses. <b>2008</b> , 29, 624-32	37
1776	Autosomal-dominant microtia linked to five tandem copies of a copy-number-variable region at chromosome 4p16. <b>2008</b> , 82, 181-7	38
1775	Disruption of neurexin 1 associated with autism spectrum disorder. <b>2008</b> , 82, 199-207	457
1774	Molecular cytogenetic analysis and resequencing of contactin associated protein-like 2 in autism spectrum disorders. <b>2008</b> , 82, 165-73	427
1773	A defect in the TUSC3 gene is associated with autosomal recessive mental retardation. <b>2008</b> , 82, 1158-64	107
1772	[New chromosomal syndromes]. <b>2008</b> , 56, 380-7	3
1771	Gene dosage methods as diagnostic tools for the identification of chromosome abnormalities. <b>2008</b> , 56, 345-53	22

1770	[Management of the CNVs in constitutional human genetics using array CGH]. <b>2008</b> , 56, 354-61	0
1769	Are animal models as good as we think?. <b>2008</b> , 69, 2-9	79
1768	Methods to detect CNVs in the human genome. <b>2008</b> , 123, 313-21	28
1767	Methods to detect and analyze copy number variations at the genome-wide and locus-specific levels. <b>2008</b> , 123, 333-42	20
1766	Studying copy number variations using a nanofluidic platform. <b>2008</b> , 36, e116	86
1765	A 580 kb microdeletion in 17q21.32 associated with mental retardation, microcephaly, cleft palate, and cardiac malformation. <b>2008</b> , 51, 74-80	4
1764	Prospective screening of patients with unexplained mental retardation using subtelomeric MLPA strongly increases the detection rate of cryptic unbalanced chromosomal rearrangements. <b>2008</b> , 51, 93-105	14
1763	Concurrent transposition of distal 6p and 20q to the 22q telomere: a recurrent benign chromosomal variant. <b>2008</b> , 51, 148-55	2
1762	Inherited 18q23 duplication in a fetus with multiple congenital anomalies. <b>2008</b> , 51, 231-8	5
1761	Genetic causality in schizophrenia and bipolar disorder: out with the old and in with the new. <b>2008</b> , 18, 229-34	45
1760	Prediction of individual genetic risk of complex disease. <b>2008</b> , 18, 257-63	129
1759	Analysis of the multi-copy gene family FAM90A as a copy number variant in different ethnic backgrounds. <b>2008</b> , 420, 113-7	3
1758	Pharmacogenetics: improving drug and dose selection. <b>2008</b> , 8, 639-46	18
1757	Intron delays and transcriptional timing during development. <b>2008</b> , 14, 324-30	81
1756	Population structure in copy number variation and SNPs in the CCL4L chemokine gene. <b>2008</b> , 9, 279-88	19
1755	Rapid large-scale expansion of functional mesenchymal stem cells from unmanipulated bone marrow without animal serum. <b>2008</b> , 14, 185-96	153
1754	Pharmacogenetic testing in psychiatry: a review of features and clinical realities. <b>2008</b> , 28, 599-617	62
1753	A preliminary attempt to personalize risperidone dosing using drug-drug interactions and genetics: part I. <b>2008</b> , 49, 258-70	26

1752	A preliminary attempt to personalize risperidone dosing using drug-drug interactions and genetics: part II. <b>2008</b> , 49, 347-61	37
1751	Variation in inflammation-related genes and risk of incident nonfatal myocardial infarction or ischemic stroke. <b>2008</b> , 198, 166-73	65
1750	Prevalence in the United States of selected candidate gene variants: Third National Health and Nutrition Examination Survey, 1991-1994. <b>2009</b> , 169, 54-66	72
1749	Genetics of infantile seizures with paroxysmal dyskinesia: the infantile convulsions and choreoathetosis (ICCA) and ICCA-related syndromes. <b>2008</b> , 45, 773-9	22
1748	Genetic mapping in human disease. <b>2008</b> , 322, 881-8	1086
1747	A segmental maximum a posteriori approach to genome-wide copy number profiling. <b>2008</b> , 24, 751-8	28
1746	Considerations regarding the genetics of obesity. <b>2008</b> , 16 Suppl 3, S33-9	33
1745	Genomic deletions correlate with underexpression of novel candidate genes at six loci in pediatric pilocytic astrocytoma. <b>2008</b> , 10, 757-72	33
1744	Schizophrenia aetiology: do gene-environment interactions hold the key?. <b>2008</b> , 102, 21-6	50
1743	Whole population, genome-wide mapping of hidden relatedness. <b>2009</b> , 19, 318-26	326
1742	Copy number variation and evolution in humans and chimpanzees. <b>2008</b> , 18, 1698-710	178
1741	Trypsinogen copy number mutations in patients with idiopathic chronic pancreatitis. <b>2008</b> , 6, 82-8	65
1740	Analysis of protocadherin alpha gene enhancer polymorphism in bipolar disorder and schizophrenia. <b>2008</b> , 102, 210-9	51
1739	Molecular subtypes of diffuse large B-cell lymphoma arise by distinct genetic pathways. <b>2008</b> , 105, 13520-5	746
1738	A binary format for genetic data designed for large whole genome studies that enable both marker and strand based analyses. <b>2008</b> ,	
1737	Chromosome copy number variation and breast cancer risk. <b>2008</b> , 123, 183-7	14
1736	The quality of DNA extracted from liquid or dried blood is not adversely affected by storage at 4 degrees C for up to 24 h. <b>2008</b> , 37 Suppl 1, i7-10	18
1735	Excessive genomic DNA copy number variation in the Li-Fraumeni cancer predisposition syndrome. <b>2008</b> , 105, 11264-9	155

1734	Experimental design and data analysis for array comparative genomic hybridization. <b>2008</b> , 26, 923-8	18
1733	Defensins and the dynamic genome: what we can learn from structural variation at human chromosome band 8p23.1. <b>2008</b> , 18, 1686-97	68
1732	Paternal genetic and epigenetic influences on IVF outcome. <b>2008</b> , 3, 359-367	2
1731	Bayesian detection of recurrent copy number alterations across multiple array samples. <b>2008</b> ,	1
1730	High frequency of submicroscopic genomic aberrations detected by tiling path array comparative genome hybridisation in patients with isolated congenital heart disease. <b>2008</b> , 45, 704-9	102
1729	The genomic architecture of segmental duplications and associated copy number variants in dogs. <b>2009</b> , 19, 491-9	122
1728	Quencher-free molecular beacons: a new strategy in fluorescence based nucleic acid analysis. <b>2008</b> , 37, 648-63	182
1727	Estimation and assessment of raw copy numbers at the single locus level. <b>2008</b> , 24, 759-67	120
1726	Browsing HapMap Data Using the Genome Browser. <b>2008</b> , 2008, pdb.prot5023	7
1725	Gene copy number variations in Asian patients with congenital bilateral absence of the vas deferens. <b>2009</b> , 24, 748-55	20
1724	Single nucleotide polymorphism-based genome-wide chromosome copy change, loss of heterozygosity, and aneuploidy in Barrett's esophagus neoplastic progression. <b>2008</b> , 1, 413-23	65
1723	Pharmacogenomics and drug toxicity. <b>2008</b> , 359, 856-8	35
1722	Mapping DNA structural variation in dogs. <b>2009</b> , 19, 500-9	109
1721	Extensive genomic copy number variation in embryonic stem cells. <b>2008</b> , 105, 17453-6	64
1720	MOCSphaser: a haplotype inference tool from a mixture of copy number variation and single nucleotide polymorphism data. <b>2008</b> , 24, 1645-6	14
1719	Association of breast cancer resistance protein/ABCG2 phenotypes and novel promoter and intron 1 single nucleotide polymorphisms. <b>2008</b> , 36, 780-95	65
1718	Copy-number variations associated with autism spectrum disorder. <b>2008</b> , 9, 1143-54	12
1717	Analysis of copy number variation in the rhesus macaque genome identifies candidate loci for evolutionary and human disease studies. <b>2008</b> , 17, 1127-36	96

1716	Comparative Genomics. <b>2008</b> ,	2
1715	Segmental duplications and evolutionary plasticity at tumor chromosome break-prone regions. <b>2008</b> , 18, 370-9	37
1714	Germline copy number variation in control populations. <b>2008</b> , 123, 211-23	1
1713	Copy number variation in metabolic phenotypes. <b>2008</b> , 123, 169-75	16
1712	Copy variations in schizophrenia and bipolar disorder. <b>2008</b> , 123, 27-35	7
1711	HIV-1/AIDS susceptibility and copy number variation in CCL3L1, a gene encoding a natural ligand for HIV-1 co-receptor CCR5. <b>2008</b> , 123, 156-60	29
1710	CNVs of human genes and their implication in pharmacogenetics. <b>2008</b> , 123, 195-204	25
1709	Human copy number polymorphic genes. <b>2008</b> , 123, 234-43	37
1708	Copy number variation on the human Y chromosome. <b>2008</b> , 123, 253-62	51
1707	Genomic drift and copy number variation of chemosensory receptor genes in humans and mice. <b>2008</b> , 123, 263-9	11
1706	Expression divergence and copy number variation in the human genome. <b>2008</b> , 123, 278-82	3
1705	The evolutionary significance of copy number variation in the human genome. <b>2008</b> , 123, 283-7	19
1704	Comparative analysis of copy number variation in primate genomes. <b>2008</b> , 123, 288-96	3
1703	Detection, breakpoint identification and detailed characterisation of a CNV at the FRA16D site using SNP assays. <b>2008</b> , 123, 322-32	4
1702	Human genes involved in copy number variation: mechanisms of origin, functional effects and implications for disease. <b>2008</b> , 123, 17-26	53
1701	Pharmacogenomics: candidate gene identification, functional validation and mechanisms. <b>2008</b> , 17, R174-9	49
1700	Chromosomal breakpoints in primary colon cancer cluster at sites of structural variants in the genome. <b>2008</b> , 68, 1284-95	63
1699	Construction, characterization and FISH mapping of a bacterial artificial chromosome library of Chinese pangolin ( <i>Manis pentadactyla</i> ). <b>2008</b> , 122, 55-60	4



1698	The role of rare structural variants in the genetics of autism spectrum disorders. <b>2008</b> , 123, 36-43	21
1697	Molecular and clinical characterization of de novo and familial cases with microduplication 3q29: guidelines for copy number variation case reporting. <b>2008</b> , 123, 65-78	35
1696	Benign copy number changes in clinical cytogenetic diagnostics by array CGH. <b>2008</b> , 123, 94-101	9
1695	The emerging role of structural variations in common disorders: initial findings and discovery challenges. <b>2008</b> , 123, 108-17	5
1694	Phenotypes, genotypes and disease susceptibility associated with gene copy number variations: complement C4 CNVs in European American healthy subjects and those with systemic lupus erythematosus. <b>2008</b> , 123, 131-41	51
1693	Copy number variation of Fc gamma receptor genes and disease predisposition. <b>2008</b> , 123, 161-8	19
1692	Large-scale copy number variants (CNVs) detected in different ethnic human populations. <b>2008</b> , 123, 224-33	4
1691	Human subtelomeric copy number variations. <b>2008</b> , 123, 244-52	32
1690	Copy number variation in the mouse genome: implications for the mouse as a model organism for human disease. <b>2008</b> , 123, 297-306	16
1689	Inducing segmental aneuploid mosaicism in the mouse through targeted asymmetric sister chromatid event of recombination. <b>2008</b> , 180, 51-9	13
1688	Genome study of kidney disease in the age of post genome-sequencing. <b>2008</b> , 8, 173-83	5
1687	Clinical puzzle: Barrett's oesophagus. <b>2008</b> , 1, 26-31	7
1686	Copy number variation in the autism genome. <b>2008</b> , 2, 417-28	1
1685	Constitutional DNA copy number changes in ICSI children. <b>2009</b> , 24, 233-40	9
1684	Effect of genetic divergence in identifying ancestral origin using HAPAA. <b>2008</b> , 18, 676-82	56
1683	Analysis of copy number variants and segmental duplications in the human genome: Evidence for a change in the process of formation in recent evolutionary history. <b>2008</b> , 18, 1865-74	114
1682	Copy number alterations in pancreatic cancer identify recurrent PAK4 amplification. <b>2008</b> , 7, 1793-802	101
1681	Human trash ESTs--sequences from cDNA collection that are not aligned to genome assembly. <b>2008</b> , 6, 759-73	3

1680	Genetic variants of the copy number polymorphic beta-defensin locus are associated with sporadic prostate cancer. <b>2008</b> , 29, 83-92	22
1679	LPA and PLG sequence variation and kringle IV-2 copy number in two populations. <b>2008</b> , 66, 199-209	26
1678	DNA Polymorphisms: Tools for Detection. <b>2008</b> , 1	
1677	High-resolution mapping of DNA breakpoints to define true recurrences among ipsilateral breast cancers. <b>2008</b> , 100, 48-58	54
1676	African genetic diversity: implications for human demographic history, modern human origins, and complex disease mapping. <b>2008</b> , 9, 403-33	472
1675	Regulation of Gene Expression in the Tumor Environment. <b>2008</b> ,	
1674	Different selective pressures shape the molecular evolution of color vision in chimpanzee and human populations. <b>2008</b> , 25, 2735-43	31
1673	Small-scale copy number variation and large-scale changes in gene expression. <b>2008</b> , 105, 16659-64	71
1672	DupMasker: a tool for annotating primate segmental duplications. <b>2008</b> , 18, 1362-8	24
1671	CNVs and genetic medicine (excitement and consequences of a rediscovery). <b>2008</b> , 123, 7-16	16
1670	The dynamic nature of eukaryotic genomes. <b>2008</b> , 25, 787-94	106
1669	CCL3L1 gene-containing segmental duplications and polymorphisms in CCR5 affect risk of systemic lupus erythaematosus. <b>2008</b> , 67, 1076-83	77
1668	Sequencing human-gibbon breakpoints of synteny reveals mosaic new insertions at rearrangement sites. <b>2009</b> , 19, 178-90	26
1667	On the frequency of copy number variants. <b>2008</b> , 24, 2350-5	5
1666	GenoSNP: a variational Bayes within-sample SNP genotyping algorithm that does not require a reference population. <b>2008</b> , 24, 2209-14	56
1665	DNA sequence-based phenotypic association analysis. <b>2008</b> , 60, 195-217	16
1664	Haploinsufficiency of DNA Damage Response Genes and their Potential Influence in Human Genomic Disorders. <b>2008</b> , 9, 137-46	13
1663	High-resolution copy-number variation map reflects human olfactory receptor diversity and evolution. <b>2008</b> , 4, e1000249	87

1662	Repetitive element-mediated recombination as a mechanism for new gene origination in <i>Drosophila</i> . <b>2008</b> , 4, e3	66
1661	CNVDetector: locating copy number variations using array CGH data. <b>2008</b> , 24, 2773-5	10
1660	The pressure of finding human hypertension genes: new tools, old dilemmas. <b>2008</b> , 22, 821-8	23
1659	CGHweb: a tool for comparing DNA copy number segmentations from multiple algorithms. <b>2008</b> , 24, 1014-5	41
1658	A novel mechanistic spectrum underlies glaucoma-associated chromosome 6p25 copy number variation. <b>2008</b> , 17, 3446-58	33
1657	Complex rearrangements lead to novel chimeric gene fusion polymorphisms at the <i>Arabidopsis thaliana</i> MAF2-5 flowering time gene cluster. <b>2009</b> , 26, 699-711	35
1656	Detection of copy number alterations in metastatic melanoma by a DNA fluorescence in situ hybridization probe panel and array comparative genomic hybridization: a southwest oncology group study (S9431). <b>2008</b> , 14, 2927-35	42
1655	Genetic analysis of human traits in vitro: drug response and gene expression in lymphoblastoid cell lines. <b>2008</b> , 4, e1000287	182
1654	Segmental duplications arise from Pol32-dependent repair of broken forks through two alternative replication-based mechanisms. <b>2008</b> , 4, e1000175	145
1653	Molecular cytogenetics and cytogenomics of brain diseases. <b>2008</b> , 9, 452-65	65
1652	Adaptive copy number evolution in malaria parasites. <b>2008</b> , 4, e1000243	144
1651	Lack of C20orf133 and FLRT3 mutations in 43 patients with Kabuki syndrome in Japan. <b>2008</b> , 45, 479-80	10
1650	Reduced purifying selection prevails over positive selection in human copy number variant evolution. <b>2008</b> , 18, 1711-23	64
1649	Copy number variations and risk for schizophrenia in 22q11.2 deletion syndrome. <b>2008</b> , 17, 4045-53	132
1648	Copy number variation at the 7q11.23 segmental duplications is a susceptibility factor for the Williams-Beuren syndrome deletion. <b>2008</b> , 18, 683-94	55
1647	Genetic loci associated with plasma concentration of low-density lipoprotein cholesterol, high-density lipoprotein cholesterol, triglycerides, apolipoprotein A1, and Apolipoprotein B among 6382 white women in genome-wide analysis with replication. <b>2008</b> , 1, 21-30	103
1646	A fast and flexible method for the segmentation of aCGH data. <b>2008</b> , 24, i139-45	53
1645	A naturally occurring gene amplification leading to sulfonamide and trimethoprim resistance in <i>Streptococcus agalactiae</i> . <b>2008</b> , 190, 672-80	40

1644	Copy number variation at the breakpoint region of isochromosome 17q. <b>2008</b> , 18, 1724-32	41
1643	THE APPLICATION OF MICROARRAY BASED COMPARATIVE GENOMIC HYBRIDIZATION IN PRENATAL DIAGNOSIS. <b>2008</b> , 19, 119-133	3
1642	Transcriptome sequencing of malignant pleural mesothelioma tumors. <b>2008</b> , 105, 3521-6	126
1641	The genetics of SLE: an update in the light of genome-wide association studies. <b>2008</b> , 47, 1603-11	93
1640	wuHMM: a robust algorithm to detect DNA copy number variation using long oligonucleotide microarray data. <b>2008</b> , 36, e41	21
1639	Analysis of genome-wide copy number variation in Irish and Dutch ALS populations. <b>2008</b> , 17, 3392-8	37
1638	Modeling genetic inheritance of copy number variations. <b>2008</b> , 36, e138	60
1637	How segmental duplications shape our genome: recent evolution of ABCC6 and PKD1 Mendelian disease genes. <b>2008</b> , 25, 2601-13	31
1636	Whole-genome amplification enables accurate genotyping for microarray-based high-density single nucleotide polymorphism array. <b>2008</b> , 17, 3499-508	19
1635	Hidden copy number variation in the HapMap population. <b>2008</b> , 105, 10067-72	5
1634	Perilobar nephrogenic rests are nonobligate molecular genetic precursor lesions of insulin-like growth factor-II-associated Wilms tumors. <b>2008</b> , 14, 7635-44	22
1633	Analysis of copy number variation using quantitative interspecies competitive PCR. <b>2008</b> , 36, e112	9
1632	Selection on major components of angiosperm genomes. <b>2008</b> , 320, 484-6	56
1631	Extending genome-wide association studies to copy-number variation. <b>2008</b> , 17, R135-42	133
1630	Familial pulmonary alveolar proteinosis caused by mutations in CSF2RA. <b>2008</b> , 205, 2703-10	229
1629	Copy number of FCGR3B, which is associated with systemic lupus erythematosus, correlates with protein expression and immune complex uptake. <b>2008</b> , 205, 1573-82	183
1628	Update on the genetics of stroke and cerebrovascular disease 2007. <b>2008</b> , 39, 252-4	5
1627	Frequency of germline genomic homozygosity associated with cancer cases. <b>2008</b> , 299, 1437-45	40

1626	Bacterial artificial chromosome-emulation oligonucleotide arrays for targeted clinical array-comparative genomic hybridization analyses. <b>2008</b> , 10, 278-89	86
1625	CCAAT/enhancer binding protein alpha (C/EBPalpha) in adipose tissue regulates genes in lipid and glucose metabolism and a genetic variation in C/EBPalpha is associated with serum levels of triglycerides. <b>2008</b> , 93, 4880-6	54
1624	Enhanced detection of clinically relevant genomic imbalances using a targeted plus whole genome oligonucleotide microarray. <b>2008</b> , 10, 415-29	132
1623	Sparse representation and Bayesian detection of genome copy number alterations from microarray data. <b>2008</b> , 24, 309-18	99
1622	Population genetics of human copy number variations. <b>2008</b> ,	
1621	Is gene discovery research or diagnosis?. <b>2008</b> , 10, 385-90	7
1620	Smoking-related genomic signatures in non-small cell lung cancer. <b>2008</b> , 178, 1164-72	33
1619	Ancestry-related differences in gene expression: findings may enhance understanding of health disparities between populations. <b>2008</b> , 9, 489-92	13
1618	Ultraconserved elements: analyses of dosage sensitivity, motifs and boundaries. <b>2008</b> , 180, 2277-93	28
1617	Regulatory variation and evolution: implications for disease. <b>2008</b> , 61, 295-306	2
1616	Global patterns of variation in allele and haplotype frequencies and linkage disequilibrium across the CYP2E1 gene. <b>2008</b> , 8, 349-56	16
1615	Immunology, Phenotype First: How Mutations Have Established New Principles and Pathways in Immunology. <b>2008</b> ,	1
1614	Expression profiles of genes regulating dairy cow fertility: recent findings, ongoing activities and future possibilities. <b>2008</b> , 2, 1158-67	16
1613	Genetic insights into sepsis: what have we learned and how will it help?. <b>2008</b> , 14, 1900-11	21
1612	Parental-origin-determination fluorescence in situ hybridization distinguishes homologous human chromosomes on a single-cell level. <b>2008</b> , 21, 189-200	26
1611	Contemplating effects of genomic structural variation. <b>2008</b> , 10, 639-47	65
1610	Statistical issues in the analysis of DNA Copy Number Variations. <b>2008</b> , 1, 368-95	20
1609	Chromosomal lesions and uniparental disomy detected by SNP arrays in MDS, MDS/MPD, and MDS-derived AML. <b>2008</b> , 111, 1534-42	281

1608	A genome-wide scan for common genetic variants with a large influence on warfarin maintenance dose. <b>2008</b> , 112, 1022-7	368
1607	Pathogenesis of the systemic inflammatory syndrome and acute lung injury: role of iron mobilization and decompartmentalization. <b>2008</b> , 294, L161-74	38
1606	Hidden Markov models for the assessment of chromosomal alterations using high-throughput SNP arrays. <b>2008</b> , 2, 687-713	43
1605	Bibliography. <b>2008</b> , 383-412	
1604	Structure of the NF1 Gene Region and Mechanisms Underlying Gross NF1 Deletions. <b>2008</b> , 89-102	1
1603	New insights into the genetics of body weight. <b>2008</b> , 11, 378-84	19
1602	Systemic lupus erythematosus genetics: what's new?. <b>2008</b> , 3, 103-107	1
1601	Germ Line Polymorphisms and Drug Response. 329-383	
1600	Genetic Epidemiology and Molecular Genetics of Psychiatric Disorders. <b>2008</b> , 257-274	
1599	The HapMap Resource is Providing New Insights into Ourselves and its Application to Pharmacogenomics. <b>2008</b> , 2, 15-23	43
1598	Epistasis between the MHC and the RCA alpha block in primary Sjögren syndrome. <b>2008</b> , 67, 849-54	10
1597	Medical genetics diagnostic evaluation of the child with global developmental delay or intellectual disability. <b>2008</b> , 21, 117-22	41
1596	Oligonucleotide microarrays for clinical diagnosis of copy number variation. <b>2008</b> , Chapter 8, Unit 8.12	6
1595	Hypertension and genome-wide association studies: combining high fidelity phenotyping and hypercontrols. <b>2008</b> , 26, 1275-81	29
1594	The fat tail of obesity as told by the genome. <b>2008</b> , 11, 366-70	19
1593	Analysis of protocadherin alpha gene deletion variant in bipolar disorder and schizophrenia. <b>2008</b> , 18, 110-5	16
1592	. <b>2008</b> ,	1
1591	Design Issues in Toxicogenomics: The Application of Genomic Technologies for Mechanistic and Predictive Research. 87-99	

1590 . 2008,

2

1589 Microarrays in Systems Neurobiology and Translational Neuroscience [From Genome Research to Clinical Applications. 1-34

1588 Rejoinder: Quantifying the Fraction of Missing Information for Hypothesis Testing in Statistical and Genetic Studies. 2008, 23,

1587 Biomonitoring of human exposure to prestige oil: effects on DNA and endocrine parameters. 2008, 2, 83-92

18

1586 Concordant gene expression in leukemia cells and normal leukocytes is associated with germline cis-SNPs. 2008, 3, e2144

6

1585 Mathematical analysis of copy number variation in a DNA sample using digital PCR on a nanofluidic device. 2008, 3, e2876

211

1584 Small deletion variants have stable breakpoints commonly associated with alu elements. 2008, 3, e3104

49

1583 Functional copy-number alterations in cancer. 2008, 3, e3179

129

1582 Computing power and sample size for case-control association studies with copy number polymorphism: application of mixture-based likelihood ratio test. 2008, 3, e3475

12

1581 Quantitative analysis of single nucleotide polymorphisms within copy number variation. 2008, 3, e3906

27

1580 A snapshot of CNVs in the pig genome. 2008, 3, e3916

97

1579 Principios de citogenética clínica. 2008, 59-83

1578 ?!? HIV/AIDS?????????????. 2008, 15, 39-50

1577 Inverse relationship between genetic diversity and epigenetic complexity. 2008,

1

1576 3rd Asia Pacific Nutrigenomics Conference. 2008, 1, 179-195

1575 Integrating Epigenomics into Pharmacogenomic Studies. 2008, 2008, 7-14

18

1574 A HapMap harvest of insights into the genetics of common disease. 2008, 118, 1590-605

683

1573 . 2009,

3

1572 Concepts of Population Genomics. **2009**, 22-32

1571 Comparing two diagnostic laboratory tests for several microdeletions causing mental retardation syndromes: multiplex ligation-dependent amplification vs fluorescent in situ hybridization. **2009**, 29, 71-6 17

1570 Combining chromosomal arm status and significantly aberrant genomic locations reveals new cancer subtypes. **2009**, 7, 91-104

1569 Unusual Types of  $\beta$ -thalassemia. 296-320 1

1568 Comparison of multiplex ligation-dependent probe amplification and real-time PCR accuracy for gene copy number quantification using the beta-defensin locus. **2009**, 47, 1023-8 21

1567 Linkage disequilibrium between two high-frequency deletion polymorphisms: implications for association studies involving the glutathione-S transferase (GST) genes. **2009**, 5, e1000472 38

1566 Maize inbreds exhibit high levels of copy number variation (CNV) and presence/absence variation (PAV) in genome content. **2009**, 5, e1000734 389

1565 Androgen receptor copy number variation and androgenetic alopecia: a case-control study. **2009**, 4, e5081 12

1564 Fidelity of SNP array genotyping using Epstein Barr virus-transformed B-lymphocyte cell lines: implications for genome-wide association studies. **2009**, 4, e6915 30

1563 A genome-wide analysis of array-based comparative genomic hybridization (CGH) data to detect intra-species variations and evolutionary relationships. **2009**, 4, e7978 10

1562 Fetal karyotyping: what should we be offering and how?. 147-158

1561 Possible genetic polymorphisms related to sex steroid metabolism and dementia in women. 143-152

1560 Genetics related to sex steroids: implications for Alzheimer's disease. 153-161

1559 Inverse relationship between genetic diversity and epigenetic complexity. **2009**, 14 14

1558  $\beta$ KGEM: **2009**, 371-382 1

1557 Epigenetics, genomic mutations and cognitive function. **2009**, 14, 377-90 28

1556 Methods for optimizing statistical analyses in pharmacogenomics research. **2009**, 2, 559-570 8

1555 Cancer cytogenetics in the zebrafish. **2009**, 6, 355-60 9



1554	Prediction of cancer outcome using DNA microarray technology: past, present and future. <b>2009</b> , 3, 157-65	11
1553	Clinical application of microarray-based comparative genomic hybridization in prenatal diagnosis. <b>2009</b> , 4, 81-92	
1552	The birth of new genes by RNA- and DNA-mediated duplication during mammalian evolution. <b>2009</b> , 16, 1429-44	7
1551	Nature's genetic gradients and the clinical phenotype. <b>2009</b> , 2, 537-9	26
1550	Allelic recombination between distinct genomic locations generates copy number diversity in human beta-defensins. <b>2009</b> , 106, 853-8	53
1549	Application of signal processing techniques for estimating regions of copy number variations in human meningioma DNA. <b>2009</b> , 2009, 6973-6	1
1548	Joint estimation of copy number variation and reference intensities on multiple DNA arrays using GADA. <b>2009</b> , 25, 1223-30	35
1547	ABYSS: a parallel assembler for short read sequence data. <b>2009</b> , 19, 1117-23	2508
1546	Pharmacogenomics: the promise of personalized medicine for CNS disorders. <b>2009</b> , 34, 159-72	65
1545	Microarray-based genomic DNA profiling technologies in clinical molecular diagnostics. <b>2009</b> , 55, 659-69	47
1544	Copy number variants, diseases and gene expression. <b>2009</b> , 18, R1-8	324
1543	A comprehensive profile of DNA copy number variations in a Korean population: identification of copy number invariant regions among Koreans. <b>2009</b> , 41, 618-28	8
1542	Genomic duplication resulting in increased copy number of genes encoding the sister chromatid cohesion complex conveys clinical consequences distinct from Cornelia de Lange. <b>2009</b> , 46, 626-34	44
1541	Refining the 22q11.2 deletion breakpoints in DiGeorge syndrome by aCGH. <b>2009</b> , 124, 113-20	53
1540	Aneuploidy: from a physiological mechanism of variance to Down syndrome. <b>2009</b> , 89, 887-920	86
1539	Population genomic inferences from sparse high-throughput sequencing of two populations of <i>Drosophila melanogaster</i> . <b>2009</b> , 1, 449-65	49
1538	High-resolution mapping and analysis of copy number variations in the human genome: a data resource for clinical and research applications. <b>2009</b> , 19, 1682-90	293
1537	Global distribution of genomic diversity underscores rich complex history of continental human populations. <b>2009</b> , 19, 795-803	129

1536	Integrated study of copy number states and genotype calls using high-density SNP arrays. <b>2009</b> , 37, 5365-77	93
1535	Single nucleotide polymorphism arrays: a decade of biological, computational and technological advances. <b>2009</b> , 37, 4181-93	270
1534	Hybridization modeling of oligonucleotide SNP arrays for accurate DNA copy number estimation. <b>2009</b> , 37, e117	11
1533	A geometric approach for classification and comparison of structural variants. <b>2009</b> , 25, i222-30	123
1532	Characterization of six human disease-associated inversion polymorphisms. <b>2009</b> , 18, 2555-66	97
1531	Rare pathogenic microdeletions and tandem duplications are microhomology-mediated and stimulated by local genomic architecture. <b>2009</b> , 18, 3579-93	126
1530	Evolution of F-box genes in plants: different modes of sequence divergence and their relationships with functional diversification. <b>2009</b> , 106, 835-40	215
1529	[Copy-number variation: a new pattern of structural diversity in genome]. <b>2009</b> , 31, 339-47	
1528	Genome-wide Association Analysis Based on Copy Number Variations*. <b>2009</b> , 36, 968-977	1
1527	Organization, Variation and Expression of the Human Genome as a Foundation of Genomic and Personalized Medicine. <b>2009</b> , 4-21	7
1526	Quantitative real-time polymerase chain reaction for the verification of genomic imbalances detected by microarray-based comparative genomic hybridization. <b>2009</b> , 13, 751-60	23
1525	Whirly proteins maintain plastid genome stability in Arabidopsis. <b>2009</b> , 106, 14693-8	152
1524	Identification of small gains and losses in single cells after whole genome amplification on tiling oligo arrays. <b>2009</b> , 37, e105	59
1523	OKCAM: an ontology-based, human-centered knowledgebase for cell adhesion molecules. <b>2009</b> , 37, D251-60	17
1522	Targets of balancing selection in the human genome. <b>2009</b> , 26, 2755-64	199
1521	Array comparative genomic hybridization profiling analysis reveals deoxyribonucleic acid copy number variations associated with premature ovarian failure. <b>2009</b> , 94, 4540-6	66
1520	Genetic association studies in ischaemic stroke: replication failure and prospects. <b>2009</b> , 27, 290-4	20
1519	An alternative approach to medical genetics based on modern evolutionary biology. Part 2: retroviral symbiosis. <b>2009</b> , 102, 324-31	7

1518	DNA copy number profiles of primary tumors as predictors of response to chemotherapy in advanced colorectal cancer. <b>2009</b> , 20, 1048-56	36
1517	Genomics and Pharmacogenomics in Anticancer Drug Development and Clinical Response. <b>2009</b> ,	2
1516	Epigenetic profiling in schizophrenia and major mental disorders. <b>2009</b> , 60, 5-11	21
1515	Personalized clinical laboratory diagnostics. <b>2009</b> , 47, 95-119	11
1514	Array comparative genomic hybridisation in clinical diagnostics: principles and applications / Array-CGH in der klinischen Diagnostik: Prinzipien und Anwendungen. <b>2009</b> , 33, 255-266	
1513	The chromosomal constitution of postmitotic neurons, assessed by neuronal nuclear transfer into oocytes and in ES cell lines derived from them. <b>2009</b> , 125, 201-12	2
1512	High-throughput multiplex sequencing to discover copy number variants in Drosophila. <b>2009</b> , 182, 935-41	22
1511	Markov Models for inferring copy number variations from genotype data on Illumina platforms. <b>2009</b> , 68, 1-22	12
1510	Genome-wide Mapping of Copy Number Variations Using SNP Arrays. <b>2009</b> , 36, 246-251	8
1509	The 'Whole Genome Age'. <b>2009</b> , 36, 244-245	
1508	Copy number variation of testis-specific protein, Y-encoded (TSPY) in 14 different breeds of cattle ( <i>Bos taurus</i> ). <b>2009</b> , 3, 205-13	37
1507	Preimplantation genetic diagnosis using fluorescent in situ hybridization for cancer predisposition syndromes caused by microdeletions. <b>2009</b> , 24, 1522-8	17
1506	Human-specific genes may offer a unique window into human cell signaling. <b>2009</b> , 2, pe59	28
1505	Copy number variation and susceptibility to human disorders (Review). <b>2009</b> , 2, 143-7	11
1504	Genome-wide association study of acute post-surgical pain in humans. <b>2009</b> , 10, 171-9	56
1503	Pathogenesis of autism: a patchwork of genetic causes. <b>2009</b> , 4, 591-599	7
1502	Copy number variation in intron 1 of SOX5 causes the Pea-comb phenotype in chickens. <b>2009</b> , 5, e1000512	162
1501	Novel sequence variations in the brain-derived neurotrophic factor gene and association with major depression and antidepressant treatment response. <b>2009</b> , 66, 488-97	137

1500	Recurrent reciprocal deletions and duplications of 16p13.11: the deletion is a risk factor for MR/MCA while the duplication may be a rare benign variant. <b>2009</b> , 46, 223-32	210
1499	A 3-bp deletion of mitochondrial DNA tRNALys observed in lymphoblastoid cells. <b>2009</b> , 54, 612-3	2
1498	A Bayesian segmentation approach to ascertain copy number variations at the population level. <b>2009</b> , 25, 1669-79	15
1497	Genetic control of global gene expression levels in the intestinal mucosa: a human twin study. <b>2009</b> , 38, 73-9	19
1496	Analysis of a large cluster of SLC22 transporter genes, including novel USTs, reveals species-specific amplification of subsets of family members. <b>2009</b> , 38, 116-24	22
1495	Genetic architecture of coronary artery disease in the genome-wide era: implications for the emerging "golden dozen" loci. <b>2009</b> , 35, 671-82	29
1494	Genotype to phenotype-discovery and characterization of novel genomic disorders in a "genotype-first" era. <b>2009</b> , 11, 836-42	30
1493	Whole genome scanning: resolving clinical diagnosis and management amidst complex data. <b>2009</b> , 66, 357-63	54
1492	Recent advances in coeliac disease genetics. <b>2009</b> , 58, 473-6	19
1491	A single-array preprocessing method for estimating full-resolution raw copy numbers from all Affymetrix genotyping arrays including GenomeWideSNP 5 & 6. <b>2009</b> , 25, 2149-56	126
1490	Forging links between human mental retardation-associated CNVs and mouse gene knockout models. <b>2009</b> , 5, e1000531	39
1489	A microhomology-mediated break-induced replication model for the origin of human copy number variation. <b>2009</b> , 5, e1000327	592
1488	Integrated associations of genotypes with multiple blood biomarkers linked to coronary heart disease risk. <b>2009</b> , 18, 2305-16	39
1487	CCL3L Copy number variation and the co-evolution of primate and viral genomes. <b>2009</b> , 5, e1000359	9
1486	Meiotic recombination generates rich diversity in NK cell receptor genes, alleles, and haplotypes. <b>2009</b> , 19, 757-69	87
1485	Sequencing genomes: from individuals to populations. <b>2009</b> , 8, 367-78	13
1484	A novel approach to detect copy number variation using segmentation and genetic algorithm. <b>2009</b> , ,	
1483	SNPNexus: a web database for functional annotation of newly discovered and public domain single nucleotide polymorphisms. <b>2009</b> , 25, 655-61	146

1482	Identification of candidate genes for sporadic amyotrophic lateral sclerosis by array comparative genomic hybridization. <b>2009</b> , 10, 162-9	12
1481	Evolution of olfactory receptor genes in primates dominated by birth-and-death process. <b>2009</b> , 1, 258-64	31
1480	Copy number variation in the Framingham Heart Study. <b>2009</b> , 3 Suppl 7, S133	4
1479	Genomic and transcriptional co-localization of protein-coding and long non-coding RNA pairs in the developing brain. <b>2009</b> , 5, e1000617	305
1478	Integrating sequencing technologies in personal genomics: optimal low cost reconstruction of structural variants. <b>2009</b> , 5, e1000432	13
1477	Genome-wide association study suggested copy number variation may be associated with body mass index in the Chinese population. <b>2009</b> , 54, 199-202	70
1476	A single-sample method for normalizing and combining full-resolution copy numbers from multiple platforms, labs and analysis methods. <b>2009</b> , 25, 861-7	32
1475	Disruption of the neurexin 1 gene is associated with schizophrenia. <b>2009</b> , 18, 988-96	376
1474	Contribution of copy number variation in the regulation of complement activation locus to development of age-related macular degeneration. <b>2009</b> , 50, 5070-9	39
1473	The evolution of meiosis from mitosis. <b>2009</b> , 181, 3-12	105
1472	Copy number variation influences gene expression and metabolic traits in mice. <b>2009</b> , 18, 4118-29	79
1471	Expression quantitative trait loci detected in cell lines are often present in primary tissues. <b>2009</b> , 18, 4296-303	41
1470	A common copy number variation on chromosome 6 association with the gene expression level of endothelin 1 in transformed B lymphocytes from three racial groups. <b>2009</b> , 2, 483-8	7
1469	Genomewide association studies: history, rationale, and prospects for psychiatric disorders. <b>2009</b> , 166, 540-56	355
1468	MALDI-TOF MS genotyping of polymorphisms related to 1-carbon metabolism using common and mass-modified terminators. <b>2009</b> , 55, 139-49	11
1467	Using reads to annotate the genome: influence of length, background distribution, and sequence errors on prediction capacity. <b>2009</b> , 37, e104	17
1466	An alternative approach to medical genetics based on modern evolutionary biology. Part 5: epigenetics and genomics. <b>2009</b> , 102, 531-7	2
1465	Genome-wide comparisons of variation in linkage disequilibrium. <b>2009</b> , 19, 1849-60	53

1464	Chromosomal instability and copy number alterations in Barrett's esophagus and esophageal adenocarcinoma. <b>2009</b> , 15, 3305-14	85
1463	Association of a germ-line copy number variation at 2p24.3 and risk for aggressive prostate cancer. <b>2009</b> , 69, 2176-9	68
1462	Microalterations of inherently unstable genomic regions in rat mammary carcinomas as revealed by long oligonucleotide array-based comparative genomic hybridization. <b>2009</b> , 69, 5159-67	12
1461	Distinguishing among evolutionary models for the maintenance of gene duplicates. <b>2009</b> , 100, 605-17	264
1460	Segmental duplications mediate novel, clinically relevant chromosome rearrangements. <b>2009</b> , 18, 2957-62	52
1459	Copy-number variation: the end of the human genome?. <b>2009</b> , 27, 448-54	44
1458	Complex human chromosomal and genomic rearrangements. <b>2009</b> , 25, 298-307	209
1457	Robustness: mechanisms and consequences. <b>2009</b> , 25, 395-403	249
1456	Rare structural variants in schizophrenia: one disorder, multiple mutations; one mutation, multiple disorders. <b>2009</b> , 25, 528-35	213
1455	Copy-number variants in neurodevelopmental disorders: promises and challenges. <b>2009</b> , 25, 536-44	102
1454	A case study of "disorganized development" and its possible relevance to genetic determinants of aging. <b>2009</b> , 130, 350-6	16
1453	Copy number estimation algorithms and fluorescence in situ hybridization to describe copy number alterations in human tumors. <b>2009</b> , 59, 218-28	4
1452	eCOMPAGT -- efficient combination and management of phenotypes and genotypes for genetic epidemiology. <b>2009</b> , 10, 139	6
1451	Accounting for uncertainty when assessing association between copy number and disease: a latent class model. <b>2009</b> , 10, 172	18
1450	Detection of recurrent copy number alterations in the genome: taking among-subject heterogeneity seriously. <b>2009</b> , 10, 308	11
1449	An experimental loop design for the detection of constitutional chromosomal aberrations by array CGH. <b>2009</b> , 10, 380	13
1448	CNV-seq, a new method to detect copy number variation using high-throughput sequencing. <b>2009</b> , 10, 80	416
1447	SNP@Evolution: a hierarchical database of positive selection on the human genome. <b>2009</b> , 9, 221	23

1446	Definition, conservation and epigenetics of housekeeping and tissue-enriched genes. <b>2009</b> , 10, 269	103
1445	Quadruplex MAPH: improvement of throughput in high-resolution copy number screening. <b>2009</b> , 10, 453	3
1444	A comparative analysis of DNA barcode microarray feature size. <b>2009</b> , 10, 471	11
1443	Detection of pathogenic copy number variants in children with idiopathic intellectual disability using 500 K SNP array genomic hybridization. <b>2009</b> , 10, 526	27
1442	Analysis of recent segmental duplications in the bovine genome. <b>2009</b> , 10, 571	67
1441	The pitfalls of platform comparison: DNA copy number array technologies assessed. <b>2009</b> , 10, 588	79
1440	RExPrimer: an integrated primer designing tool increases PCR effectiveness by avoiding 3' SNP-in-primer and mis-priming from structural variation. <b>2009</b> , 10 Suppl 3, S4	17
1439	Genomic sister-disorders of neurodevelopment: an evolutionary approach. <b>2009</b> , 2, 81-100	30
1438	The discovery of genes implicated in myocardial infarction. <b>2009</b> , 7 Suppl 1, 305-7	14
1437	Cancer gene discovery in mouse and man. <b>2009</b> , 1796, 140-61	8
1436	Comment on "Altered DNA copy number in patients with different seizure disorder type: by array-CGH" by Kim HS et al. Brain & Development 2007;29:639-643. <b>2009</b> , 31, 94	
1435	Microfluidic digital PCR enables rapid prenatal diagnosis of fetal aneuploidy. <b>2009</b> , 200, 543.e1-7	78
1434	Neurobeachin (NBEA) is a target of recurrent interstitial deletions at 13q13 in patients with MGUS and multiple myeloma. <b>2009</b> , 37, 234-44	26
1433	Identified hidden genomic changes in mantle cell lymphoma using high-resolution single nucleotide polymorphism genomic array. <b>2009</b> , 37, 937-46	34
1432	Next-generation sequencing approaches in genetic rodent model systems to study functional effects of human genetic variation. <b>2009</b> , 583, 1668-73	16
1431	High-resolution genomic profiling of pediatric lymphoblastic lymphomas reveals subtle differences with pediatric acute lymphoblastic leukemias in the B-lineage. <b>2009</b> , 191, 27-33	25
1430	Severe combined immunodeficiency (SCID) and attention deficit hyperactivity disorder (ADHD) associated with a Coronin-1A mutation and a chromosome 16p11.2 deletion. <b>2009</b> , 131, 24-30	111
1429	Estudio del componente genético de la cardiopatía isquémica: de los estudios de ligamiento al genotipado integral del genoma. <b>2009</b> , 9, 24-38	2

1428 Genetic and Epigenetic Features of Stem Cells. 169-202

1427	Genome-wide association studies and the genetic dissection of complex traits. <b>2009</b> , 84, 504-15	55
1426	Epigenetics and the embodiment of race: developmental origins of US racial disparities in cardiovascular health. <b>2009</b> , 21, 2-15	449
1425	Molecular (SNP) analyses of overlapping hemizygous deletions of 10q25.3 to 10qter in four patients: evidence for HMX2 and HMX3 as candidate genes in hearing and vestibular function. <b>2009</b> , 149A, 669-80	35
1424	Split hand-foot malformation, tetralogy of Fallot, mental retardation and a 1 Mb 19p deletion-evidence for further heterogeneity?. <b>2009</b> , 149A, 975-81	16
1423	A child with terminal 14q deletion syndrome: consideration of genotype-phenotype correlations. <b>2009</b> , 149A, 1012-8	18
1422	Aberrant GRIA3 transcripts with multi-exon duplications in a family with X-linked mental retardation. <b>2009</b> , 149A, 1280-9	35
1421	Mutation analysis of SOX9 and single copy number variant analysis of the upstream region in eight patients with campomelic dysplasia and acampomelic campomelic dysplasia. <b>2009</b> , 149A, 2882-5	16
1420	No pathogenic rearrangement within the DISC 1 gene in psychosis. <b>2009</b> , 150B, 148-50	2
1419	Bayesian EM algorithm for scoring polymorphic deletions from SNP data and application to a common CNV on 8q24. <b>2009</b> , 33, 357-68	7
1418	Adapting the logical basis of tests for Hardy-Weinberg Equilibrium to the real needs of association studies in human and medical genetics. <b>2009</b> , 33, 569-80	6
1417	Emerging themes and new challenges in defining the role of structural variation in human disease. <b>2009</b> , 30, 135-44	73
1416	SNP array mapping of chromosome 20p deletions: genotypes, phenotypes, and copy number variation. <b>2009</b> , 30, 371-8	51
1415	Genomic microarrays in mental retardation: a practical workflow for diagnostic applications. <b>2009</b> , 30, 283-92	126
1414	An integrated approach for measuring copy number variation at the FCGR3 (CD16) locus. <b>2009</b> , 30, 477-84	59
1413	Partial deletion of the MAPT gene: a novel mechanism of FTDP-17. <b>2009</b> , 30, E591-602	32
1412	Copy number variation at the FCGR locus includes FCGR3A, FCGR2C and FCGR3B but not FCGR2A and FCGR2B. <b>2009</b> , 30, E640-50	119
1411	Molecular karyotyping of patients with unexplained mental retardation by SNP arrays: a multicenter study. <b>2009</b> , 30, 1082-92	61



1410	Searching genetic risk factors for schizophrenia and bipolar disorder: learn from the past and back to the future. <b>2009</b> , 30, 1139-52	44
1409	Deletions in the VPS13B (COH1) gene as a cause of Cohen syndrome. <b>2009</b> , 30, E845-54	46
1408	Candidate gene copy number analysis by PCR and multicapillary electrophoresis. <b>2009</b> , 30, 1098-101	9
1407	Microarray-based genomic profiling reveals novel genomic aberrations in follicular lymphoma which associate with patient survival and gene expression status. <b>2009</b> , 48, 39-54	60
1406	Construction and application of a zebrafish array comparative genomic hybridization platform. <b>2009</b> , 48, 155-70	20
1405	Novel mechanisms of gene disruption at the medulloblastoma isodicentric 17p11 breakpoint. <b>2009</b> , 48, 121-31	8
1404	Increase in gene dosage is a mechanism of HIF-1alpha constitutive expression in head and neck squamous cell carcinomas. <b>2009</b> , 48, 441-54	16
1403	Genome-wide high-resolution analysis of DNA copy number alterations in NF1-associated malignant peripheral nerve sheath tumors using 32K BAC array. <b>2009</b> , 48, 897-907	44
1402	The life and death of gene families. <b>2009</b> , 31, 29-39	166
1401	Analysis of severely affected patients with dihydropyrimidine dehydrogenase deficiency reveals large intragenic rearrangements of DPYD and a de novo interstitial deletion del(1)(p13.3p21.3). <b>2009</b> , 125, 581-90	40
1400	Genome-wide association studies in ADHD. <b>2009</b> , 126, 13-50	316
1399	Detection of disease-associated deletions in case-control studies using SNP genotypes with application to rheumatoid arthritis. <b>2009</b> , 126, 303-15	7
1398	Reduced TFAP2A function causes variable optic fissure closure and retinal defects and sensitizes eye development to mutations in other morphogenetic regulators. <b>2009</b> , 126, 791-803	53
1397	Sustained viral activity of epstein-Barr virus contributes to cellular immortalization of lymphoblastoid cell lines. <b>2009</b> , 27, 143-8	21
1396	Population genetic models of duplicated genes. <b>2009</b> , 137, 19-37	51
1395	The genetic architecture of blood pressure variation. <b>2009</b> , 3, 418-425	11
1394	Genetics of ischemic stroke: inheritance of a sporadic disorder. <b>2009</b> , 9, 19-27	5
1393	Development and application of genotyping technologies. <b>2009</b> , 52, 17-23	6

1392	Progress in the detection of human genome structural variations. <b>2009</b> , 52, 560-7	9
1391	Novel copy number variants in children with autism and additional developmental anomalies. <b>2009</b> , 1, 292-301	31
1390	[Progress in locating the genetic causes of schizophrenia]. <b>2009</b> , 80, 6, 8, 10-1	3
1389	Unravelling the genetic basis of renal diseases; from single gene to multifactorial disorders. <b>2010</b> , 220, 198-216	25
1388	High-resolution array genomic hybridization in prenatal diagnosis. <b>2009</b> , 29, 20-8	61
1387	Copy number variation in African Americans. <b>2009</b> , 10, 15	24
1386	BAC array CGH in patients with Velocardiofacial syndrome-like features reveals genomic aberrations on chromosome region 1q21.1. <b>2009</b> , 10, 144	27
1385	Identification of subtelomeric genomic imbalances and breakpoint mapping with quantitative PCR in 296 individuals with congenital defects and/or mental retardation. <b>2009</b> , 2, 10	5
1384	Human genetics and resistance to parasitic infection. <b>2009</b> , 31, 221-4	3
1383	Copy number variation in the human genome and its implication in autoimmunity. <b>2009</b> , 156, 12-6	71
1382	Functional and clinical consequences of Fc receptor polymorphic and copy number variants. <b>2009</b> , 157, 244-54	119
1381	Intravenous immunoglobulins--understanding properties and mechanisms. <b>2009</b> , 158 Suppl 1, 2-13	163
1380	The future of genetics in psychology and psychiatry: microarrays, genome-wide association, and non-coding RNA. <b>2009</b> , 50, 63-71	46
1379	The effects of genetic polymorphisms in the organic cation transporters OCT1, OCT2, and OCT3 on the renal clearance of metformin. <b>2009</b> , 86, 299-306	254
1378	Transmitted duplication of 8p23.1-8p23.2 associated with speech delay, autism and learning difficulties. <b>2009</b> , 17, 37-43	50
1377	Characterization of deletions at 9p affecting the candidate regions for sex reversal and deletion 9p syndrome by MLPA. <b>2009</b> , 17, 1439-47	68
1376	A novel microdeletion syndrome involving 5q14.3-q15: clinical and molecular cytogenetic characterization of three patients. <b>2009</b> , 17, 1592-9	81
1375	Copy number variation in the CCL4L gene is associated with susceptibility to acute rejection in lung transplantation. <b>2009</b> , 10, 254-9	20

1374	Reproducible association with type 1 diabetes in the extended class I region of the major histocompatibility complex. <b>2009</b> , 10, 323-33	39
1373	Eczema genetics: current state of knowledge and future goals. <b>2009</b> , 129, 543-52	114
1372	Accurate detection of uniparental disomy and microdeletions by SNP array analysis in myelodysplastic syndromes with normal cytogenetics. <b>2009</b> , 23, 1605-13	77
1371	Let's celebrate human genetic diversity. <i>Nature</i> , <b>2009</b> , 461, 726-8	50.4 14
1370	Human genetics: Sharp focus on the variable genome. <i>Nature</i> , <b>2009</b> , 461, 735-6	50.4 3
1369	Deletion of the late cornified envelope LCE3B and LCE3C genes as a susceptibility factor for psoriasis. <b>2009</b> , 41, 211-5	405
1368	Segmental copy number variation shapes tissue transcriptomes. <b>2009</b> , 41, 424-9	245
1367	The impact of copy number variation on local gene expression in mouse hematopoietic stem and progenitor cells. <b>2009</b> , 41, 430-7	103
1366	Acquired mutations in TET2 are common in myelodysplastic syndromes. <b>2009</b> , 41, 838-42	598
1365	The DNA replication FoSTeS/MMBIR mechanism can generate genomic, genic and exonic complex rearrangements in humans. <b>2009</b> , 41, 849-53	333
1364	De novo copy number variants identify new genes and loci in isolated sporadic tetralogy of Fallot. <b>2009</b> , 41, 931-5	325
1363	Personalized copy number and segmental duplication maps using next-generation sequencing. <b>2009</b> , 41, 1061-7	543
1362	CCL3L1 and HIV/AIDS susceptibility. <b>2009</b> , 15, 1110-2	60
1361	CCL3L1 and HIV/AIDS susceptibility. <b>2009</b> , 15, 1112-5	57
1360	Experimental aspects of copy number variant assays at CCL3L1. <b>2009</b> , 15, 1115-7	63
1359	Reply to: "Experimental aspects of copy number variant assays at CCL3L1". <b>2009</b> , 15, 1117-20	22
1358	BreakDancer: an algorithm for high-resolution mapping of genomic structural variation. <b>2009</b> , 6, 677-81	1062
1357	Computational methods for discovering structural variation with next-generation sequencing. <b>2009</b> , 6, S13-20	421

1356	Array painting: a protocol for the rapid analysis of aberrant chromosomes using DNA microarrays. <b>2009</b> , 4, 1722-36	22
1355	Plasma lipoproteins: genetic influences and clinical implications. <b>2009</b> , 10, 109-21	304
1354	Human genetic variation and its contribution to complex traits. <b>2009</b> , 10, 241-51	778
1353	Mechanisms of change in gene copy number. <b>2009</b> , 10, 551-64	845
1352	Genetics of human gene expression: mapping DNA variants that influence gene expression. <b>2009</b> , 10, 595-604	185
1351	Computational analysis of human genome polymorphism. <b>2009</b> , 43, 260-268	4
1350	Extended genetic analysis of BTNL2 in sarcoidosis. <b>2009</b> , 73, 59-61	9
1349	Nuclear and mitochondrial genome defects in autisms. <b>2009</b> , 1151, 102-32	50
1348	Clinical utility of array CGH for the detection of chromosomal imbalances associated with mental retardation and multiple congenital anomalies. <b>2009</b> , 1151, 157-66	96
1347	Genes, cognition, and communication: insights from neurodevelopmental disorders. <b>2009</b> , 1156, 1-18	76
1346	Duplication of MER115 on chromosome 4 in patients with primary biliary cirrhosis. <b>2009</b> , 29, 375-83	7
1345	Genetic recombination as a major cause of mutagenesis in the human globin gene clusters. <b>2009</b> , 42, 1839-50	16
1344	Sample degradation leads to false-positive copy number variation calls in multiplex real-time polymerase chain reaction assays. <b>2009</b> , 386, 288-90	21
1343	Determination of CYP2D6 gene copy number by multiplex polymerase chain reaction analysis. <b>2009</b> , 389, 74-6	12
1342	Controlling oligonucleotide surface density in light-directed DNA array fabrication. <b>2009</b> , 25, 6570-5	19
1341	Genetic susceptibility to gastrointestinal cancer: minireview of the genomewide studies. <b>2009</b> , 16, 1783-8	1
1340	Identification of chromosome 7 inversion breakpoints in an autistic family narrows candidate region for autism susceptibility. <b>2009</b> , 2, 258-66	8
1339	High-throughput methods for SNP genotyping. <b>2009</b> , 578, 245-54	46

1338	Population analysis of large copy number variants and hotspots of human genetic disease. <b>2009</b> , 84, 148-61	454
1337	Replication stress induces genome-wide copy number changes in human cells that resemble polymorphic and pathogenic variants. <b>2009</b> , 84, 339-50	114
1336	DECIPHER: Database of Chromosomal Imbalance and Phenotype in Humans Using Ensembl Resources. <b>2009</b> , 84, 524-33	1126
1335	Hip geometry variation is associated with bone mineralization pathway gene variants: The Framingham Study. <b>2010</b> , 25, 1564-71	19
1334	Combinatorial algorithms for structural variation detection in high-throughput sequenced genomes. <b>2009</b> , 19, 1270-8	230
1333	(Epi)genomics and neurodevelopment in schizophrenia: monozygotic twins discordant for schizophrenia augment the search for disease-related (epi)genomic alterations. <b>2009</b> , 52, 8-19	20
1332	MEIS1 intronic risk haplotype associated with restless legs syndrome affects its mRNA and protein expression levels. <b>2009</b> , 18, 1065-74	73
1331	A prominent role for segmental duplications in modeling eukaryotic genomes. <b>2009</b> , 332, 254-66	24
1330	Array-CGH detection of a de novo 0.8Mb deletion in 19q13.32 associated with mental retardation, cardiac malformation, cleft lip and palate, hearing loss and multiple dysmorphic features. <b>2009</b> , 52, 62-6	12
1329	A new large deletion in the DFNB1 locus causes nonsyndromic hearing loss. <b>2009</b> , 52, 195-200	38
1328	Functional phenotyping and genotyping of circulating tumor cells from patients with castration resistant prostate cancer. <b>2009</b> , 277, 164-73	79
1327	A fast modified protocol for random-access ultra-high density whole-genome scan: a tool for personalized genomic medicine, positional mapping, and cytogenetic analysis. <b>2009</b> , 406, 31-5	5
1326	Analysis of eighteen deletion breakpoints in the parkin gene. <b>2009</b> , 389, 181-6	12
1325	The emerging role of synaptic cell-adhesion pathways in the pathogenesis of autism spectrum disorders. <b>2009</b> , 32, 402-12	215
1324	Recent insights into the molecular genetics of dementia. <b>2009</b> , 32, 451-61	47
1323	Genetic association analysis of copy-number variation (CNV) in human disease pathogenesis. <b>2009</b> , 93, 22-6	145
1322	Multiplex Parologue Ratio Tests for accurate measurement of multiallelic CNVs. <b>2009</b> , 93, 98-103	37
1321	Optimizing comparative genomic hybridization probes for genotyping and SNP detection in Plasmodium falciparum. <b>2009</b> , 93, 543-50	31

1320	A genome-wide survey of copy number variations in Han Chinese residing in Taiwan. <b>2009</b> , 94, 241-6	22
1319	Copy number variants (CNVs) in primate species using array-based comparative genomic hybridization. <b>2009</b> , 49, 18-25	18
1318	Chromosomal microarray interpretation: what is a child neurologist to do?. <b>2009</b> , 41, 391-8	19
1317	The genetics of human autoimmune disease. <b>2009</b> , 33, 290-9	68
1316	Phenotypic suppression of the Drosophila mitochondrial disease-like mutant tko(25t) by duplication of the mutant gene in its natural chromosomal context. <b>2009</b> , 9, 353-63	12
1315	Great genotypic and phenotypic diversities associated with copy-number variations of complement C4 and RP-C4-CYP21-TNX (RCCX) modules: a comparison of Asian-Indian and European American populations. <b>2009</b> , 46, 1289-303	40
1314	Development of single nucleotide polymorphism markers for Atlantic cod ( <i>Gadus morhua</i> ) using expressed sequences. <b>2009</b> , 296, 7-14	23
1313	Copy number variation in human health, disease, and evolution. <b>2009</b> , 10, 451-81	804
1312	Challenges for CNV interpretation in clinical molecular karyotyping: lessons learned from a 1001 sample experience. <b>2009</b> , 52, 398-403	84
1311	Cytokine Polymorphisms and Immunosenescence. <b>2009</b> , 631-658	
1310	The future (or lack of future) of personalized prescription in psychiatry. <b>2009</b> , 59, 81-9	68
1309	The dystrobrevin binding protein 1 (DTNBP1) gene is associated with schizophrenia in the Irish Case Control Study of Schizophrenia (ICCS) sample. <b>2009</b> , 115, 245-53	30
1308	Large-scale genomic analysis of ovarian carcinomas. <b>2009</b> , 3, 157-64	29
1307	High-Throughput Single Nucleotide Polymorphisms Genotyping Technologies. <b>2009</b> ,	0
1306	The miR-106b-25 polycistron, activated by genomic amplification, functions as an oncogene by suppressing p21 and Bim. <b>2009</b> , 136, 1689-700	233
1305	Genome-wide analysis of pancreatic cancer using microarray-based techniques. <b>2009</b> , 9, 13-24	45
1304	Array comparative genomic hybridization in prenatal diagnosis: another experience. <b>2009</b> , 25, 277-84	63
1303	Low-affinity Fcγ receptors, autoimmunity and infection. <b>2009</b> , 11, e24	55

1302	Ovarian aging: mechanisms and clinical consequences. <b>2009</b> , 30, 465-93	629
1301	The human Major Histocompatibility Complex as a paradigm in genomics research. <b>2009</b> , 8, 379-94	73
1300	The genetic architecture of Down syndrome phenotypes revealed by high-resolution analysis of human segmental trisomies. <b>2009</b> , 106, 12031-6	280
1299	Contribution of Fcγ receptor IIIA gene 158V/F polymorphism and copy number variation to the risk of ACPA-positive rheumatoid arthritis. <b>2009</b> , 68, 1775-80	42
1298	Single Nucleotide Polymorphisms. <b>2009</b> ,	23
1297	The neurobiology of individual differences in complex behavioral traits. <b>2009</b> , 32, 225-47	213
1296	Molecular, Clinical and Environmental Toxicology. <b>2009</b> ,	10
1295	Predictive genetic testing for coronary artery disease. <b>2009</b> , 46, 343-60	11
1294	Microarray Analysis of the Physical Genome. <b>2009</b> ,	2
1293	Key concepts in human genetics: understanding the complex phenotype. <b>2009</b> , 54, 1-10	4
1292	Nature versus nurture in determining athletic ability. <b>2009</b> , 54, 11-27	6
1291	Fluorescence In Situ Hybridization (FISH) [Application Guide]. <b>2009</b> ,	25
1290	Array CGH: Opening New Horizons. <b>2009</b> , 421-437	1
1289	Tandem repeats modify the structure of human genes hosted in segmental duplications. <b>2009</b> , 10, R137	15
1288	High resolution discovery and confirmation of copy number variants in 90 Yoruba Nigerians. <b>2009</b> , 10, R125	49
1287	Deep short-read sequencing of chromosome 17 from the mouse strains A/J and CAST/Ei identifies significant germline variation and candidate genes that regulate liver triglyceride levels. <b>2009</b> , 10, R112	32
1286	Use of high-density tiling microarrays to identify mutations globally and elucidate mechanisms of drug resistance in Plasmodium falciparum. <b>2009</b> , 10, R21	114
1285	PEMer: a computational framework with simulation-based error models for inferring genomic structural variants from massive paired-end sequencing data. <b>2009</b> , 10, R23	201

1284	Parasite-host interaction in malaria: genetic clues and copy number variation. <b>2009</b> , 1, 82	4
1283	Copy number variations and cancer. <b>2009</b> , 1, 62	219
1282	Genomic disorders ten years on. <b>2009</b> , 1, 42	112
1281	A kernel-based integration of genome-wide data for clinical decision support. <b>2009</b> , 1, 39	54
1280	Genetics of rheumatic disease. <b>2009</b> , 11, 248	18
1279	Triple X syndrome in a patient with partial lipodystrophy discovered using a high-density oligonucleotide microarray: a case report. <b>2009</b> , 3, 8867	2
1278	The HLA genomic loci map: expression, interaction, diversity and disease. <b>2009</b> , 54, 15-39	441
1277	Cytogenetics in Reproduction. <b>2009</b> , 777-799	
1276	Introduction to microarray technology. <b>2009</b> , 529, 1-22	27
1275	Textbook of Personalized Medicine. <b>2009</b> ,	55
1274	Constructing genomic maps of positive selection in humans: where do we go from here?. <b>2009</b> , 19, 711-22	318
1273	Genetics of chronic obstructive pulmonary disease: a succinct review, future avenues and prospective clinical applications. <b>2009</b> , 10, 655-67	17
1272	Pharmacogenetics: from discovery to patient care. <b>2009</b> , 66, 625-37	49
1271	Copy number alterations that predict metastatic capability of human breast cancer. <b>2009</b> , 69, 3795-801	68
1270	Sequence and structural variation in a human genome uncovered by short-read, massively parallel ligation sequencing using two-base encoding. <b>2009</b> , 19, 1527-41	401
1269	Parent's attitudes towards full-scale prenatal testing for genetic disorders. <b>2009</b> , 30, 42-7	3
1268	Copy Number Variants: Distribution in Patients with Coronary Atherosclerosis. <b>2009</b> , 23, 1095-1100	2
1267	Molecular copy-number counting: potential of single-molecule diagnostics. <b>2009</b> , 9, 309-12	5



1266	Periventricular heterotopia, mental retardation, and epilepsy associated with 5q14.3-q15 deletion. <b>2009</b> , 72, 784-92	98
1265	Molecular markers of breast axillary lymph node metastasis. <b>2009</b> , 9, 441-54	15
1264	Mobile elements create structural variation: analysis of a complete human genome. <b>2009</b> , 19, 1516-26	220
1263	A Computational Approach to Detect CNVs Using High-throughput Sequencing. <b>2009</b> ,	
1262	Pharmacogenetics in heart failure: promises and challenges. <b>2009</b> , 10, 1713-25	8
1261	Next-generation sequencing: a transformative tool for vaccinology. <b>2009</b> , 8, 963-7	19
1260	Methodological issues in molecular genetic studies of mental disorders. <b>2009</b> , 5, 49-69	11
1259	Genetic Variants in Male Infertility. <b>2009</b> , 113-127	
1258	Genomic copy number variation, human health, and disease. <b>2009</b> , 374, 340-50	143
1257	Genotyping technologies for genetic research. <b>2009</b> , 10, 117-33	164
1256	Identification of genomic alterations associated with the aggressiveness of pancreatic cancer using an ultra-high-resolution CGH array. <b>2009</b> , 9, 267-72	7
1255	Genetics of type 1 diabetes and autoimmune thyroid disease. <b>2009</b> , 38, 289-301, vii-viii	17
1254	Detection of chromosome aneuploidies in chorionic villus samples by multiplex ligation-dependent probe amplification. <b>2009</b> , 11, 17-24	18
1253	New insights into nutrition and cognitive neuroscience. <b>2009</b> , 68, 408-15	62
1252	Comparing CNV detection methods for SNP arrays. <b>2009</b> , 8, 353-66	150
1251	Copy number variations associated with idiopathic autism identified by whole-genome microarray-based comparative genomic hybridization. <b>2009</b> , 19, 177-85	32
1250	The evolution of human segmental duplications and the core duplicon hypothesis. <b>2009</b> , 74, 355-62	48
1249	Structural chromosomal variations in neurological diseases. <b>2009</b> , 15, 245-53	8

1248	Molecular Biological Analyses and Molecular Pathology in Clinical Chemistry. <b>2009</b> ,	
1247	Using GWAS Data to Identify Copy Number Variants Contributing to Common Complex Diseases. <b>2009</b> , 24,	9
1246	Genome-wide Association Studies. <b>2009</b> ,	0
1245	[Methods for the analysis of large gene deletions and their application in some hereditary diseases]. <b>2009</b> , 150, 2258-64	1
1244	Personalized medicine: A transformative approach is needed. <b>2009</b> , 180, 911-913	4
1243	Genome-wide profiling of follicular lymphoma by array comparative genomic hybridization reveals prognostically significant DNA copy number imbalances. <b>2009</b> , 113, 137-48	99
1242	Uniparental disomies, homozygous deletions, amplifications, and target genes in mantle cell lymphoma revealed by integrative high-resolution whole-genome profiling. <b>2009</b> , 113, 3059-69	147
1241	HapMap scanning of novel human minor histocompatibility antigens. <b>2009</b> , 113, 5041-8	46
1240	Genes and osteoporosis: time for a change in strategy. <b>2009</b> , 4, 221-233	2
1239	Association tests and software for copy number variant data. <b>2009</b> , 3, 191-4	6
1238	Mouse models of genomic syndromes as tools for understanding the basis of complex traits: an example with the smith-magenis and the potocki-lupski syndromes. <b>2009</b> , 10, 259-68	12
1237	Use of cell lines in the investigation of pharmacogenetic loci. <b>2009</b> , 15, 3782-95	31
1236	Grouping preprocess for haplotype inference from SNP and CNV data. <b>2009</b> , 197, 012009	1
1235	The human genome puzzle - the role of copy number variation in somatic mosaicism. <b>2010</b> , 11, 426-31	40
1234	Finding Recurrent Copy Number Alteration Regions: A Review of Methods. <b>2010</b> , 5, 1-17	31
1233	Rare copy number variants: a point of rarity in genetic risk for bipolar disorder and schizophrenia. <b>2010</b> , 67, 318-27	154
1232	Genetics of congenital heart disease. <b>2010</b> , 6, 91-7	120
1231	Controlled somatic and germline copy number variation in the mouse model. <b>2010</b> , 11, 470-80	2

1230	Absence of AVPR2 copy number variation in eunatremic and dysnatremic subjects in non-Hispanic Caucasian populations. <b>2010</b> , 40, 121-7	2
1229	Retinoic Acid Induced 1, RAI1: A Dosage Sensitive Gene Related to Neurobehavioral Alterations Including Autistic Behavior. <b>2010</b> , 11, 607-17	34
1228	TFG, a target of chromosome translocations in lymphoma and soft tissue tumors, fuses to GPR128 in healthy individuals. <b>2010</b> , 95, 20-6	45
1227	Nature and Nurture: Genetic Influences and Gene-Environment Interactions in Depression. <b>2010</b> , 6, 82-90	1
1226	Molecular Biomarkers in Schizophrenia – Implications for Clinical Practice. <b>2010</b> , 6, 114-121	
1225	Evolution of detoxifying systems: the role of environment and population history in shaping genetic diversity at human CYP2D6 locus. <b>2010</b> , 20, 485-99	23
1224	"Genes to society"--the logic and process of the new curriculum for the Johns Hopkins University School of Medicine. <b>2010</b> , 85, 498-506	57
1223	Recent advances in the genetics of rheumatoid arthritis. <b>2010</b> , 22, 109-18	81
1222	Copy number variations and cancer susceptibility. <b>2010</b> , 22, 55-63	58
1221	Male with mosaicism for supernumerary ring X chromosome: analysis of phenotype and characterization of genotype using array comparative genome hybridization. <b>2010</b> , 21, 1369-75	7
1220	Genome dynamics are influenced by food source in <i>Allogromia laticollaris</i> strain CSH (Foraminifera). <b>2010</b> , 2, 678-85	15
1219	DNA sequence variants and the practice of medicine. <b>2010</b> , 25, 182-5	2
1218	The personal genome and the practice of cardiovascular medicine. <b>2010</b> , 6, 13-20	2
1217	Genome-wide scan of copy number variation in late-onset Alzheimer's disease. <b>2010</b> , 19, 69-77	98
1216	Genome-wide scanning reveals complex etiology of oculo-auriculo-vertebral spectrum. <b>2010</b> , 222, 311-8	19
1215	High-density screening reveals a different spectrum of genomic aberrations in chronic lymphocytic leukemia patients with 'stereotyped' IGHV3-21 and IGHV4-34 B-cell receptors. <b>2010</b> , 95, 1519-25	40
1214	'Mendelian CNVs'; Causing Mental Retardation and Developmental Disorders. <b>2010</b> , 114-125	
1213	Clinical omics analysis of colorectal cancer incorporating copy number aberrations and gene expression data. <b>2010</b> , 9, 147-61	30

1212 Change Point Methods in Genetics. **2010**, 1

1211 Copy Number Variations. **2010**, 21-33

1210 SNP array analysis in hematologic malignancies: avoiding false discoveries. **2010**, 115, 4157-61 82

1209 On a novel coalescent model for genome-wide evolution of copy number variations. **2010**, 4, 300-15 4

1208 Statistical methods for detecting natural selection from genomic data. **2010**, 85, 359-76 21

1207 Quantitative Genomics of Female Reproduction. **2010**, 23-51 1

1206 Genomewide Association Studies: History, Rationale, and Prospects for Psychiatric Disorders. **2010**, 8, 417-434 1

1205 Increased genomic copy number of DEFA1/DEFA3 is associated with susceptibility to severe sepsis in Chinese Han population. **2010**, 112, 1428-34 37

1204 microRNA and cancer. **2010**, 12, 309-17 120

1203 Diversity of human copy number variation and multicopy genes. **2010**, 330, 641-6 491

1202 Consensus statement: chromosomal microarray is a first-tier clinical diagnostic test for individuals with developmental disabilities or congenital anomalies. **2010**, 86, 749-64 1831

1201 A definitive haplotype map as determined by genotyping duplicated haploid genomes finds a predominant haplotype preference at copy-number variation events. **2010**, 86, 918-28 6

1200 Mechanisms of genomic instabilities underlying two common fragile-site-associated loci, PARK2 and DMD, in germ cell and cancer cell lines. **2010**, 87, 75-89 71

1199 Fine-scale survey of X chromosome copy number variants and indels underlying intellectual disability. **2010**, 87, 173-88 93

1198 Identification of copy number variation hotspots in human populations. **2010**, 87, 494-504 36

1197 Population differences in the rate of proliferation of international HapMap cell lines. **2010**, 87, 829-33 18

1196 Recurrent distal 7q11.23 deletion including HIP1 and YWHAG identified in patients with intellectual disabilities, epilepsy, and neurobehavioral problems. **2010**, 87, 857-65 48

1195 Carbohydrate metabolic pathway genes associated with quantitative trait loci (QTL) for obesity and type 2 diabetes: identification by data mining. **2010**, 5, 942-9 4

1194	Structural variation in the human genome and its role in disease. <b>2010</b> , 61, 437-55	827
1193	The single nucleotide polymorphisms gene but not the copy number variation of Fcgr3B is associated with lupus nephritis in Chinese people. <b>2010</b> , 19, 662-4	7
1192	Genome-wide association studies and beyond. <b>2010</b> , 31, 9-20 4 p following 20	96
1191	Analysis of copy number variations of BS69 in multiple types of hematological malignancies. <b>2010</b> , 89, 959-64	8
1190	Allelic genome structural variations in maize detected by array comparative genome hybridization. <b>2010</b> , 120, 355-67	77
1189	CBF gene copy number variation at Frost Resistance-2 is associated with levels of freezing tolerance in temperate-climate cereals. <b>2010</b> , 121, 21-35	121
1188	Genetic origins of pediatric heart disease. <b>2010</b> , 31, 422-9	37
1187	Genome-wide copy number variation association study suggested VPS13B gene for osteoporosis in Caucasians. <b>2010</b> , 21, 579-87	23
1186	New technologies in the genetic approach to sudden cardiac death in the young. <b>2010</b> , 203, 15-24	17
1185	Nutritional genomics era: opportunities toward a genome-tailored nutritional regimen. <b>2010</b> , 21, 457-67	25
1184	Copy number variations are a rare cause of non-CMT1A Charcot-Marie-Tooth disease. <b>2010</b> , 257, 735-41	21
1183	Genetics of osteoporosis: accelerating pace in gene identification and validation. <b>2010</b> , 127, 249-85	77
1182	Copy number variants at Williams-Beuren syndrome 7q11.23 region. <b>2010</b> , 128, 3-26	112
1181	Genetic determinants of autism in individuals with deletions of 18q. <b>2010</b> , 128, 155-64	18
1180	Breakpoint determination of 15 large deletions in Peutz-Jeghers subjects. <b>2010</b> , 128, 373-82	25
1179	Rare variation at the TNFAIP3 locus and susceptibility to rheumatoid arthritis. <b>2010</b> , 128, 627-33	27
1178	HNF1B alterations associated with congenital anomalies of the kidney and urinary tract. <b>2010</b> , 25, 1073-9	62
1177	Connecting the Human Variome Project to nutrigenomics. <b>2010</b> , 5, 275-283	4

1176	The genome-wide association study--a new era for common polygenic disorders. <b>2010</b> , 3, 173-82	22
1175	The presence of the UGT2B17 gene is associated with lung cancer in male Chinese Han smokers. <b>2010</b> , 32, 13-17	2
1174	The genetic basis of non-syndromic intellectual disability: a review. <b>2010</b> , 2, 182-209	162
1173	Copy number variations are not modifiers of phenotypic expression in a pair of identical twins carrying a BRCA1 mutation. <b>2010</b> , 123, 901-5	9
1172	Integrating the multiple dimensions of genomic and epigenomic landscapes of cancer. <b>2010</b> , 29, 73-93	39
1171	Genome-wide association studies in economics and entrepreneurship research: promises and limitations. <b>2010</b> , 35, 1-18	34
1170	Systems biology and heart failure: concepts, methods, and potential research applications. <b>2010</b> , 15, 371-98	16
1169	Epilepsy: insights into causes and treatment dilemmas. <b>2010</b> , 9, 9-11	
1168	Rapid Sequence Homology Assessment by Subsampling the Genome Space Using Difference Sets. <b>2010</b> , 56, 756-770	1
1167	Comprehensive copy number variant (CNV) analysis of neuronal pathways genes in psychiatric disorders identifies rare variants within patients. <b>2010</b> , 44, 971-8	60
1166	Evolutionary biology looks at behavior genetics. <b>2010</b> , 49, 289-295	11
1165	A mobile threat to genome stability: The impact of non-LTR retrotransposons upon the human genome. <b>2010</b> , 20, 211-21	145
1164	Single nucleotide polymorphisms of DNA repair genes as predictors of radioresponse. <b>2010</b> , 20, 232-40	63
1163	Mutational bias shaping fly copy number variation: implications for genome evolution. <b>2010</b> , 26, 243-7	20
1162	Human genome diversity: frequently asked questions. <b>2010</b> , 26, 285-95	75
1161	Reconstructing CNV genotypes using segregation analysis: combining pedigree information with CNV assay. <b>2010</b> , 42, 34	3
1160	Identification of recurrent regions of Copy-Number Variants across multiple individuals. <b>2010</b> , 11, 147	11
1159	Conditional random pattern model for copy number aberration detection. <b>2010</b> , 11, 200	2

1158	Partitioning of copy-number genotypes in pedigrees. <b>2010</b> , 11, 226	2
1157	CNstream: a method for the identification and genotyping of copy number polymorphisms using Illumina microarrays. <b>2010</b> , 11, 264	13
1156	A classification model for distinguishing copy number variants from cancer-related alterations. <b>2010</b> , 11, 297	13
1155	CONAN: copy number variation analysis software for genome-wide association studies. <b>2010</b> , 11, 318	15
1154	R-Gada: a fast and flexible pipeline for copy number analysis in association studies. <b>2010</b> , 11, 380	44
1153	Detection of copy number variation from array intensity and sequencing read depth using a stepwise Bayesian model. <b>2010</b> , 11, 539	5
1152	CNV Workshop: an integrated platform for high-throughput copy number variation discovery and clinical diagnostics. <b>2010</b> , 11, 74	45
1151	Identification of copy number variations and common deletion polymorphisms in cattle. <b>2010</b> , 11, 232	113
1150	Copy number variation in the bovine genome. <b>2010</b> , 11, 284	121
1149	A critical assessment of cross-species detection of gene duplicates using comparative genomic hybridization. <b>2010</b> , 11, 304	7
1148	Genetic structure of the Spanish population. <b>2010</b> , 11, 326	43
1147	An initial map of chromosomal segmental copy number variations in the chicken. <b>2010</b> , 11, 351	81
1146	Copy number variation and cytidine analogue cytotoxicity: a genome-wide association approach. <b>2010</b> , 11, 357	16
1145	Deletions of immunoglobulin heavy chain and T cell receptor gene regions are uniquely associated with lymphoid blast transformation of chronic myeloid leukemia. <b>2010</b> , 11, 41	36
1144	Copy number variation in the porcine genome inferred from a 60 k SNP BeadChip. <b>2010</b> , 11, 593	86
1143	Copy number variation in the genomes of twelve natural isolates of <i>Caenorhabditis elegans</i> . <b>2010</b> , 11, 62	57
1142	An initial comparative map of copy number variations in the goat ( <i>Capra hircus</i> ) genome. <b>2010</b> , 11, 639	96
1141	Analysis of copy loss and gain variations in Holstein cattle autosomes using BeadChip SNPs. <b>2010</b> , 11, 673	66

1140	An integrative multi-dimensional genetic and epigenetic strategy to identify aberrant genes and pathways in cancer. <b>2010</b> , 4, 67	48
1139	A simple way to evaluate self-designed probes for tumor specific Multiplex Ligation-dependent Probe Amplification (MLPA). <b>2010</b> , 3, 179	2
1138	Bayesian estimation of genomic copy number with single nucleotide polymorphism genotyping arrays. <b>2010</b> , 3, 350	
1137	The evolution of human genetic and phenotypic variation in Africa. <b>2010</b> , 20, R166-73	139
1136	Ciliary trafficking: CEP290 guards a gated community. <b>2010</b> , 20, R928-31	31
1135	Reproductive strategies: how big is your love?. <b>2010</b> , 20, R925-8	4
1134	Comprehensive genetic analyses of PLP1 in patients with Pelizaeus-Merzbacher disease applied by array-CGH and fiber-FISH analyses identified new mutations and variable sizes of duplications. <b>2010</b> , 32, 171-9	26
1133	Micro-RNAs and copy number changes: new levels of gene regulation in acute myeloid leukemia. <b>2010</b> , 184, 21-5	9
1132	Subtelomeric rearrangements and copy number variations in people with intellectual disabilities. <b>2010</b> , 54, 938-42	8
1131	Disseminated peritoneal leiomyomatosis after laparoscopic supracervical hysterectomy with characteristic molecular cytogenetic findings of uterine leiomyoma. <b>2010</b> , 49, 1152-60	57
1130	On the genome-wide analysis of copy number variants in family-based designs: methods for combining family-based and population-based information for testing dichotomous or quantitative traits, or completely ascertained samples. <b>2010</b> , 34, 582-90	6
1129	Complete ascertainment of intragenic copy number mutations (CNMs) in the CFTR gene and its implications for CNM formation at other autosomal loci. <b>2010</b> , 31, 421-8	29
1128	Assessment of complement C4 gene copy number using the paralog ratio test. <b>2010</b> , 31, 866-74	20
1127	Genes, mutations, and human inherited disease at the dawn of the age of personalized genomics. <b>2010</b> , 31, 631-55	138
1126	Genomic copy number variations in three Southeast Asian populations. <b>2010</b> , 31, 851-7	20
1125	Missense mutations in the AFG3L2 proteolytic domain account for ~1.5% of European autosomal dominant cerebellar ataxias. <b>2010</b> , 31, 1117-24	69
1124	Detection of clinically relevant exonic copy-number changes by array CGH. <b>2010</b> , 31, 1326-42	195
1123	Evolutionary Genomics Leads the Way. <b>2010</b> , 1-16	



1122	Common recurrent microduplication syndromes: diagnosis and management in clinical practice. <b>2010</b> , 152A, 1066-78	25
1121	De novo 12;17 translocation upstream of SOX9 resulting in 46,XX testicular disorder of sex development. <b>2010</b> , 152A, 422-6	29
1120	Delineation of a 1.65 Mb critical region for hemihyperplasia and digital anomalies on Xq25. <b>2010</b> , 152A, 453-8	9
1119	A homozygous deletion of 8q24.3 including the NIBP gene associated with severe developmental delay, dysgenesis of the corpus callosum, and dysmorphic facial features. <b>2010</b> , 152A, 1268-72	18
1118	Genomic alterations in biliary atresia suggest region of potential disease susceptibility in 2q37.3. <b>2010</b> , 152A, 886-95	48
1117	EGR3 as a potential susceptibility gene for schizophrenia in Korea. <b>2010</b> , 153B, 1355-60	27
1116	Nonadaptive processes in primate and human evolution. <b>2010</b> , 143 Suppl 51, 13-45	14
1115	Protection of human genomic DNA from mechanical stress by reversible folding transition. <b>2010</b> , 11, 340-3	14
1114	Multiple mechanisms induce ectopic expression of LYL1 in subsets of T-ALL cell lines. <b>2010</b> , 34, 521-8	20
1113	Genetic variation, Fcγ receptors, KIRs and infection: the evolution of autoimmunity. <b>2010</b> , 22, 715-22	20
1112	Comparing the retention mechanisms of tandem duplicates and retrogenes in human and mouse genomes. <b>2010</b> , 42, 24	6
1111	Genetic copy number variants in sib pairs both affected with schizophrenia. <b>2010</b> , 17, 2	23
1110	A copy number variation in human NCF1 and its pseudogenes. <b>2010</b> , 11, 13	18
1109	Little ROCK is a ROCK1 pseudogene expressed in human smooth muscle cells. <b>2010</b> , 11, 22	2
1108	Functional and cellular characterization of human Retinoic Acid Induced 1 (RAI1) mutations associated with Smith-Magenis Syndrome. <b>2010</b> , 11, 63	26
1107	The use of array-CGH in a cohort of Greek children with developmental delay. <b>2010</b> , 3, 22	20
1106	Clinical application of whole-genome array CGH during prenatal diagnosis: Study of 25 selected pregnancies with abnormal ultrasound findings or apparently balanced structural aberrations. <b>2010</b> , 3, 24	37
1105	Candidate gene study of HOXB1 in autism spectrum disorder. <b>2010</b> , 1, 9	6

1104	Pharmacogenomics: a systems approach. <b>2010</b> , 2, 3-22	40
1103	Multiple correspondence discriminant analysis: an application to detect stratification in copy number variation. <b>2010</b> , 29, 3284-93	6
1102	Single-molecule genomics. <b>2010</b> , 220, 297-306	26
1101	A retrospective study by oligonucleotide array-CGH analysis in 50 fetuses with multiple malformations. <b>2010</b> , 30, 333-41	60
1100	The impact of human copy number variation on a new era of genetic testing. <b>2010</b> , 117, 391-8	41
1099	Genetic variations associated with psoriasis and psoriatic arthritis found by genome-wide association. <b>2010</b> , 23, 101-13	28
1098	Segmentation and estimation for SNP microarrays: a Bayesian multiple change-point approach. <b>2010</b> , 66, 675-83	4
1097	Cytokine regulation of immune responses to <i>Porphyromonas gingivalis</i> . <b>2010</b> , 54, 160-94	39
1096	A locus for an auditory processing deficit and language impairment in an extended pedigree maps to 12p13.31-q14.3. <b>2010</b> , 9, 545-61	21
1095	Correlation between genotype and supernumerary tooth formation in cleidocranial dysplasia. <b>2010</b> , 13, 197-202	32
1094	Barcode technology in yeast: application to pharmacogenomics. <b>2010</b> , 10, 1083-9	13
1093	Massive parallel sequencing in animal genetics: wherefroms and wheretos. <b>2010</b> , 41, 561-9	37
1092	Prognostic significance of genetic alterations detected by high-density single nucleotide polymorphism array in gastric cancer. <b>2010</b> , 101, 1261-9	21
1091	Copy number variations of the human histamine H4 receptor gene are associated with systemic lupus erythematosus. <b>2010</b> , 163, 935-40	29
1090	Deciphering the molecular basis of venous thromboembolism: where are we and where should we go?. <b>2010</b> , 148, 495-506	12
1089	Poor prognosis in familial acute myeloid leukaemia with combined biallelic CEBPA mutations and downstream events affecting the ATM, FLT3 and CDX2 genes. <b>2010</b> , 150, 382-5	13
1088	Comparison of familial and sporadic chronic lymphocytic leukaemia using high resolution array comparative genomic hybridization. <b>2010</b> , 151, 336-45	11
1087	Copy number variation in chemokine superfamily: the complex scene of CCL3L-CCL4L genes in health and disease. <b>2010</b> , 162, 41-52	34

1086	High-resolution genomic profiling of human papillomavirus-associated vulval neoplasia. <b>2010</b> , 102, 1044-51	6
1085	Arsenic-related DNA copy-number alterations in lung squamous cell carcinomas. <b>2010</b> , 103, 1277-83	35
1084	Recurrent copy number changes in mentally retarded children harbour genes involved in cellular localization and the glutamate receptor complex. <b>2010</b> , 18, 39-46	36
1083	High-resolution SNP arrays in mental retardation diagnostics: how much do we gain?. <b>2010</b> , 18, 178-85	44
1082	Identification of ANKRD11 and ZNF778 as candidate genes for autism and variable cognitive impairment in the novel 16q24.3 microdeletion syndrome. <b>2010</b> , 18, 429-35	83
1081	A new set of markers for human identification based on 32 polymorphic Alu insertions. <b>2010</b> , 18, 808-14	17
1080	Association of copy number variation in the FCGR3B gene with risk of autoimmune diseases. <b>2010</b> , 11, 155-60	66
1079	Identification of acquired copy number alterations and uniparental disomies in cytogenetically normal acute myeloid leukemia using high-resolution single-nucleotide polymorphism analysis. <b>2010</b> , 24, 438-49	104
1078	Association of common copy number variants at the glutathione S-transferase genes and rare novel genomic changes with schizophrenia. <b>2010</b> , 15, 1023-33	65
1077	Origins and functional impact of copy number variation in the human genome. <i>Nature</i> , <b>2010</b> , 464, 704-12	50.4 1467
1076	Genome-wide association study of CNVs in 16,000 cases of eight common diseases and 3,000 shared controls. <i>Nature</i> , <b>2010</b> , 464, 713-20	50.4 639
1075	Integrating common and rare genetic variation in diverse human populations. <i>Nature</i> , <b>2010</b> , 467, 52-8	50.4 2135
1074	Nucleotide-resolution analysis of structural variants using BreakSeq and a breakpoint library. <b>2010</b> , 28, 47-55	136
1073	High-resolution DNA analysis of human embryonic stem cell lines reveals culture-induced copy number changes and loss of heterozygosity. <b>2010</b> , 28, 371-7	223
1072	Quantitative comparison of genome-wide DNA methylation mapping technologies. <b>2010</b> , 28, 1106-14	486
1071	Discovery of common Asian copy number variants using integrated high-resolution array CGH and massively parallel DNA sequencing. <b>2010</b> , 42, 400-5	167
1070	Mutation spectrum revealed by breakpoint sequencing of human germline CNVs. <b>2010</b> , 42, 385-91	192
1069	Genome-wide association study identifies new HLA class II haplotypes strongly protective against narcolepsy. <b>2010</b> , 42, 786-9	145

1068	Copy number variation and human genome maps. <b>2010</b> , 42, 365-6	19
1067	Chipping away at the genetics of smoking behavior. <b>2010</b> , 42, 366-8	38
1066	Public data archives for genomic structural variation. <b>2010</b> , 42, 813-4	67
1065	Gene-environment interaction influences the reactivity of autoantibodies to citrullinated antigens in rheumatoid arthritis. <b>2010</b> , 42, 814-6; author reply 816	57
1064	Characterization of missing human genome sequences and copy-number polymorphic insertions. <b>2010</b> , 7, 365-71	114
1063	The evolution of gene duplications: classifying and distinguishing between models. <b>2010</b> , 11, 97-108	833
1062	Methodological challenges of genome-wide association analysis in Africa. <b>2010</b> , 11, 149-60	143
1061	Missing heritability and strategies for finding the underlying causes of complex disease. <b>2010</b> , 11, 446-50	1230
1060	Genome destabilization by homologous recombination in the germ line. <b>2010</b> , 11, 182-95	159
1059	FcgammaRIIB, FcgammaRIIIB, and systemic lupus erythematosus. <b>2010</b> , 1183, 69-88	71
1058	Impact of Mendelian inheritance in cardiovascular disease. <b>2010</b> , 1214, 122-37	10
1057	DISC1 duplication in two brothers with autism and mild mental retardation. <b>2010</b> , 77, 389-94	24
1056	Gene copy number variation and common human disease. <b>2010</b> , 77, 201-13	85
1055	Mandibulofacial dysostosis, microtia, and limb anomalies in a newborn: a new form of acrofacial dysostosis syndrome?. <b>2010</b> , 78, 570-4	11
1054	. <b>2010</b> ,	7
1053	Introduction to Conservation Genetics. 539-594	
1052	The MAOA Gene Predicts Credit Card Debt. <b>2010</b> ,	9
1051	Dosage sensitivity shapes the evolution of copy-number varied regions. <b>2010</b> , 5, e9474	76

1050	A male with unilateral microphthalmia reveals a role for TMX3 in eye development. <b>2010</b> , 5, e10565	27
1049	High-throughput high-resolution class I HLA genotyping in East Africa. <b>2010</b> , 5, e10751	8
1048	Genome-wide profiling of structural genomic variations in Korean HapMap individuals. <b>2010</b> , 5, e11417	6
1047	Association of CCR2-CCR5 haplotypes and CCL3L1 copy number with Kawasaki Disease, coronary artery lesions, and IVIG responses in Japanese children. <b>2010</b> , 5, e11458	19
1046	Novel association strategy with copy number variation for identifying new risk Loci of human diseases. <b>2010</b> , 5, e12185	19
1045	Elusive copy number variation in the mouse genome. <b>2010</b> , 5, e12839	22
1044	Genetic variation of the Fc gamma receptor 3B gene and association with rheumatoid arthritis. <b>2010</b> , 5, e13173	22
1043	New copy number variations in schizophrenia. <b>2010</b> , 5, e13422	72
1042	Analyses of copy number variation of GK rat reveal new putative type 2 diabetes susceptibility loci. <b>2010</b> , 5, e14077	10
1041	Does collocation inform the impact of collaboration?. <b>2010</b> , 5, e14279	68
1040	The effect of algorithms on copy number variant detection. <b>2010</b> , 5, e14456	27
1039	Microarray-based maps of copy-number variant regions in European and sub-Saharan populations. <b>2010</b> , 5, e15246	20
1038	Pharmacogenetics in breast cancer hormone therapy. <b>2010</b> , 4, 242-247	
1037	Design and generation of MLPA probe sets for combined copy number and small-mutation analysis of human genes: EGFR as an example. <b>2010</b> , 10, 2003-18	21
1036	Targeted interrogation of copy number variation using SCIMMkit. <b>2010</b> , 26, 120-2	6
1035	Waved aCGH: to smooth or not to smooth. <b>2010</b> , 38, e94	23
1034	Failure to confirm CNVs as of aetiological significance in twin pairs discordant for schizophrenia. <b>2010</b> , 13, 455-60	21
1033	Copy number variations in East-Asian population and their evolutionary and functional implications. <b>2010</b> , 19, 1001-8	73

1032	Independent and population-specific association of risk variants at the IRGM locus with Crohn's disease. <b>2010</b> , 19, 1828-39	79
1031	Copy number variation, chromosome rearrangement, and their association with recombination during avian evolution. <b>2010</b> , 20, 503-11	112
1030	Copy number variation of FCGR3A rather than FCGR3B and FCGR2B is associated with susceptibility to anti-GBM disease. <b>2010</b> , 22, 45-51	45
1029	Detecting structural variations in the human genome using next generation sequencing. <b>2010</b> , 9, 405-15	48
1028	PhosSNP for systematic analysis of genetic polymorphisms that influence protein phosphorylation. <b>2010</b> , 9, 623-34	65
1027	Reference-unbiased copy number variant analysis using CGH microarrays. <b>2010</b> , 38, e190	20
1026	Phenotype-genotype correlation in a familial IGF1R microdeletion case. <b>2010</b> , 47, 492-8	34
1025	Visualizing chromosome mosaicism and detecting ethnic outliers by the method of "rare" heterozygotes and homozygotes (RHH). <b>2010</b> , 19, 2539-53	1
1024	The effect of translocation-induced nuclear reorganization on gene expression. <b>2010</b> , 20, 554-64	78
1023	Founder population-specific HapMap panel increases power in GWA studies through improved imputation accuracy and CNV tagging. <b>2010</b> , 20, 1344-51	40
1022	Gene copy-number polymorphism in nature. <b>2010</b> , 277, 3213-21	106
1021	Genetic variation of genes involved in dihydrotestosterone metabolism and the risk of prostate cancer. <b>2010</b> , 19, 229-39	42
1020	Genome-wide SNP array analysis in patients with features of sotos syndrome. <b>2010</b> , 73, 265-74	19
1019	Whole genome survey of copy number variation in the spontaneously hypertensive rat: relationship to quantitative trait loci, gene expression, and blood pressure. <b>2010</b> , 55, 1231-8	18
1018	Unravelling the genetics of spermatogenic failure. <b>2010</b> , 139, 303-7	41
1017	The clinical context of copy number variation in the human genome. <b>2010</b> , 12, e8	134
1016	Detecting copy number variation with mated short reads. <b>2010</b> , 20, 1613-22	130
1015	Patrocles: a database of polymorphic miRNA-mediated gene regulation in vertebrates. <b>2010</b> , 38, D640-51	121

1014	CCL3L1 copy number is a strong genetic determinant of HIV seropositivity in Caucasian intravenous drug users. <b>2010</b> , 201, 730-9	25
1013	Population-genetic nature of copy number variations in the human genome. <b>2010</b> , 19, 761-73	34
1012	A bayesian analysis for identifying DNA copy number variations using a compound poisson process. <b>2010</b> , 2010, 268513	1
1011	Metabolic syndrome components in murine models. <b>2010</b> , 10, 25-40	18
1010	Performance of MLPA as a screening method for aneuploidy in uncultured amniocytes. <b>2010</b> , 11, 199-203	
1009	Genes associated with multiple sclerosis: 15 and counting. <b>2010</b> , 10, 857-61	18
1008	Coronary heart disease risk prediction in the era of genome-wide association studies: current status and what the future holds. <b>2010</b> , 121, 2235-48	49
1007	Copy number, linkage disequilibrium and disease association in the FCGR locus. <b>2010</b> , 19, 3282-94	103
1006	Inferring combined CNV/SNP haplotypes from genotype data. <b>2010</b> , 26, 1437-45	29
1005	Current Clinical and Pharmaceutical Applications of Microarrays: From Disease Biomarkers Discovery to Automated Diagnostics. <b>2010</b> , 15, 405-413	8
1004	A very fast and accurate method for calling aberrations in array-CGH data. <b>2010</b> , 11, 515-8	14
1003	Low dimensional chaos in the AT and GC skew profiles of DNA sequences. <b>2010</b> , 19, 090508	1
1002	Can the genetics of type 1 and type 2 diabetes shed light on the genetics of latent autoimmune diabetes in adults?. <b>2010</b> , 31, 183-93	46
1001	Loss-of-function variants in the genomes of healthy humans. <b>2010</b> , 19, R125-30	141
1000	Copy number variant detection in inbred strains from short read sequence data. <b>2010</b> , 26, 565-7	40
999	Application of OMICS technologies in occupational and environmental health research; current status and projections. <b>2010</b> , 67, 136-43	72
998	Classification of genome-wide copy number variations and their associated SNP and gene networks analysis. <b>2010</b> ,	
997	A double-layered mixture model for the joint analysis of DNA copy number and gene expression data. <b>2010</b> , 17, 121-37	15

996	Application of Nexus copy number software for CNV detection and analysis. <b>2010</b> , Chapter 4, Unit 4.14.1-28	18
995	Classification of pathogenic or benign status of CNVs detected by microarray analysis. <b>2010</b> , 10, 717-21	10
994	Association of higher DEFB4 genomic copy number with Crohn's disease. <b>2010</b> , 105, 354-9	75
993	Association of TLR7 copy number variation with susceptibility to childhood-onset systemic lupus erythematosus in Mexican population. <b>2010</b> , 69, 1861-5	80
992	GATA4 mutations in 357 unrelated patients with congenital heart malformation. <b>2010</b> , 14, 797-802	47
991	Duplication of the class I cytosolic small heat shock protein gene and potential functional divergence revealed by sequence variations flanking the {alpha}-crystallin domain in the genus <i>Rhododendron</i> (Ericaceae). <b>2010</b> , 105, 57-69	6
990	A survey of cancer cell lines reveals highly structured and hierarchical relationships within and between DNA and mRNA that may be the result of selection. <b>2010</b> , 14, 91-7	5
989	Dynamics and processes of copy number instability in human gamma-globin genes. <b>2010</b> , 107, 8304-9	15
988	The association between copy number variations in glutathione S-transferase M1 and T1 and age-related cataract in a Han Chinese population. <b>2010</b> , 51, 3924-8	24
987	The impact of genetic architecture on genome-wide evaluation methods. <b>2010</b> , 185, 1021-31	487
986	FACADE: a fast and sensitive algorithm for the segmentation and calling of high resolution array CGH data. <b>2010</b> , 38, e157	14
985	High-resolution human genome structure by single-molecule analysis. <b>2010</b> , 107, 10848-53	146
984	Disruption of ST5 is associated with mental retardation and multiple congenital anomalies. <b>2010</b> , 47, 91-8	9
983	Copy number variations in schizophrenia: critical review and new perspectives on concepts of genetics and disease. <b>2010</b> , 167, 899-914	156
982	Simultaneous detection of trisomies 13, 18, and 21 with multiplex ligation-dependent probe amplification-based real-time PCR. <b>2010</b> , 56, 1451-9	15
981	Strong synaptic transmission impact by copy number variations in schizophrenia. <b>2010</b> , 107, 10584-9	165
980	Detecting copy number variations from array CGH data based on a conditional random field model. <b>2010</b> , 8, 295-314	12
979	Multi-objective tag SNPs selection using evolutionary algorithms. <b>2010</b> , 26, 1446-52	13



978	MouseIndelDB: a database integrating genomic indel polymorphisms that distinguish mouse strains. <b>2010</b> , 38, D600-6	18
977	Lower linkage disequilibrium at CNVs is due to both recurrent mutation and transposing duplications. <b>2010</b> , 27, 103-11	23
976	Integrative classification and analysis of multiple arrayCGH datasets with probe alignment. <b>2010</b> , 26, 2313-20	6
975	Accurate distinction of pathogenic from benign CNVs in mental retardation. <b>2010</b> , 6, e1000752	42
974	Gene expression variability within and between human populations and implications toward disease susceptibility. <b>2010</b> , 6, e1000910	64
973	Web-based, participant-driven studies yield novel genetic associations for common traits. <b>2010</b> , 6, e1000993	332
972	Structural variation analysis with strobe reads. <b>2010</b> , 26, 1291-8	30
971	Duplications of the critical Rubinstein-Taybi deletion region on chromosome 16p13.3 cause a novel recognisable syndrome. <b>2010</b> , 47, 155-61	45
970	Deletion of Late Cornified Envelope 3B and 3C genes is not associated with atopic dermatitis. <b>2010</b> , 130, 2057-61	24
969	Exploiting sequence similarity to validate the sensitivity of SNP arrays in detecting fine-scaled copy number variations. <b>2010</b> , 26, 1007-14	1
968	The genetic interpretation of area under the ROC curve in genomic profiling. <b>2010</b> , 6, e1000864	239
967	Genome-wide pharmacogenetics of antidepressant response in the GENDEP project. <b>2010</b> , 167, 555-64	280
966	Genome-wide copy number variation in epilepsy: novel susceptibility loci in idiopathic generalized and focal epilepsies. <b>2010</b> , 6, e1000962	348
965	FCGR3B copy number variation is not associated with lupus nephritis in a Chinese population. <b>2010</b> , 19, 158-61	11
964	Phenotypic consequences of copy number variation: insights from Smith-Magenis and Potocki-Lupski syndrome mouse models. <b>2010</b> , 8, e1000543	122
963	Association study of single nucleotide polymorphisms on chromosome 19q13 with abdominal aortic aneurysm. <b>2010</b> , 61, 243-7	10
962	Experimental approaches for identifying schizophrenia risk genes. <b>2010</b> , 4, 587-610	4
961	Organization, Variation and Expression of the Human Genome. <b>2010</b> , 13-26	1

960	Submicroscopic genomic alterations in Silver-Russell syndrome and Silver-Russell-like patients. <b>2010</b> , 47, 816-22	60
959	Comparative analyses of seven algorithms for copy number variant identification from single nucleotide polymorphism arrays. <b>2010</b> , 38, e105	83
958	Did You Know?. <b>2010</b> , 85, 506	
957	Severe Progressive Autism Associated with Two de novo Changes: A 2.6-Mb 2q31.1 Deletion and a Balanced t(14;21)(q21.1;p11.2) Translocation with Long-Range Epigenetic Silencing of LRFN5 Expression. <b>2010</b> , 1, 46-57	30
956	Translating genomic analyses into improved management of coronary artery disease. <b>2010</b> , 6, 507-21	5
955	Neues zur Genetik der Schizophrenie. <b>2010</b> , 4, 297-304	
954	Analysis of X chromosome genomic DNA sequence copy number variation associated with premature ovarian failure (POF). <b>2010</b> , 25, 2139-50	39
953	Ancestry and disease in the age of genomic medicine. <b>2010</b> , 363, 1551-8	139
952	Pervasive gene content variation and copy number variation in maize and its undomesticated progenitor. <b>2010</b> , 20, 1689-99	236
951	Microindel detection in short-read sequence data. <b>2010</b> , 26, 722-9	82
950	An effective model for natural selection in promoters. <b>2010</b> , 20, 685-92	22
949	Entwicklungen in der Neurogenetik am Beispiel der Schizophrenie. <b>2010</b> , 4, 121-130	
948	A novel microdeletion/microduplication syndrome of 19p13.13. <b>2010</b> , 12, 503-11	34
947	Genome-wide association analysis of copy number variations in subarachnoid aneurysmal hemorrhage. <b>2010</b> , 55, 726-30	7
946	Prenatal detection of cryptic rearrangements by multiplex ligation probe amplification in fetuses with ultrasound abnormalities. <b>2010</b> , 12, 376-80	7
945	Natural diversity in flowering responses of <i>Arabidopsis thaliana</i> caused by variation in a tandem gene array. <b>2010</b> , 186, 263-76	28
944	SplittingHeirs. <b>2010</b> ,	1
943	Computational Systems-Biology and Bioinformatics. <b>2010</b> ,	

942	Determinants of renal disease variability in ADPKD. <b>2010</b> , 17, 131-9	43
941	Pharmacogenetics in heart failure: how it will shape the future. <b>2010</b> , 6, 1-10	1
940	References. 203-222	
939	Contrasting methods of quantifying fine structure of human recombination. <b>2010</b> , 11, 45-64	23
938	DNA fluorocode: A single molecule, optical map of DNA with nanometre resolution. <b>2010</b> , 1, 453	73
937	Confirmed rare copy number variants implicate novel genes in schizophrenia. <b>2010</b> , 38, 445-51	110
936	Molecular Cytogenetics in Molecular Diagnostics. <b>2010</b> , 133-153	1
935	Empirical evaluation of oligonucleotide probe selection for DNA microarrays. <b>2010</b> , 5, e9921	13
934	Early identification of cardiovascular risk using genomics and proteomics. <b>2010</b> , 7, 309-17	49
933	Trends and Statistical Challenges in Genomewide Association Studies. <b>2010</b> , 283-308	
932	Application of genetic/genomic approaches to allergic disorders. <b>2010</b> , 126, 425-36; quiz 437-8	15
931	Examination of copy number variations of CHST9 in multiple types of hematologic malignancies. <b>2010</b> , 203, 176-9	8
930	Screening for common copy-number variants in cancer genes. <b>2010</b> , 203, 316-23	2
929	Whole genome sequencing. <b>2010</b> , 628, 215-26	116
928	NOTCH2 in breast cancer: association of SNP rs11249433 with gene expression in ER-positive breast tumors without TP53 mutations. <b>2010</b> , 9, 113	44
927	Copy number variation of the SELENBP1 gene in schizophrenia. <b>2010</b> , 6, 40	8
926	GenEthica. <b>2010</b> , 9, 155-159	
925	Novel candidate cancer genes identified by a large-scale cross-species comparative oncogenomics approach. <b>2010</b> , 70, 883-95	36

924	Genome-wide oligonucleotide array comparative genomic hybridization for etiological diagnosis of mental retardation: a multicenter experience of 1499 clinical cases. <b>2010</b> , 12, 204-12	55
923	Genetic influences modulating the radiological severity of rheumatoid arthritis. <b>2010</b> , 69, 476-82	19
922	The discovery of human genetic variations and their use as disease markers: past, present and future. <b>2010</b> , 55, 403-15	66
921	Theory and methods in cultural neuroscience. <b>2010</b> , 5, 356-61	60
920	Subsampling methods for genomic inference. <b>2010</b> , 4,	50
919	Analysis of copy number variations among diverse cattle breeds. <b>2010</b> , 20, 693-703	214
918	Karyomapping: a universal method for genome wide analysis of genetic disease based on mapping crossovers between parental haplotypes. <b>2010</b> , 47, 651-8	275
917	Genomic microarrays in mental retardation: from copy number variation to gene, from research to diagnosis. <b>2010</b> , 47, 289-97	121
916	Molecular Diagnostics and Cytogenetic Testing. <b>2010</b> , 61-95	
915	Somatic genomic variations in early human prenatal development. <b>2010</b> , 11, 397-401	15
914	Somatic genome variations in health and disease. <b>2010</b> , 11, 387-96	79
913	Evaluation of array comparative genomic hybridization for genetic analysis of chorionic villus sampling from pregnancy loss in comparison to karyotyping and multiplex ligation-dependent probe amplification. <b>2010</b> , 14, 421-4	22
912	Human Genome Sequence and Variation. <b>2010</b> , 31-53	3
911	Human Gene Mutation: Mechanisms and Consequences. <b>2010</b> , 319-363	5
910	Population Genetic Principles and Human Populations. <b>2010</b> , 487-506	
909	Human Evolution. <b>2010</b> , 529-555	
908	Ensembl Genome Browser. <b>2010</b> , 923-939	3
907	Sequence variations at the human leukocyte antigen-linked olfactory receptor cluster do not influence female preferences for male odors. <b>2010</b> , 71, 100-3	8

906	Lower copy numbers of the chemokine CCL3L1 gene in patients with chronic hepatitis C. <b>2010</b> , 52, 153-9	21
905	Genome-wide high-resolution screening in Dupuytren's disease reveals common regions of DNA copy number alterations. <b>2010</b> , 35, 1172-1183.e7	13
904	Nebulin: a study of protein repeat evolution. <b>2010</b> , 402, 38-51	38
903	A preliminary investigation into a genetic basis for cis-3-hexen-1-ol odour perception: A genome-wide association approach. <b>2010</b> , 21, 121-131	36
902	Epimutations and cancer predisposition: importance and mechanisms. <b>2010</b> , 20, 290-8	51
901	Germline copy number variation and cancer risk. <b>2010</b> , 20, 282-9	90
900	The evolution of mammalian chemokine genes. <b>2010</b> , 21, 253-62	109
899	Genome-wide association studies in atherothrombosis. <b>2010</b> , 21, 74-8	24
898	Sperm FISH analysis in two healthy infertile brothers with t(15;18) unbalanced translocation: Implications for genetic counselling and reproductive management. <b>2010</b> , 53, 127-32	7
897	Two neighboring microdeletions of 5q13.2 in a child with oculo-auriculo-vertebral spectrum. <b>2010</b> , 53, 153-8	10
896	Additional cryptic CNVs in mentally retarded patients with apparently balanced karyotypes. <b>2010</b> , 53, 227-33	19
895	Segmental copy-number gain within the region of isopentenyl diphosphate isomerase genes in sporadic amyotrophic lateral sclerosis. <b>2010</b> , 402, 438-42	15
894	A human genome structural variation sequencing resource reveals insights into mutational mechanisms. <b>2010</b> , 143, 837-47	206
893	Neurogenetics: advancing the "next-generation" of brain research. <b>2010</b> , 68, 165-73	35
892	Resumen de la evolución de las técnicas de citogenética y genética molecular para la identificación de las alteraciones genéticas del desarrollo embrionario. <b>2010</b> , 36, 520-525	1
891	Identification of human specific gene duplications relative to other primates by array CGH and quantitative PCR. <b>2010</b> , 95, 203-9	6
890	Accurate and objective copy number profiling using real-time quantitative PCR. <b>2010</b> , 50, 262-70	242
889	[Fetal chromosome technique by microarray-based comparative genomic hybridization]. <b>2010</b> , 17, 1119-23	0

888	A short primer on the functional analysis of copy number variation for biomedical scientists. <b>2010</b> , 628, 119-35	6
887	Sex and age interaction with genetic association of atherogenic uric acid concentrations. <b>2010</b> , 210, 474-8	28
886	Exploring the landscape of the genome. <b>2010</b> , 628, 21-38	4
885	Genetic variation analysis for biomedical researchers: a primer. <b>2010</b> , 628, 1-20	11
884	RECONSTRUCTING DNA COPY NUMBER BY PENALIZED ESTIMATION AND IMPUTATION. <b>2010</b> , 4, 1749-1773	21
883	A gene family-based method for interspecies comparisons of sequencing-based transcriptomes and its use in environmental adaptation analysis. <b>2010</b> , 37, 205-18	3
882	A genome wide association study between copy number variation (CNV) and human height in Chinese population. <b>2010</b> , 37, 779-85	12
881	Copy number changes of CNV regions in intersubspecific crosses of the house mouse. <b>2010</b> , 27, 1845-56	24
880	Nutrigenomics: Integrating Genomic Approaches into Nutrition Research. <b>2010</b> , 347-363	8
879	Clinical genetic testing for patients with autism spectrum disorders. <b>2010</b> , 125, e727-35	281
878	DNA Microarrays and Genetic Testing. <b>2010</b> , 247-265	1
877	Effect of genome-wide association studies, direct-to-consumer genetic testing, and high-speed sequencing technologies on predictive genetic counselling for cancer risk. <b>2010</b> , 11, 890-8	27
876	Targeted chromosomal deletions in human cells using zinc finger nucleases. <b>2010</b> , 20, 81-9	206
875	Deletion of YWHAЕ in a patient with periventricular heterotopias and pronounced corpus callosum hypoplasia. <b>2010</b> , 47, 132-6	32
874	Microarrays for personalized genomic medicine. <b>2010</b> , 52, 1-18	15
873	Chromosomes. <b>2010</b> , 55-138	1
872	POD-FISH: a new technique for parental origin determination based on copy number variation polymorphism. <b>2010</b> , 659, 291-8	11
871	Development of animal models for schizophrenia. <b>2010</b> , 3, 22-6	30

- 870 DNA Sequencing for the Detection of Human Genome Variation. **2010**, 27-37
- 869 Genome-Wide Association Studies and Genotyping Technologies. **2010**, 38-45
- 868 Bioinformatics for next generation sequencing data. **2010**, 1, 294-307 57
- 867 Computational Biology. **2010**, 1
- 866 Towards a comprehensive structural variation map of an individual human genome. **2010**, 11, R52 204
- 865 Copy Number Variation and Human Health. **2010**, 46-59
- 864 Behavioral Neurobiology of Schizophrenia and Its Treatment. **2010**, 6
- 863 Ohnologs in the human genome are dosage balanced and frequently associated with disease. **2010**, 107, 9270-4 191
- 862 Inversion variants in the human genome: role in disease and genome architecture. **2010**, 2, 11 47
- 861 Evolution in health and medicine Sackler colloquium: Genomic disorders: a window into human gene and genome evolution. **2010**, 107 Suppl 1, 1765-71 47
- 860 The role of copy number variation in schizophrenia. **2010**, 10, 25-32 50
- 859 Innovative diagnostic technologies and their significance for personalized medicine. **2010**, 14, 141-7 18
- 858 The quest for genetic risk factors for Crohn's disease in the post-GWAS era. **2011**, 3, 13 10
- 857 Copy number variation in common disease. **2011**, 21, 1-4 26
- 856 Model based clustering approach for identifying structural variation using next generation sequencing data. **2011**,
- 855 Mapping candidate regions and genes for congenital anomalies of the kidneys and urinary tract (CAKUT) by array-based comparative genomic hybridization. **2011**, 26, 136-43 52
- 854 A genomic portrait of human microsatellite variation. **2011**, 28, 303-12 71
- 853 Intragenic higher order repeats in neuroblastoma breakpoint family genes distinguish humans from chimpanzees. **2011**, 28, 1877-92 15

852	Interpretation of array comparative genome hybridization data: a major challenge. <b>2011</b> , 135, 222-7	27
851	Copy number variations at the Prader-Willi syndrome region on chromosome 15 and associations with obesity in whites. <b>2011</b> , 19, 1229-34	20
850	Array comparative genomic hybridization analysis identifies recurrent gain of chromosome 2p25.3 involving the ACP1 and MYCN genes in chronic lymphocytic leukemia. <b>2011</b> , 11 Suppl 1, S17-24	7
849	Screening for copy number variation in genes associated with the long QT syndrome: clinical relevance. <b>2011</b> , 57, 40-7	69
848	The small interferon-induced transmembrane genes and proteins. <b>2011</b> , 31, 183-97	122
847	Overview of Genotyping. <b>2011</b> , 1-23	3
846	DNA Chip Analysis in Genome Discovery. <b>2011</b> , 24-42	2
845	Computational Modeling of Gene Translation and its Potential Applications in Individualized Medicine. <b>2011</b> , 487-503	
844	Clinical application of array-based comparative genomic hybridization by two-stage screening for 536 patients with mental retardation and multiple congenital anomalies. <b>2011</b> , 56, 110-24	20
843	Using MACS to identify peaks from ChIP-Seq data. <b>2011</b> , Chapter 2, Unit 2.14	149
842	Structural variation in two human genomes mapped at single-nucleotide resolution by whole genome de novo assembly. <b>2011</b> , 29, 723-30	99
841	Copy number variations of EphA3 are associated with multiple types of hematologic malignancies. <b>2011</b> , 11, 50-3	26
840	Genetic Basis of Human Biodiversity: An Update. <b>2011</b> , 97-119	1
839	Personalized medicine: progress and promise. <b>2011</b> , 12, 217-44	183
838	Bioinformatics for copy number variation data. <b>2011</b> , 719, 235-49	2
837	Use of matrix-assisted laser desorption/ionization time-of-flight mass spectrometry for multiplex genotyping. <b>2011</b> , 53, 1-29	18
836	A copy number variation morbidity map of developmental delay. <b>2011</b> , 43, 838-46	931
835	Two Faces of Evil: Cancer and Neurodegeneration. <b>2011</b> ,	



834	Cancer Cytogenetics. <b>2011</b> ,	1
833	Functional enrichment analysis with structural variants: pitfalls and strategies. <b>2011</b> , 135, 277-85	21
832	Copy number polymorphisms and anticancer pharmacogenomics. <b>2011</b> , 12, R46	23
831	Evolution of Fungi and Fungal-Like Organisms. <b>2011</b> ,	4
830	High frequency of submicroscopic chromosomal deletions in patients with idiopathic congenital eye malformations. <b>2011</b> , 151, 1087-1094.e45	17
829	Analysis of copy number variants in the cattle genome. <b>2011</b> , 482, 73-7	31
828	[New technologies for genome analysis: Which use in prenatal diagnosis]. <b>2011</b> , 39, 32-41	
827	A parallel SNP array study of genomic aberrations associated with mental retardation in patients and general population in Estonia. <b>2011</b> , 54, 136-43	7
826	Charcot-Marie-Tooth caused by a copy number variation in myelin protein zero. <b>2011</b> , 54, e580-3	22
825	Characterisation of the FAM69 family of cysteine-rich endoplasmic reticulum proteins. <b>2011</b> , 406, 471-7	10
824	Oligonucleotide microarrays in constitutional genetic diagnosis. <b>2011</b> , 11, 521-32	11
823	Copy-number changes in prenatal diagnosis. <b>2011</b> , 11, 579-92	18
822	Joint segmentation, calling, and normalization of multiple CGH profiles. <b>2011</b> , 12, 413-28	70
821	Copy number variants in pharmacogenetic genes. <b>2011</b> , 17, 244-51	70
820	IL28B single nucleotide polymorphisms in the treatment of hepatitis C. <b>2011</b> , 55, 692-701	86
819	Recombination rates in admixed individuals identified by ancestry-based inference. <b>2011</b> , 43, 847-53	85
818	Data-driven approach to detect common copy-number variations and frequency profiles in a population-based Korean cohort. <b>2011</b> , 19, 1167-72	7
817	Genome-wide association: from confounded to confident. <b>2011</b> , 17, 174-84	4

816	A first comparative map of copy number variations in the sheep genome. <b>2011</b> , 97, 158-65	78
815	Inside the CBF locus in Poaceae. <b>2011</b> , 180, 39-45	44
814	Discovery and development of integrative biological markers for schizophrenia. <b>2011</b> , 95, 686-702	25
813	Unraveling the biological mechanisms in Alzheimer's disease--lessons from genomics. <b>2011</b> , 35, 340-7	11
812	Glyoxalase in ageing. <b>2011</b> , 22, 293-301	133
811	Glyoxalase in diabetes, obesity and related disorders. <b>2011</b> , 22, 309-17	178
810	ERBB2 gene amplification in oral squamous cell malignancies: a correlation with tumor progression and gene expression. <b>2011</b> , 112, 90-5	7
809	Identification des biomarqueurs. <b>2011</b> , 78, S161-S164	1
808	Efficient change point detection for genomic sequences of continuous measurements. <b>2011</b> , 27, 161-6	80
807	Human copy number variation and complex genetic disease. <b>2011</b> , 45, 203-26	271
806	Genome-wide association studies and type 2 diabetes. <b>2011</b> , 10, 52-60	82
805	Molecular karyotyping: from microscope to SNP arrays. <b>2011</b> , 76, 208-13	21
804	7 Evolution of the Plant-Symbiotic Fungal Phylum, Glomeromycota. <b>2011</b> , 163-185	39
803	A user's guide to the encyclopedia of DNA elements (ENCODE). <b>2011</b> , 9, e1001046	1060
802	SNP array analysis in constitutional and cancer genome diagnostics--copy number variants, genotyping and quality control. <b>2011</b> , 135, 212-21	34
801	Evolution in the Genus Homo. <b>2011</b> , 42, 47-69	35
800	Array-based Comparative Genomic Hybridization and Its Application to Cancer Genomes and Human Genetics. <b>2011</b> , 10, 77	2
799	. <b>2011</b> ,	16

798	Copy Number Variant Association Studies. <b>2011</b> , 215-230	1
797	Family-based Association Methods. <b>2011</b> , 231-250	
796	Identifying Variations Within Unstable Regions of the Genome Reveal Autism Associated Patterns. <b>2011</b> ,	1
795	Genome-Wide Association Studies of Copy Number Variation in Autism Spectrum Disorder. <b>2011</b> ,	
794	Chromosomal Position Effects and Gene Variegation: Impact in Pathologies. <b>2011</b> , 77-105	
793	SNCA Gene Multiplication: A Model Mechanism of Parkinson Disease. <b>2011</b> ,	2
792	Genetics of childhood obesity. <b>2011</b> , 2011, 845148	38
791	A Method to Assess Linkage Disequilibrium between CNVs and SNPs Inside Copy Number Variable Regions. <b>2011</b> , 2, 17	4
790	Genetic damage and male reproduction. 17-49	3
789	Accuracy of CNV Detection from GWAS Data. <b>2011</b> , 6, e14511	57
788	The characterisation of three types of genes that overlie copy number variable regions. <b>2011</b> , 6, e14814	19
787	MicroRNA genes derived from repetitive elements and expanded by segmental duplication events in mammalian genomes. <b>2011</b> , 6, e17666	69
786	The genetic effect of copy number variations on the risk of type 2 diabetes in a Korean population. <b>2011</b> , 6, e19091	17
785	Copy number variation across European populations. <b>2011</b> , 6, e23087	20
784	Very few RNA and DNA sequence differences in the human transcriptome. <b>2011</b> , 6, e25842	64
783	myKaryoView: a light-weight client for visualization of genomic data. <b>2011</b> , 6, e26345	4
782	GALC deletions increase the risk of primary open-angle glaucoma: the role of Mendelian variants in complex disease. <b>2011</b> , 6, e27134	32
781	A map of copy number variations in Chinese populations. <b>2011</b> , 6, e27341	40

780	SNPs array karyotyping reveals a novel recurrent 20p13 amplification in primary myelofibrosis. <b>2011</b> , 6, e27560	4
779	Genome-wide signatures of 'rearrangement hotspots' within segmental duplications in humans. <b>2011</b> , 6, e28853	9
778	Use of array genomic hybridization technology for constitutional genetic diagnosis in Canada. <b>2011</b> , 16, 211-2	3
777	The Sonoda-Tajima Cell Collection: a human genetics research resource with emphasis on South American indigenous populations. <b>2011</b> , 3, 272-83	5
776	Clinical applicability of sequence variations in genes related to drug metabolism. <b>2011</b> , 12, 445-54	9
775	Genomic Diversity in Pig ( <i>Sus scrofa</i> ) and its Comparison with Human and other Livestock. <b>2011</b> , 12, 138-46	25
774	Use of Array Genomic Hybridization Technology in Prenatal Diagnosis in Canada. <b>2011</b> , 33, 1256-1259	30
773	Ensembl BioMarts: a hub for data retrieval across taxonomic space. <b>2011</b> , 2011, bar030	751
772	Segmental copy number loss of SFMBT1 gene in elderly individuals with ventriculomegaly: a community-based study. <b>2011</b> , 50, 297-303	25
771	Genetics of Aneurysms and Arteriovenous Malformations. <b>2011</b> , 1292-1300	
770	Classical Genetic Studies of Schizophrenia. <b>2011</b> , 245-268	1
769	Detecting simultaneous variant intervals in aligned sequences. <b>2011</b> , 5,	47
768	Human genetics and genomics a decade after the release of the draft sequence of the human genome. <b>2011</b> , 5, 577-622	65
767	X-chromosome duplications in males with mental retardation: pathogenic or benign variants?. <b>2011</b> , 79, 71-8	5
766	High-resolution molecular karyotyping in patients with developmental delay and/or multiple congenital anomalies in a clinical setting. <b>2011</b> , 79, 147-57	23
765	RUNX2 analysis of Danish cleidocranial dysplasia families. <b>2011</b> , 79, 254-63	17
764	Bringing immunoglobulin knowledge up to date: how should we treat today?. <b>2011</b> , 166, 16-25	16
763	Investigation of the HIN200 locus in UK SLE families identifies novel copy number variants. <b>2011</b> , 75, 383-97	5

762	Genome structural variation discovery and genotyping. <b>2011</b> , 12, 363-76		947
761	A decade's perspective on DNA sequencing technology. <i>Nature</i> , <b>2011</b> , 470, 198-203	50.4	605
760	Copy number variation and selection during reprogramming to pluripotency. <i>Nature</i> , <b>2011</b> , 471, 58-62	50.4	753
759	Comprehensive assessment of array-based platforms and calling algorithms for detection of copy number variants. <b>2011</b> , 29, 512-20		333
758	Clinical assessment and genomic landscape of a consanguineous family with three Kallmann syndrome descendants. <b>2011</b> , 13, 166-71		7
757	Integrated genetic and genomic approach in the Singapore Translational and Clinical Research in Psychosis Study: an overview. <b>2011</b> , 5, 91-9		3
756	Rapid clinical-scale propagation of mesenchymal stem cells using cultures initiated with immunoselected bone marrow CD105+ cells. <b>2011</b> , 15, 1983-8		9
755	Association of Gene Copy Number Variation and Rheumatoid Arthritis Susceptibility. <b>2020</b> , 2020, 7189626		2
754	Genome-wide characterization of copy number variations in the host genome in genetic resistance to Marek's disease using next generation sequencing. <b>2020</b> , 21, 77		1
753	A Deep Learning Framework to Predict Tumor Tissue-of-Origin Based on Copy Number Alteration. <b>2020</b> , 8, 701		12
752	Human genetic diversity in health and disease. <b>2020</b> , 123-136		1
751	Genetic copy number variants, cognition and psychosis: a meta-analysis and a family study. <b>2021</b> , 26, 5307-5319		5
750	New Insights into Potocki-Shaffer Syndrome: Report of Two Novel Cases and Literature Review. <b>2020</b> , 10,		2
749	Analysis of Chromosomal Copy Number in First-Trimester Pregnancy Loss Using Next-Generation Sequencing. <b>2020</b> , 11, 545856		5
748	Chromosomal abnormality, laboratory techniques, tools and databases in molecular Cytogenetics. <b>2020</b> , 47, 9055-9073		1
747	Interplay of microRNAs to genetic, epigenetic, copy number variations of cervical cancer related genes. <b>2020</b> , 142, 103184		1
746	Fuzzy methods for the detection of copy number variations in comparative genomic hybridization arrays. <b>2020</b> , 27, 3647-3654		1
745	DINTD: Detection and Inference of Tandem Duplications From Short Sequencing Reads. <b>2020</b> , 11, 924		1

744	Application value of NIPT for uncommon fetal chromosomal abnormalities. <b>2020</b> , 13, 39	4
743	Both Methylation and Copy Number Variation Participated in the Varied Expression of PRAME in Multiple Myeloma. <b>2020</b> , 13, 7545-7553	0
742	Relevance of Copy Number Variation at Chromosome X in Male Fetuses Inherited from the Mother May Be Ascertained by Including Male Relatives from the Maternal Lineage in Addition to Trio Analyses. <b>2020</b> , 11,	1
741	Pan-genome: A promising resource for noncoding RNA discovery in plants. <b>2020</b> , 13, e20046	7
740	Copy number variants outperform SNPs to reveal genotype-temperature association in a marine species. <b>2020</b> , 29, 4765-4782	25
739	Detection of InDel and CNV of gene and their associations with bovine growth traits. <b>2020</b> , 1-8	2
738	Identification and population genetic analyses of copy number variations in six domestic goat breeds and Bezoar ibexes using next-generation sequencing. <b>2020</b> , 21, 840	0
737	A Novel XGBoost Method to Identify Cancer Tissue-of-Origin Based on Copy Number Variations. <b>2020</b> , 11, 585029	9
736	Susceptibility to Malaria: Influence of Combined Polymorphisms of IgG3 Gm Allotypes and Fc Gamma Receptors IIA, IIIA, and IIIB. <b>2020</b> , 11, 608016	2
735	Copy Number Analysis Reveal Genetic Risks of Penile Cancer. <b>2020</b> , 10, 596261	0
734	Tunability enhancement of gene regulatory motifs through competition for regulatory protein resources. <b>2020</b> , 102, 052410	3
733	Effects of Agricultural Fungicide Use on <i>Aspergillus fumigatus</i> Abundance, Antifungal Susceptibility, and Population Structure. <b>2020</b> , 11,	13
732	Whole genome variants across 57 pig breeds enable comprehensive identification of genetic signatures that underlie breed features. <b>2020</b> , 11, 115	1
731	Genetic Risk Profiling in Parkinson's Disease and Utilizing Genetics to Gain Insight into Disease-Related Biological Pathways. <b>2020</b> , 21,	9
730	Lessons Learned from CNV Analysis of Major Birth Defects. <b>2020</b> , 21,	3
729	Are mental disorders orphan diseases?. <b>2020</b> , 45, 75-78	0
728	Copy number variant analysis and expression profiling of the olfactory receptor-rich 11q11 region in obesity predisposition. <b>2020</b> , 25, 100656	1
727	Understanding the Early Evolutionary Stages of a Tandem <i>Drosophilamelanogaster</i> -Specific Gene Family: A Structural and Functional Population Study. <b>2020</b> , 37, 2584-2600	6

726	Twin Research in the Post-Genomic Era: Dissecting the Pathophysiological Effects of Adversity and the Social Environment. <b>2020</b> , 21,	3
725	Couples experiences of receiving uncertain results following prenatal microarray or exome sequencing: A mixed-methods systematic review. <b>2020</b> , 40, 1028-1039	13
724	High confidence copy number variants identified in Holstein dairy cattle from whole genome sequence and genotype array data. <b>2020</b> , 10, 8044	6
723	The diagnostic yield of intellectual disability: combined whole genome low-coverage sequencing and medical exome sequencing. <b>2020</b> , 13, 70	5
722	Novel copy number variations of the CHRM3 gene associated with gene expression and growth traits in Chinese Datong yak ( <i>Bos grunniens</i> ). <b>2020</b> , 48, 156-165	1
721	Population Structure, Stratification, and Introgression of Human Structural Variation. <b>2020</b> , 182, 189-199.e15	23
720	Genomic Structural Diversity in Local Goats: Analysis of Copy-Number Variations. <b>2020</b> , 10,	3
719	Insight into the Possible Formation Mechanism of the Intersex Phenotype of Lanzhou Fat-Tailed Sheep Using Whole-Genome Resequencing. <b>2020</b> , 10,	4
718	Whole-genome resequencing of wild and domestic sheep identifies genes associated with morphological and agronomic traits. <b>2020</b> , 11, 2815	48
717	Copy number variations of TOP2B gene are associated with growth traits in Chinese sheep breeds. <b>2020</b> , 1-5	0
716	Importance of Genetic Studies of Cardiometabolic Disease in Diverse Populations. <b>2020</b> , 126, 1816-1840	7
715	Integrating Genome-Wide CNVs Into QTLs and High Confidence GWAScore Regions Identified Positional Candidates for Sheep Economic Traits. <b>2020</b> , 11, 569	4
714	Genome-wide high-resolution mapping of mitotic DNA synthesis sites and common fragile sites by direct sequencing. <b>2020</b> , 30, 1009-1023	21
713	Characterization of the primate TRIM gene family reveals the recent evolution in primates. <b>2020</b> , 295, 1281-1294	3
712	Genome-wide detection of copy number variants in European autochthonous and commercial pig breeds by whole-genome sequencing of DNA pools identified breed-characterising copy number states. <b>2020</b> , 51, 541-556	7
711	1Q12 Loci Movement in the Interphase Nucleus Under the Action of ROS Is an Important Component of the Mechanism That Determines Copy Number Variation of Satellite III (1q12) in Health and Schizophrenia. <b>2020</b> , 8, 386	4
710	Whole genome resequencing of the Iranian native dogs and wolves to unravel variome during dog domestication. <b>2020</b> , 21, 207	1
709	CNVmap: A Method and Software To Detect and Map Copy Number Variants from Segregation Data. <b>2020</b> , 214, 561-576	1

708	Three-dimensional genome: developmental technologies and applications in precision medicine. <b>2020</b> , 65, 497-511	16
707	Sensitivity to gene dosage and gene expression affects genes with copy number variants observed among neuropsychiatric diseases. <b>2020</b> , 13, 55	7
706	Population Structure, and Selection Signatures Underlying High-Altitude Adaptation Inferred From Genome-Wide Copy Number Variations in Chinese Indigenous Cattle. <b>2019</b> , 10, 1404	7
705	Characterization of A Homozygous Deletion of Steroid Hormone Biosynthesis Genes in Horse Chromosome 29 as A Risk Factor for Disorders of Sex Development and Reproduction. <b>2020</b> , 11,	5
704	NanoVar: accurate characterization of patients' genomic structural variants using low-depth nanopore sequencing. <b>2020</b> , 21, 56	27
703	eBreCaP: extreme learning-based model for breast cancer survival prediction. <b>2020</b> , 14, 160-169	9
702	Ancient Patrilineal Lines and Relatively High ECA Y Diversity Preserved in Indigenous Horses Revealed With Novel Y-Chromosome Markers. <b>2020</b> , 11, 467	0
701	Exposure to the Heavy-Metal Lead Induces DNA Copy Number Alterations in Zebrafish Cells. <b>2020</b> , 33, 2047-2053	1
700	A genome-wide survey of copy number variations reveals an asymmetric evolution of duplicated genes in rice. <b>2020</b> , 18, 73	2
699	Population-specific profiling of CCL3L1 copy number of the three major ethnic groups in Malaysia and the implication on HIV susceptibility. <b>2020</b> , 754, 144821	2
698	The Association of the Copy Number Variation of the Gene with Growth Traits of Chinese Cattle. <b>2020</b> , 10,	7
697	CRSCNV: A Cross-Model-Based Statistical Approach to Detect Copy Number Variations in Sequence Data. <b>2020</b> , 8, 2302-2312	1
696	Easy One-Step Amplification and Labeling Procedure for Copy Number Variation Detection. <b>2020</b> , 66, 463-473	5
695	The Integration of Multiple Nuclear-Encoded Transgenes in the Green Alga Results in Higher Transcription Levels. <b>2019</b> , 10, 1784	6
694	Cytogenetic and molecular diagnostic testing associated with prenatal and postnatal birth defects. <b>2020</b> , 112, 293-306	6
693	Pathogenic copy number variants are detected in a subset of patients with gastrointestinal malformations. <b>2020</b> , 8, e1084	2
692	Rare copy number variants in over 100,000 European ancestry subjects reveal multiple disease associations. <b>2020</b> , 11, 255	17
691	Cancer LncRNA Census reveals evidence for deep functional conservation of long noncoding RNAs in tumorigenesis. <b>2020</b> , 3, 56	77



690	Haplotype-resolved genomes provide insights into structural variation and gene content in Angus and Brahman cattle. <b>2020</b> , 11, 2071	38
689	Copy number variation in human genomes from three major ethno-linguistic groups in Africa. <b>2020</b> , 21, 289	3
688	Sudden Cardiac Death and Copy Number Variants: What Do We Know after 10 Years of Genetic Analysis?. <b>2020</b> , 47, 102281	10
687	A Roadmap for Understanding the Evolutionary Significance of Structural Genomic Variation. <b>2020</b> , 35, 561-572	57
686	The combination between gene copy number variation and growth traits in Chinese cattle. <b>2021</b> , 32, 683-687	0
685	Copy Number Variations and Expression Levels of Guanylate-Binding Protein 6 Gene Associated with Growth Traits of Chinese Cattle. <b>2020</b> , 10,	1
684	Comparative analysis of genome editing systems, Cas9 and BE3, in silkworms. <b>2020</b> , 158, 486-492	1
683	Hierarchical discovery of large-scale and focal copy number alterations in low-coverage cancer genomes. <b>2020</b> , 21, 147	3
682	Clinical Integration of Genome Diagnostics for Congenital Anomalies of the Kidney and Urinary Tract. <b>2020</b> , 16, 128-137	11
681	Genome-Wide Analysis of Copy Number Variation in Latin American Parkinson's Disease Patients. <b>2021</b> , 36, 434-441	4
680	Genetic control of tumor development in malformation syndromes. <b>2021</b> , 185, 324-335	0
679	Systematic identification of genetic systems associated with phenotypes in patients with rare genomic copy number variations. <b>2021</b> , 140, 457-475	5
678	A genome-wide study of the relationship between chromosomal abnormalities and gene expression in colorectal tumors. <b>2021</b> , 60, 250-262	4
677	Identification of copy number variants by NGS-based NIPT at low sequencing depth. <b>2021</b> , 256, 297-301	4
676	A copy number variant at the HPDA-D12 locus confers compact plant architecture in cotton. <b>2021</b> , 229, 2091-2103	8
675	Clinical relevance of blood-based ctDNA analysis: mutation detection and beyond. <b>2021</b> , 124, 345-358	77
674	Copy-number variation of the NPHP1 gene in patients with juvenile Nephronophthisis. <b>2021</b> , 76, 16-24	1
673	A Comprehensive Analysis of Unique and Recurrent Copy Number Variations in Alzheimer's Disease and its Related Disorders. <b>2020</b> , 17, 926-938	

672	Comprehensive Assessment of Copy Number Alterations Uncovers Recurrent and Copy Gain in Medullary Thyroid Carcinoma. <b>2021</b> , 13,	2
671	Network-based analysis of allele frequency distribution among multiple populations identifies adaptive genomic structural variants.	0
670	Copy Number Variation in the Human Genome. <b>2021</b> , 275-300	
669	Mutations and Polymorphisms: What Is The Difference?. <b>2021</b> , 1-21	4
668	Genetic Analysis of Multiple Myeloma Identifies Cytogenetic Alterations Implicated in Disease Complexity and Progression. <b>2021</b> , 13,	2
667	Molecular combing solutions to characterize replication kinetics and genome rearrangements. <b>2021</b> , 47-71	0
666	Gene signatures of 6-methyladenine regulators in women with lung adenocarcinoma and development of a risk scoring system: a retrospective study using the cancer genome atlas database. <b>2021</b> , 13, 3957-3968	1
665	Statistical Considerations on NGS Data for Inferring Copy Number Variations. <b>2021</b> , 2243, 27-58	0
664	Applications of Machine Learning in Healthcare.	5
663	Quantitative evaluation of PTPN22 copy number variation by digital droplet PCR and association with type 2 diabetes risk. <b>2021</b> , 68, 153-162	
662	The genetic landscape of polycystic kidney disease in Ireland. <b>2021</b> , 29, 827-838	3
661	Conservation of copy number profiles during engraftment and passaging of patient-derived cancer xenografts. <b>2021</b> , 53, 86-99	44
660	Rapid parallel adaptation despite gene flow in silent crickets. <b>2021</b> , 12, 50	8
659	A global analysis of CNVs in Chinese indigenous fine-wool sheep populations using whole-genome resequencing. <b>2021</b> , 22, 78	4
658	Genomic disorders in the genomics era. <b>2021</b> , 35-59	
657	CNVIntegrate: the first multi-ethnic database for identifying copy number variations associated with cancer. <b>2021</b> , 2021,	2
656	MLIP genotype as a predictor of pharmacological response in primary open-angle glaucoma and ocular hypertension. <b>2021</b> , 11, 1583	0
655	A map of copy number variations in the Tunisian population: a valuable tool for medical genomics in North Africa. <b>2021</b> , 6, 3	1

654	Identification of Copy Number Variation Among Nonsyndromic Cleft Lip and or Without Cleft Palate With Hypodontia: A Genome-Wide Association Study. <b>2021</b> , 12, 637306	2
653	Copy Number Variations of Glycoside Hydrolase 45 Genes in <i>Bursaphelenchus xylophilus</i> and Their Impact on the Pathogenesis of Pine Wilt Disease. <b>2021</b> , 12, 275	1
652	A 44-kb deleted-type copy number variation is associated with decreasing complement component activity and calf mortality in Japanese Black cattle. <b>2021</b> , 22, 107	0
651	A Mechanism Leading to Changes in Copy Number Variations Affected by Transcriptional Level Might Be Involved in Evolution, Embryonic Development, Senescence, and Oncogenesis Mediated by Retrotransposons. <b>2021</b> , 9, 618113	0
650	Whole-Genome Diversification Analysis of the Hornbeam Species Reveals Speciation and Adaptation Among Closely Related Species. <b>2021</b> , 12, 581704	
649	Impaired Reproductive Function in Equines: From Genetics to Genomics. <b>2021</b> , 11,	5
648	Investigating the Effect of Imputed Structural Variants from Whole-Genome Sequence on Genome-Wide Association and Genomic Prediction in Dairy Cattle. <b>2021</b> , 11,	1
647	Genetic architecture of Tourette syndrome: our current understanding. <b>2021</b> , 51, 2201-2209	2
646	DNA copy number variation: Main characteristics, evolutionary significance, and pathological aspects. <b>2021</b> , 44, 548-559	10
645	Aquila enables reference-assisted diploid personal genome assembly and comprehensive variant detection based on linked reads. <b>2021</b> , 12, 1077	2
644	Chances and Challenges of New Genetic Screening Technologies (NIPT) in Prenatal Medicine from a Clinical Perspective: A Narrative Review. <b>2021</b> , 12,	4
643	Ultrasound wave exchange between COVID-19 virus and host cells. <b>2021</b> , 33,	
642	Polymorphisms of AMY1A gene and their association with growth, carcass traits and feed intake efficiency in chickens. <b>2021</b> , 113, 583-594	2
641	The haplolethality paradox of the wupA gene in <i>Drosophila</i> . <b>2021</b> , 17, e1009108	
640	Identification of deleterious recessive haplotypes and candidate deleterious recessive mutations in Japanese Black cattle. <b>2021</b> , 11, 6687	3
639	Thermal adaptation rather than demographic history drives genetic structure inferred by copy number variants in a marine fish. <b>2021</b> , 30, 1624-1641	4
638	DNA replication, transcription, and H3K56 acetylation regulate copy number and stability at tandem repeats. <b>2021</b> ,	1
637	A Single-Run Next-Generation Sequencing (NGS) Assay for the Simultaneous Detection of Both Gene Mutations and Large Chromosomal Abnormalities in Patients with Myelodysplastic Syndromes (MDS) and Related Myeloid Neoplasms. <b>2021</b> , 13,	0

636	Classical, Molecular, and Genomic Cytogenetics of the Pig, a Clinical Perspective. <b>2021</b> , 11,	3
635	Prenatal Diagnosis of Chromosomal Abnormalities: From Karyotype to Microarray. <b>2021</b> , 547-571	0
634	Re-evaluation of single nucleotide variants and identification of structural variants in a cohort of 45 sudden unexplained death cases. <b>2021</b> , 135, 1341-1349	4
633	PRENATAL TANIDA MĀRODZĀNĀ ROLĪ157-168	
632	A genome-wide scan of copy number variants in three Iranian indigenous river buffaloes. <b>2021</b> , 22, 305	4
631	Extensive genome-wide duplications in the eastern oyster (). <b>2021</b> , 376, 20200164	5
630	Copy number variation underlies complex phenotypes in domestic dog breeds and other canids. <b>2021</b> , 31, 762-774	0
629	Luminal A Breast Cancer Co-expression Network: Structural and Functional Alterations. <b>2021</b> , 12, 629475	4
628	Genome-wide detection of CNV regions and their potential association with growth and fatness traits in Duroc pigs. <b>2021</b> , 22, 332	5
627	Samplot: a platform for structural variant visual validation and automated filtering. <b>2021</b> , 22, 161	12
626	Detection of copy number variants in African goats using whole genome sequence data. <b>2021</b> , 22, 398	0
625	Genome-wide detection of CNVs and their association with performance traits in broilers. <b>2021</b> , 22, 354	2
624	The application of expanded noninvasive prenatal screening for genome-wide chromosomal abnormalities and genetic counseling. <b>2021</b> , 34, 2710-2716	2
623	Genomic variations and epigenomic landscape of the Medaka Inbred Kiyosu-Karlsruhe (MIKK) panel.	3
622	Genomic characterization of world's longest selection experiment in mouse reveals the complexity of polygenic traits..	1
621	Genomics of Adaptations in Ungulates. <b>2021</b> , 11,	
620	Genome-Wide Detection of Copy Number Variations and Their Association With Distinct Phenotypes in the World's Sheep. <b>2021</b> , 12, 670582	2
619	Pervasive cis effects of variation in copy number of large tandem repeats on local DNA methylation and gene expression. <b>2021</b> , 108, 809-824	8

618	Expanding the Scope of Non-invasive Prenatal Testing to Detect Fetal Chromosomal Copy Number Variations. <b>2021</b> , 8, 649169	2
617	Genome-wide copy number variations in a large cohort of bantu African children. <b>2021</b> , 14, 129	1
616	VarGenius-HZD allows accurate detection of rare homozygous or hemizygous deletions in targeted sequencing leveraging breadth of coverage.	
615	Genome Scan for Variable Genes Involved in Environmental Adaptations of Nubian Ibex. <b>2021</b> , 89, 448-457	
614	Genetic and pharmacological causes of germline hypermutation.	3
613	HBOS-CNV: A New Approach to Detect Copy Number Variations From Next-Generation Sequencing Data. <b>2021</b> , 12, 642473	0
612	Copy number variation and neuropsychiatric illness. <b>2021</b> , 68, 57-63	8
611	Genome-wide detection of copy number variations in Tharparkar cattle. <b>2021</b> , 1-8	3
610	Application of CRISPR-Cas9 gene editing for congenital heart disease. <b>2021</b> , 64, 269-279	4
609	Comprehensive Evaluation of Non-invasive Prenatal Screening to Detect Fetal Copy Number Variations. <b>2021</b> , 12, 665589	1
608	Population differentiated copy number variation of Bos taurus, Bos indicus and their African hybrids. <b>2021</b> , 22, 531	2
607	Benchmarking germline CNV calling tools from exome sequencing data. <b>2021</b> , 11, 14416	3
606	Integrative analysis of prognostic long non-coding RNAs with copy number variation in bladder cancer. <b>2021</b> , 22, 664-681	2
605	Copy number variations with adaptive potential in caribou (Rangifer tarandus): genome architecture and new annotated genome assembly.	
604	Genes and Pseudogenes: Complexity of the RCCX Locus and Disease. <b>2021</b> , 12, 709758	2
603	The potential of expanded noninvasive prenatal screening for detection of microdeletion and microduplication syndromes. <b>2021</b> , 41, 1332-1342	1
602	Genome structural variation in human evolution. <b>2021</b> ,	3
601	Development and Evaluation of a Droplet Digital PCR Assay for 8p23 EDefensin Cluster Copy Number Determination. <b>2021</b> , 25, 607-615	0

600	The Thousand Polish Genomes Project—a national database of Polish variant allele frequencies.	0
599	Discovery of genomic variation across a generation. <b>2021</b> , 30, R174-R186	2
598	Directed yeast genome evolution by controlled introduction of trans-chromosomal structural variations.	0
597	Profiling variable-number tandem repeat variation across populations using repeat-pangenome graphs. <b>2021</b> , 12, 4250	4
596	A Reassessment of Copy Number Variations in Congenital Heart Defects: Picturing the Whole Genome. <b>2021</b> , 12,	1
595	A genome-wide analysis of the molecular alterations occurring in the adenomatous and carcinomatous components of the same tumor based on the adenoma-carcinoma sequence. <b>2021</b> , 71, 582-593	1
594	Evidence for opposing selective forces operating on human-specific duplicated TCAF genes in Neanderthals and humans. <b>2021</b> , 12, 5118	0
593	Copy number variation in triple-negative breast cancer samples associated with lymph node metastasis. <b>2021</b> , 23, 743-753	4
592	CNV-MEANN: A Neural Network and Mind Evolutionary Algorithm-Based Detection of Copy Number Variations From Next-Generation Sequencing Data. <b>2021</b> , 12, 700874	2
591	CNest: A Novel Copy Number Association Discovery Method Uncovers 862 New Associations from 200,629 Whole Exome Sequence Datasets in the UK Biobank.	0
590	Copy number variation and expression of exportin-4 associates with severity of fibrosis in metabolic associated fatty liver disease. <b>2021</b> , 70, 103521	5
589	Genome-wide CNV investigation suggests a role for cadherin, Wnt, and p53 pathways in primary open-angle glaucoma. <b>2021</b> , 22, 590	1
588	Genomic basis of deep-water adaptation in Arctic Charr ( <i>Salvelinus alpinus</i> ) morphs. <b>2021</b> , 30, 4415-4432	5
587	The genetic consequences of dog breed formation—Accumulation of deleterious genetic variation and fixation of mutations associated with myxomatous mitral valve disease in cavalier King Charles spaniels. <b>2021</b> , 17, e1009726	0
586	ER expression associates with poor prognosis in male lung squamous carcinoma after radical resection. <b>2021</b> , 21, 1043	0
585	HandyCNV: Standardized Summary, Annotation, Comparison, and Visualization of Copy Number Variant, Copy Number Variation Region, and Runs of Homozygosity. <b>2021</b> , 12, 731355	2
584	Characterization of full-length LINE-1 insertions in 154 genomes. <b>2021</b> , 113, 3804-3810	0
583	Genome-wide CNV analysis reveals variants associated with high-altitude adaptation and meat traits in Qaidam cattle. <b>2021</b> , 54, 8-16	0

582 Reproductive genetics. **2022**, 21-46.e3

581 Prediction and classification of diabetes mellitus using genomic data. **2021**, 235-292

5

580 An atlas of CNV maps in cattle, goat and sheep. **2021**, 64, 1747-1764

8

579 Neurexins: molecular codes for shaping neuronal synapses. **2021**, 22, 137-151

17

578 RaceIQ and Genes.

1

577 Relevance of Copy Number Variation to Human Genetic Disease. 1-10

1

576 Subtelomeres: Evolution in the Human Genome.

1

575 Nucleic Acids as Detection Tools. 401-431

1

574 Developmental Instability, Mutation Load, and Neurodevelopmental Disorders. 81-110

1

573 References. 1-124

1

572 Candidate Gene and Genome-Wide Association Studies in Behavioral Medicine. **2010**, 423-441

3

571 Tissue-Specific eQTL in Zebrafish. **2020**, 2082, 239-249

1

570 Research Methods for Genetic Studies. **2008**, 181-199

1

569 Studying cancer genomics through next-generation DNA sequencing and bioinformatics. **2014**, 1168, 83-98

9

568 Application of BAC-probes to visualize copy number variants (CNVs). **2015**, 1227, 299-307

5

567 Principles of Clinical Molecular Biology. **2008**, 3-32

1

566 Comparative genomic hybridization: microarray design and data interpretation. **2009**, 529, 37-49

10

565 Concordance Between Tumor and Germline DNA. **2008**, 91-101

2

564	Comparative genomic hybridization by representational oligonucleotide microarray analysis. <b>2009</b> , 556, 33-46	3
563	Copy number variations in the human genome and strategies for analysis. <b>2010</b> , 628, 103-17	5
562	Genetic mapping and positional cloning. <b>2010</b> , 597, 13-32	10
561	Copy number variation. <b>2011</b> , 713, 167-83	3
560	Single nucleotide polymorphism microarray analysis of genetic alterations in cancer. <b>2011</b> , 730, 235-58	16
559	Molecular Basis of Cardiac Development and Congenital Heart Disease. <b>2012</b> , 317-339	2
558	DNA Copy Number Profiling in Normal and Tumor Genomes. <b>2010</b> , 259-281	10
557	Cancer Genomic and Epigenomic Variations in Sub-Saharan Africa. <b>2017</b> , 21-36	1
556	Detection of Copy Number Variations (CNVs) Based on the Coverage Depth from the Next Generation Sequencing Data. <b>2017</b> , 13-22	2
555	Identification of Deletion Polymorphisms from Haplotypes. <b>2007</b> , 354-365	7
554	Genetic dissection of host resistance to Mycobacterium tuberculosis: the sst1 locus and the lpr1 gene. <b>2008</b> , 321, 123-48	45
553	Statistical Analysis of Single Nucleotide Polymorphism Microarrays in Cancer Studies. <b>2011</b> , 225-255	3
552	Catalog of Genetic Variations (SNPs and CNVs) and Analysis Tools for Thai Genetic Studies. <b>2010</b> , 130-140	2
551	Human and Primate Subtelomeres. <b>2014</b> , 153-164	3
550	Human Genetics of Ventricular Septal Defect. <b>2016</b> , 307-328	3
549	Toxicogenomics: transcription profiling for toxicology assessment. <b>2009</b> , 99, 325-66	13
548	Genetic Discovery for Congenital Heart Defects. <b>2016</b> , 355-360	1
547	Cancer Genomics in Precision Oncology: Applications, Challenges, and Prospects. <b>2020</b> , 453-499	6



546	Principles of Clinical Cytogenetics. <b>2007</b> , 59-88	3
545	Genetics of Cerebral Cavernous Malformations. <b>2011</b> , 4127-4133	1
544	Genetic Basis of Kidney Disease. <b>2012</b> , 1554-1569	2
543	Clinical Neurogenetics. <b>2012</b> , 704-734	3
542	Association analysis of KMT2D copy number variation as a positional candidate for growth traits. <b>2020</b> , 753, 144799	3
541	The Emerging Field of Noncoding RNAs and Their Importance in Pediatric Diseases. <b>2020</b> , 221S, S11-S19	2
540	Unravelling the genetics of autism spectrum disorders. 53-111	2
539	DNA copy number and structural variation (CNV) contributions to adult and childhood obesity. <b>2020</b> , 48, 1819-1828	2
538	Probabilistic Graphical Models for Next-generation Genomics and Genetics. <b>2014</b> , 3-29	2
537	Anesthetic sensitivity: learning to fly. <b>2009</b> , 111, 5-7	2
536	Combinatorial content of CCL3L and CCL4L gene copy numbers influence HIV-AIDS susceptibility in Ukrainian children. <b>2009</b> , 23, 679-88	37
535	Review of Preferential Susceptible Genes in Microtia Patients Through Various Approaches. <b>2020</b> , 31, 538-541	5
534	Copy number variation is associated with gene expression change in archaea. <b>2018</b> , 4,	9
533	Copy number variants implicate cardiac function and development pathways in earthquake-induced stress cardiomyopathy.	1
532	Unique genomic features and deeply-conserved functions of long non-coding RNAs in the Cancer LncRNA Census (CLC).	4
531	Effects of agricultural fungicide use on <i>Aspergillus fumigatus</i> abundance, antifungal susceptibility, and population structure.	4
530	Profiling variable-number tandem repeat variation across populations using repeat-pangenome graphs.	3
529	Non-B-form DNA structures mark centromeres.	1

528	De novo assembly of two Swedish genomes reveals missing segments from the human GRCh38 reference and improves variant calling of population-scale sequencing data.	5
527	Complex haplotypes of metabolizing GSTM1 gene deletion harbors signatures of a selective sweep in East Asian populations.	1
526	Whole genome sequencing reveals high complexity of copy number variation at insecticide resistance loci in malaria mosquitoes.	2
525	Aquila: diploid personal genome assembly and comprehensive variant detection based on linked reads.	1
524	NanoVar: Accurate Characterization of Patients' Genomic Structural Variants Using Low-Depth Nanopore Sequencing.	2
523	Haplotype-Resolved Cattle Genomes Provide Insights Into Structural Variation and Adaptation.	1
522	Population Structure, Stratification and Introgression of Human Structural Variation.	1
521	De novo assembly of a Tibetan genome and identification of novel structural variants associated with high altitude adaptation.	1
520	CNVmap: a method and software to detect and map copy number variants from segregation data.	1
519	Conservation of copy number profiles during engraftment and passaging of patient-derived cancer xenografts.	2
518	The C20orf133 gene is disrupted in a patient with Kabuki syndrome. <b>2009</b> , 2009,	4
517	Genetic basis of human congenital anomalies of the kidney and urinary tract. <b>2018</b> , 128, 4-15	50
516	Functional and population genetic features of copy number variations in two dairy cattle populations. <b>2020</b> , 21, 89	12
515	Histone Methylation and the Initiation of Cancer. <b>2008</b> , 109-150	6
514	Accurately assessing the risk of schizophrenia conferred by rare copy-number variation affecting genes with brain function. <b>2010</b> , 6, e1001097	118
513	Identifying signatures of natural selection in Tibetan and Andean populations using dense genome scan data. <b>2010</b> , 6, e1001116	395
512	Whole Genome Sequencing Identifies a 78 kb Insertion from Chromosome 8 as the Cause of Charcot-Marie-Tooth Neuropathy CMTX3. <b>2016</b> , 12, e1006177	15
511	A constitutional translocation t(1;17)(p36.2;q11.2) in a neuroblastoma patient disrupts the human NBPF1 and ACCN1 genes. <b>2008</b> , 3, e2207	39

510	Cataloging coding sequence variations in human genome databases. <b>2008</b> , 3, e3575	11
509	Geographical affinities of the HapMap samples. <b>2009</b> , 4, e4684	21
508	Genetic structures of copy number variants revealed by genotyping single sperm. <b>2009</b> , 4, e5236	3
507	Identification of copy number variants defining genomic differences among major human groups. <b>2009</b> , 4, e7230	28
506	Whole genome distribution and ethnic differentiation of copy number variation in Caucasian and Asian populations. <b>2009</b> , 4, e7958	44
505	Early embryonic chromosome instability results in stable mosaic pattern in human tissues. <b>2010</b> , 5, e9591	43
504	Malignant precursor cells pre-exist in human breast DCIS and require autophagy for survival. <b>2010</b> , 5, e10240	109
503	Genome rearrangements detected by SNP microarrays in individuals with intellectual disability referred with possible Williams syndrome. <b>2010</b> , 5, e12349	11
502	Ultra high throughput sequencing in human DNA variation detection: a comparative study on the NDUFA3-PRPF31 region. <b>2010</b> , 5, e13071	7
501	Infantile convulsions with paroxysmal dyskinesia (ICCA syndrome) and copy number variation at human chromosome 16p11. <b>2010</b> , 5, e13750	14
500	Chromosomal minimal critical regions in therapy-related leukemia appear different from those of de novo leukemia by high-resolution aCGH. <b>2011</b> , 6, e16623	21
499	Determination of beta-defensin genomic copy number in different populations: a comparison of three methods. <b>2011</b> , 6, e16768	37
498	A large expansion of the HSFY gene family in cattle shows dispersion across Yq and testis-specific expression. <b>2011</b> , 6, e17790	23
497	mRNA levels in control rat liver display strain-specific, hereditary, and AHR-dependent components. <b>2011</b> , 6, e18337	7
496	Copy number variants in patients with severe oligozoospermia and Sertoli-cell-only syndrome. <b>2011</b> , 6, e19426	105
495	A multi-sample based method for identifying common CNVs in normal human genomic structure using high-resolution aCGH data. <b>2011</b> , 6, e26975	3
494	Genome-wide mapping of copy number variation in humans: comparative analysis of high resolution array platforms. <b>2011</b> , 6, e27859	51
493	Accuracy in copy number calling by qPCR and PRT: a matter of DNA. <b>2011</b> , 6, e28910	26

492	Genome-wide association study of copy number variants suggests LTBP1 and FGD4 are important for alcohol drinking. <b>2012</b> , 7, e30860	12
491	Deletion and down-regulation of HRH4 gene in gastric carcinomas: a potential correlation with tumor progression. <b>2012</b> , 7, e31207	21
490	Genetic association studies of copy-number variation: should assignment of copy number states precede testing?. <b>2012</b> , 7, e34262	10
489	A preliminary study of copy number variation in Tibetans. <b>2012</b> , 7, e41768	22
488	Genome-wide association study identified CNP12587 region underlying height variation in Chinese females. <b>2012</b> , 7, e44292	1
487	Genome-wide identification of copy number variations in Chinese Holstein. <b>2012</b> , 7, e48732	41
486	A pilot study on collective effects of 22q13.31 deletions on gray matter concentration in schizophrenia. <b>2012</b> , 7, e52865	13
485	Widespread divergence of the CEACAM/PSG genes in vertebrates and humans suggests sensitivity to selection. <b>2013</b> , 8, e61701	21
484	Genotyping-by-sequencing (GBS): a novel, efficient and cost-effective genotyping method for cattle using next-generation sequencing. <b>2013</b> , 8, e62137	123
483	Genomic copy number variants: evidence for association with antibody response to anthrax vaccine adsorbed. <b>2013</b> , 8, e64813	5
482	Identification of genome-wide copy number variations among diverse pig breeds using SNP genotyping arrays. <b>2013</b> , 8, e68683	26
481	Genome-wide copy number variations inferred from SNP genotyping arrays using a Large White and Minzhu intercross population. <b>2013</b> , 8, e74879	16
480	Advantage of using allele-specific copy numbers when testing for association in regions with common copy number variants. <b>2013</b> , 8, e75350	5
479	Direct visualization of the highly polymorphic RNU2 locus in proximity to the BRCA1 gene. <b>2013</b> , 8, e76054	12
478	Concurrent gene signatures for han chinese breast cancers. <b>2013</b> , 8, e76421	40
477	A common copy number variation (CNV) polymorphism in the CNTNAP4 gene: association with aging in females. <b>2013</b> , 8, e79790	12
476	Structural variation-associated expression changes are paralleled by chromatin architecture modifications. <b>2013</b> , 8, e79973	23
475	CNVannotator: a comprehensive annotation server for copy number variation in the human genome. <b>2013</b> , 8, e80170	20

474	Genome-wide detection of copy number variations among diverse horse breeds by array CGH. <b>2014</b> , 9, e86860	23
473	Enhancing genome-wide copy number variation identification by high density array CGH using diverse resources of pig breeds. <b>2014</b> , 9, e87571	21
472	Genome-wide analysis of copy number variation identifies candidate gene loci associated with the progression of non-alcoholic fatty liver disease. <b>2014</b> , 9, e95604	27
471	HaplotypeCN: copy number haplotype inference with Hidden Markov Model and localized haplotype clustering. <b>2014</b> , 9, e96841	2
470	A comparison of assays for accurate copy number measurement of the low-affinity Fc gamma receptor genes FCGR3A and FCGR3B. <b>2015</b> , 10, e0116791	12
469	Global spectrum of copy number variations reveals genome organizational plasticity and proposes new migration routes. <b>2015</b> , 10, e0121846	4
468	The Porcine TSPY Gene Is Tricopy but Not a Copy Number Variant. <b>2015</b> , 10, e0131745	3
467	Genome-Wide Copy Number Variations Using SNP Genotyping in a Mixed Breed Swine Population. <b>2015</b> , 10, e0133529	10
466	CNV-CH: A Convex Hull Based Segmentation Approach to Detect Copy Number Variations (CNV) Using Next-Generation Sequencing Data. <b>2015</b> , 10, e0135895	5
465	Genomic Variants Revealed by Invariably Missing Genotypes in Nelore Cattle. <b>2015</b> , 10, e0136035	2
464	Microarray Analysis of Copy Number Variants on the Human Y Chromosome Reveals Novel and Frequent Duplications Overrepresented in Specific Haplogroups. <b>2015</b> , 10, e0137223	14
463	Identification of Copy Number Variations in Xiang and Kele Pigs. <b>2016</b> , 11, e0148565	13
462	Comprehensive Analysis of Genome Rearrangements in Eight Human Malignant Tumor Tissues. <b>2016</b> , 11, e0158995	6
461	A Segmental Copy Number Loss of the SFMBT1 Gene Is a Genetic Risk for Shunt-Responsive, Idiopathic Normal Pressure Hydrocephalus (iNPH): A Case-Control Study. <b>2016</b> , 11, e0166615	14
460	Heritable heading time variation in wheat lines with the same number of Ppd-B1 gene copies. <b>2017</b> , 12, e0183745	2
459	Whole-genome sequencing reveals mutational landscape underlying phenotypic differences between two widespread Chinese cattle breeds. <b>2017</b> , 12, e0183921	13
458	AMYCNE: Confident copy number assessment using whole genome sequencing data. <b>2018</b> , 13, e0189710	11
457	Personalized medicine: a transformative approach is needed. <b>2009</b> , 180, 911-3	6

456	Copy number variation (CNV) identification, interpretation, and database from Brazilian patients. <b>2020</b> , 43, e20190218	2
455	Integrated analysis of DNA methylation and mRNA expression profiles to identify key genes involved in the regrowth of clinically non-functioning pituitary adenoma. <b>2020</b> , 12, 2408-2427	6
454	Glyoxalase 1 copy number variation in patients with well differentiated gastro-entero-pancreatic neuroendocrine tumours (GEP-NET). <b>2017</b> , 8, 76961-76973	4
453	Identification of copy number alterations in colon cancer from analysis of amplicon-based next generation sequencing data. <b>2018</b> , 9, 20409-20425	5
452	Fibroblast growth factor receptor 2 expression, but not its genetic amplification, is associated with tumor growth and worse survival in esophagogastric junction adenocarcinoma. <b>2016</b> , 7, 19748-61	22
451	A Postgenomic Perspective on Molecular Cytogenetics. <b>2018</b> , 19, 227-239	18
450	NeuroArray, A Custom CGH Microarray to Decipher Copy Number Variants in Alzheimer's Disease. <b>2018</b> , 19, 499-504	2
449	Copy Number Variations in Adult-onset Neuropsychiatric Diseases. <b>2018</b> , 19, 420-430	18
448	Fcgbp - A Potential Viral Trap in RV144. <b>2014</b> , 8, 21-4	15
447	Genomewide Association Studies. <b>2008</b> , 225-238	3
446	Multicollinearity. <b>2010</b> , 836-843	1
445	Nature and nurture in neuropsychiatric genetics: where do we stand?. <b>2010</b> , 12, 7-23	35
444	Evaluation of Array Comparative genomic Hybridisation in prenatal diagnosis of fetal anomalies: a multicentre cohort study with cost analysis and assessment of patient, health professional and commissioner preferences for array comparative genomic hybridisation. <b>2017</b> , 4, 1-104	16
443	Shallow-depth sequencing of cell-free DNA for Hodgkin and diffuse large B-cell lymphoma (differential) diagnosis: a standardized approach with underappreciated potential. <b>2020</b> , Online ahead of print,	5
442	A genome-wide study of moyamoya-type cerebrovascular disease in the Korean population. <b>2011</b> , 50, 486-91	2
441	Genetic Profiling: Ethical Constraints upon Criminal Investigation Procedures. <b>2007</b> , 3, 236-252	2
440	On the spot: very local chromosomal rearrangements. <b>2012</b> , 4, 22	2
439	Mechanisms underlying copy number variation in F-box genes: evidence from comparison of 12 <i>Drosophila</i> species. <b>2011</b> , 19, 3-16	3

438	[Association of human microRNA related genetic variations with cancer]. <b>2011</b> , 33, 870-8	7
437	Amplifications of NCOA3 gene in colorectal cancers in a Chinese population. <b>2012</b> , 18, 855-60	4
436	Where are we today with personalized medicine?. <b>2012</b> , 50, 11-3	8
435	[High throughput genotyping for genomic cohort study]. <b>2007</b> , 40, 102-7	2
434	Identification and application of biomarkers in molecular and genomic epidemiologic research. <b>2009</b> , 42, 349-55	3
433	Recurrent benign copy number variants & issues in interpretation of variants of unknown significance identified by cytogenetic microarray in Indian patients with intellectual disability. <b>2015</b> , 142, 699-712	1
432	Identifying the risk of producing aneuploids using meiotic recombination genes as biomarkers: A copy number variation approach. <b>2017</b> , 145, 39-50	1
431	Copy number variations of five Y chromosome genes in donkeys. <b>2017</b> , 60, 391-397	1
430	Analysis of copy number variation in 8,842 Korean individuals reveals 39 genes associated with hepatic biomarkers AST and ALT. <b>2010</b> , 43, 547-53	8
429	A replication study of genome-wide CNV association for hepatic biomarkers identifies nine genes associated with liver function. <b>2011</b> , 44, 578-83	5
428	Effects of Copy Number Variations on Developmental Aspects of Children With Delayed Development. <b>2019</b> , 43, 215-223	3
427	Detection of copy number variation and selection signatures on the X chromosome in Chinese indigenous sheep with different types of tail. <b>2020</b> , 33, 1378-1386	6
426	Identification of copy number variations using high density whole-genome SNP markers in Chinese Dongxiang spotted pigs. <b>2019</b> , 1809-1815	1
425	Clinical Applications of Chromosomal Microarray Analysis. <b>2010</b> , 7, 111-118	6
424	Genome-wide Survey of Copy Number Variants Associated with Blood Pressure and Body Mass Index in a Korean Population. <b>2011</b> , 9, 152-160	2
423	Identifying Copy Number Variants under Selection in Geographically Structured Populations Based on F-statistics. <b>2012</b> , 10, 81-7	2
422	Effect of Combining Multiple CNV Defining Algorithms on the Reliability of CNV Calls from SNP Genotyping Data. <b>2012</b> , 10, 194-9	17
421	Embracing heterogeneity: coalescing the Tree of Life and the future of phylogenomics. <b>2019</b> , 7, e6399	63

- 420 Application of Restriction Site-Associated DNA Sequencing (RAD-Seq) for Copy Number Variation and Triploidy Detection in Human. **2021**, 161, 406-413
- 419 Genome-Wide Assessment Characteristics of Genes Overlapping Copy Number Variation Regions in Duroc Purebred Population. **2021**, 12, 753748 2
- 418 Genome-wide selective detection of Mile red-bone goat using next-generation sequencing technology. **2021**, 11, 14805-14812 0
- 417 Molecular Cytogenetics in the Era of Chromosomics and Cytogenomic Approaches. **2021**, 12, 720507 3
- 416 Human genome more variable than previously thought. *Nature*, 504
- 415 Chromosomal Microarray Analysis Is Replacing the Karyotype in the Evaluation of Neonates. **2007**, 2007, xix-xxii
- 414 Conceptual Evolution in Cancer Biology. **2008**, 185-208
- 413 Estimating the Relative Contributions of New Genes from Retrotransposition and Segmental Duplication Events during Mammalian Evolution. **2008**, 40-54
- 412 Microarray-basierte Analysen der genomischen Kopienzahl und der Genexpression beim Kolonkarzinom. **2008**, 23-25
- 411 Genetics and Psychiatry. **2008**, 853-883
- 410 Whole Genome Analysis by Array-Based Comparative Genomic Hybridization in Patients with Congenital Malformations. **2008**, 11, 1
- 409 Copy Number Variations in the Human Genome: Potential Source for Individual Diversity and Disease Association Studies. **2008**, 6, 1-7 6
- 408 Nucleotide Sequence Divergence between Humans and Chimpanzees.
- 407 Segmental Duplications and Their Role in the Evolution of the Human Genome. 1
- 406 Transposable Element-driven Duplications during Hominoid Genome Evolution.
- 405 Human-specific Changes of Genome Structure.
- 404 Human Genome Project: Importance in Clinical Genetics.
- 403 Development of KHapmap Browser using DAS for Korean HapMap Research. **2008**, 6, 57-63 1



402 Structural Diversity of the Human Genome and Disease Susceptibility.

401 Sequencing the Human Genome: Novel Insights into its Structure and Function.

400 Boundary Crossing Probability Computations in the Analysis of Scan Statistics. **2009**, 87-108

399 Basics of Personalized Medicine. **2009**, 1-27

2

398 Copy Number Variation and Human Health. **2009**, 108-119

397 Genome-Wide Association Studies and Genotyping Technologies. **2009**, 101-107

396 Array-Based Comparative Genomic Hybridization Application for Revealing Genomic Micro Imbalances in Congenital Malformations. **2009**, 12, 3-8

395 Genetics. **2009**, 49-55

394 Analysis of Population-Based Genetic Association Studies Applied to Cancer Susceptibility and Prognosis. **2009**, 149-191

393 DNA Sequencing for the Detection of Human Genome Variation and Polymorphism. **2009**, 88-100

392 Basic Genetics and Patterns of Inheritance. **2009**, 3-36

391 G72/G30 in Neuropsychiatric Disorders. **2009**, 91-106

390 A Linear-Time Algorithm for Analyzing Array CGH Data Using Log Ratio Triangulation. **2009**, 248-259

389 Cell Adhesion Molecules in Synaptopathies. **2009**, 141-158

388 [Relationships between copy number variations and human disease and its perspective in animal disease-resistant breeding]. **2008**, 30, 1385-91

387 Molecular Genetics of Autism.

386 Viselkedésgenetika a humán genom megfigyelésénél: kvantitatív és molekuláris genetikai alapok. **2009**, 29, 3-23

385 A CNV detection algorithm based on statistical analysis of the aligned reads. **2009**, 16D, 661-672

- 384 Comparison of Methods for Detecting and Quantifying Variation in Copy Numbers of Duplicated Genes. **2009**, 16, 1037-1046
- 383 Structural Variation in Great Ape Genomes.
- 382 Policy perspectives on the emerging pathways of personalized medicine. **2009**, 11, 377-87 7
- 381 Approaches to Identify Environmental and Epigenomic Components or Covariates of Cancer and Disease Susceptibility. **2010**, 197-219 2
- 380 Genetic Disorders. **2010**, 135-182
- 379 Sind wir polyploid?. **2010**, 247-267
- 378 12 Lichaamsmetabolisme. **2010**, 415-458
- 377 Human genetic variation and Parkinson's disease. **2010**, 3, 1-5
- 376 Molecular Biology. 1-22
- 375 Epilepsy with Complex Genetics. **2010**, 83-101
- 374 No Association between Copy Number Variation of the TCRB Gene and the Risk of Autism Spectrum Disorder in the Korean Population. **2010**, 8, 76-80
- 373 Genetic Variation Identified through Gene and Genome Sequencing. **2010**, 399-423
- 372 Multispot Array Technologies.
- 371 Prenatal Diagnosis by Microarray Analysis. 365-379
- 370 Diagnostic Genome Profiling in Mental Retardation. **2011**, 177-194
- 369 Atherosclerosis: Pathogenesis, Genetics and Experimental Models. 1
- 368 Nervous System Aging, Degeneration, and the p53 Family. **2011**, 83-93
- 367 Genetic Studies of Schizophrenia. **2011**, 333-380

366 Genetics of Affective Disorders. **2010**, 460-463

365 Cytogenetics of Primary Skin Tumors. **2011**, 57-72

364 Single Nucleotide Polymorphism and its Application in Mapping Loci Involved in Developing Human Diseases and Traits. **2011**, 113-127

363 Molecular Diagnosis of Hematopoietic Neoplasms. **2011**, 1415-1440

362 Cytogenetic Analysis and Related Techniques in Hematopathology. **2011**, 81-94

1

361 Principles of Molecular Biology. **2011**, 13-25

360 Characterising Structural Variation by Means of Next-Generation Sequencing.

359 Genome-Wide Association Study between Copy Number Variation and Trans-Gene Expression by Protein-Protein Interaction-Network. **2011**, 18D, 89-100

358 Congenital Anomalies. 162-184

357 Facultativeness Principle and Generalized Approach to the Genome and Hereditary Variability. **2012**, 259-268

356 Comparative Genomic Hybridization in the Study of Human Disease.

355 Gene Duplication and Functional Consequences. **2012**, 139-156

354 Principles of Molecular Biology. **2012**, 1209-1223

0

353 Cytogenetic Testing and Chromosomal Disorders. **2012**, 39-59

352 Exploring correlations among copy number variants. **2012**, 02, 131-135

351 The Microarray Paradigm and Its Various Implementations. **2012**, 1-8

350 Genomes and Nucleic Acid Alterations. **2012**, 1225-1229

349 Array-CGH and SNP-Arrays, the New Karyotype. **2012**, 39-52

348 Pharmacogenomics Principles: Introduction to Personalized Medicine. 1-14

347 Molecular genetics. **2012**, 222-233

346 Germline Copy Number Variation and Cancer Risk.

345 Single Nucleotide Polymorphism and its Application in Mapping Loci Involved in Developing Human Diseases and Traits. **2012**, 3, 61-75

344 The Relationship Between DNA-Repair Genes, Cellular Radiosensitivity, and the Response of Tumors and Normal Tissues to Radiotherapy. **2013**, 75-128

343 Recurrent CNVs in the Etiology of Epigenetic Neurodevelopmental Disorders. **2013**, 147-178

342 Human Genome. **2013**, 249-262

341 Untersuchung der Funktion von DNA-Sequenzen. **2013**, 211-268

340 Topics in Computational Genomics. **2013**, 69-100

339 Mapping Prostate Cancer Aggressiveness Loci. **2013**, 195-200

338 Mismeasuring Man Thirty Years Later. **2013**, 129-146

2

337 Determination of Breast Cancer Dormancy: Analysis of Circulating Free DNA Using SNP 6.0 Arrays. **2013**, 35-50

336 Pharmacogenomics of Pulmonary and Respiratory Diseases. **2013**, 507-527

335 Chromosome Microarrays. **2014**, 149-165

334 Genetics of Childhood Obesity. **2014**, 71-91

1

333 References. **2014**, 801-954

332 Genetics of Childhood Obesity. **2013**, 1-21

331 Omics Technologies Applied in Breast Cancer Research. **2014**, 3-15

330 Situations spécifiques. **2014**, 171-242

329 Autism and Schizophrenia Are Disorders of Evolvability. **2014**, 03, 161-183

1

328 [Cardiovascular pharmacogenomics]. **2014**, 84, 25-31

1

327 Genetic Copy Number Variations in Colon Mucosa Indicating Risk for Colorectal Cancer. **2014**, 05, 1354-1361

326 Detecting Copy Number Changes and Structural Rearrangements Using DNA Sequencing. **2014**, 355-378

1

325 Latest findings in the genetic architecture of schizophrenia: The contribution of genetic studies among Afrikaners. **2014**, 33,

324 Human Genetics of Diabetic Cardiovascular Complications. **2014**, 1,

323 Detection of Copy Number Variations from Array Comparative Genomic Hybridization Data Using Linear-chain Conditional Random Field Models. **2014**, 409-428

322 Molecular Diagnostics and Tumor Mutational Analysis. **2015**, 47-65

1

321 The Genetics of Specific Language Impairment (SLI). **2015**, 7-34

320 Genetics of Tourette Syndrome. **2015**, 169-189

1

319 Genetic Testing: An Industrial Perspective. **2015**, 451-462

318 Copy Number Variation in Human Health, Disease and Evolution. **2015**, 129-154

3

317 Basic Aspects. **2015**, 1-33

316 Overview of Next Generation, High-Throughput Molecular Genetic Methods. **2015**, 1-16

315 Effect of MPG gene rs2858056 polymorphism, copy number variation, and level of serum MPG protein on the risk for rheumatoid arthritis. **2015**, 10, e0120699

3

314 The Mutational Spectrum of Neurodevelopmental Disorders. 49-68

313 Translational Research Methods: Basics of Renal Molecular Biology. **2016**, 425-445

312 Prenatal Diagnosis by Microarray Analysis. 366-379

311 General aspects of aetiology, diagnostics and therapy. **2016**, 3-104

310 Copy number variation in the cattle genome. **2016**, 44, 59-68

309 22q11.2 Deletion Syndrome: A Paradigmatic Copy-Number-Variant (CNV) Disorder. **2016**, 723-730

308 Introduction to Molecular Genetics. **2016**, 3-24

307 Copy number variants in the sheep genome detected using multiple approaches.

306 Sequencing the Human Genome: Novel Insights into Its Structure and Function. 1-9

305 CrowdVariant: a crowdsourcing approach to classify copy number variants. 1

304 A computational method for detection of structural variants using Deviant Reads and read pair Orientation: DevRO.

303 Genetics and Genomic Basis of Sleep Disorders in Humans. **2017**, 322-339.e7 0

302 Spontaneous mutations and transmission distortions of genic copy number variants shape the standing genetic variation in *Picea glauca*.

301 ?????Interleukin-18 ??????. **2017**, 51, 17-24

300 Comparison of village dog and wolf genomes highlights the pivotal role of the neural crest in dog domestication. 2

299 Worldwide genetic variation of the IGHV and TRBV immune receptor gene families in humans. 2

298 Integrative DNA copy number detection and genotyping from sequencing and array-based platforms. 0

297 *Drosophila* larval brain neoplasms present tumour-type dependent genome instability.

296 KoVariome: Korean National Standard Reference Variome database of whole genomes with comprehensive SNV, indel, CNV, and SV analyses.

295 Global characterization of copy number variants in epilepsy patients from whole genome sequencing. 0

- 294 An Integrated Method of Detecting Copy Number Variation Based on Sequence Assembly. **2018**, 589-594
- 293 IsoCon: Deciphering highly similar multigene family transcripts from Iso-Seq data.
- 292 Human Genome. **2018**, 273-289
- 291 SV-plaudit: A cloud-based framework for manually curating thousands of structural variants. 1
- 290 Apport des puces  $\square$ ADN et nouveaux syndromes microdéliionnels. **2018**, 202, 693-705
- 289 CNV Neurons Are Rare in Aged Human Neocortex. 1
- 288 High coverage genome sequencing and identification of genomic variants in Bengal tiger (*Panthera tigris tigris*). 0
- 287 6q21q22.2 Deletion Syndrome with Ataxia and Congenital Ocular Motor Apraxia (Cogan's Syndrome). 4-13
- 286 Discovery of tandem and interspersed segmental duplications using high throughput sequencing. 1
- 285 A machine-learning approach for accurate detection of copy-number variants from exome sequencing.
- 284 svtools: population-scale analysis of structural variation. 0
- 283 SVIM: Structural Variant Identification using Mapped Long Reads. 3
- 282 Genetic Control of Fetal Sex Development. **2019**, 454-467
- 281 Simultaneous detection of target CNVs and SNVs of thalassemia by multiplex PCR and next-generation sequencing. **2019**, 19, 2837-2848 4
- 280 Integrating detection of copy neutral chromosomal losses in a clinical setting in leukemia and lymphoma by means of allelic imbalance and read depth ratio comparison.
- 279 Karyotypic divergence confounds cellular phenotypes in large pharmacogenomic studies.
- 278 A Simple Deep Learning Approach for Detecting Duplications and Deletions in Next-Generation Sequencing Data.
- 277 Copy number variants outperform SNPs to reveal genotype-temperature association in a marine species. 3

- 276 Thermal adaptation rather than demographic history drives genetic structure inferred by copy number variants in a marine fish.
- 275 Prenatal Diagnosis of Partial Trisomy 6q and Partial Monosomy 18p Associated with Cephalocele: A Case Report. **2020**, 23, 99-102
- 274 Similarity-based analysis of allele frequency distribution among multiple populations identifies adaptive genomic structural variants. **2021**, 1
- 273 Encyclopedia of Behavioral Medicine. **2020**, 555-555
- 272 Pervasive cis effects of variation in copy number of large tandem repeats on local epigenetics and gene expression.
- 271 Genome-wide Copy Number Variations in a Large Cohort of Bantu African Children.
- 270 Gene Copy Number Variation Does Not Reflect Structure or Environmental Selection in Two Recently Diverged California Populations of. **2020**, 10, 4591-4597 0
- 269 Giftedness, Talent, and Human Evolution: A Framework for Understanding Extreme Behavior. **2021**, 281-294 0
- 268 A proteogenomic approach to target neoantigens in solid tumors. **2020**, 17, 797-812 1
- 267 Data-driven prioritization and preclinical evaluation of therapeutic targets in glioblastoma. **2020**, 2, vdaa151 0
- 266 Viral Symbiosis in the Origins and Evolution of Life with a Particular Focus on the Placental Mammals. **2020**, 69, 3-24 0
- 265 The genomic basis of medicine. **2020**, 218-235
- 264 Whole genome resequencing of the Iranian native dogs and wolves to unravel variome during dog domestication.
- 263 Introduction to Molecular Genetics. **2020**, 3-26
- 262 Whole genome resequencing of the Iranian native dogs and wolves to unravel variome during dog domestication.
- 261 Detection of structural variations in densely-labelled optical DNA barcodes: A hidden Markov model approach. **2021**, 16, e0259670
- 260 lhybCNV: An intra-hybrid approach for CNV detection from next-generation sequencing data. **2021**, 121, 103304 2
- 259 Combining callers improves the detection of copy number variants from whole-genome sequencing. **2021**, 0



258	Diverse environmental perturbations reveal the evolution and context-dependency of genetic effects on gene expression levels.	0
257	The drifting human genome. <b>2007</b> , 104, 20147-8	7
256	Incipient Events in Human Carcinogenesis: A Concept of Forerunner Genes. <b>2008</b> , 125-146	
255	Algorithms for Computing the Length-Constrained Max-Score Segments with Applications to DNA Copy Number Data Analysis. <b>2007</b> , 834-845	
254	Post-Hybridization Quality Measures for Oligos in Genome-Wide Microarray Experiments. <b>2008</b> , 64-75	
253	The Microarray Paradigm and Its Various Implementations. <b>2012</b> , 1-8	0
252	Array-CGH and SNP-Arrays, the New Karyotype. <b>2012</b> , 39-52	0
251	Tunability enhancement of gene regulatory motifs through competition for regulatory protein resources.	
250	The haplolethality paradox of the wup gene in Drosophila.	
249	Data_Sheet_1.pdf. <b>2019</b> ,	1
248	A new wave in the genetics of psychiatric disorders: the copy number variant tsunami. <b>2009</b> , 34, 55-9	8
247	Whole genome association studies of neuropsychiatric disease: An emerging era of collaborative genetic discovery. <b>2007</b> , 3, 613-8	2
246	Tumor genome wide DNA alterations assessed by array CGH in patients with poor and excellent survival following operation for colorectal cancer. <b>2007</b> , 3, 341-55	7
245	Comparison of comparative genomic hybridization technologies across microarray platforms. <b>2009</b> , 20, 135-51	25
244	MLGA: a cost-effective approach to the diagnosis of gene deletions in eye development anomalies. <b>2009</b> , 15, 1445-8	3
243	The Barker hypothesis: how pediatricians will diagnose and prevent common adult-onset diseases. <b>2009</b> , 120, 199-207	38
242	Molecular genetics of Psoriasis (Principles, technology, gene location, genetic polymorphism and gene expression). <b>2010</b> , 4, 103-27	3
241	Cell-based Models for Discovery of Pharmacogenomic Markers of Anticancer Agent Toxicity. <b>2008</b> , 4, 1-13	6

240	Copy number variation in the complement factor H-related genes and age-related macular degeneration. <b>2011</b> , 17, 2080-92	24
239	Using the R Package crlmm for Genotyping and Copy Number Estimation. <b>2011</b> , 40, 1-32	445
238	Genomes, populations and diseases: ethnic genomics and personalized medicine. <b>2010</b> , 2, 15-30	4
237	Variation in CCL3L1 copy number in rhesus macaques ( <i>Macaca mulatta</i> ). <b>2012</b> , 62, 218-24	1
236	Detecting highly differentiated copy-number variants from pooled population sequencing. <b>2013</b> , 344-55	6
235	Genetic variation and its role in malignancy. <b>2011</b> , 7, 158-71	10
234	Genetic effects of a 13q31.1 microdeletion detected by noninvasive prenatal testing (NIPT). <b>2014</b> , 7, 7003-11	7
233	Copy number variation of the Lipoprotein(a) (LPA) gene is associated with coronary artery disease in a southern Han Chinese population. <b>2014</b> , 7, 3669-77	10
232	Genetics of Vascular Dementia. <b>2010</b> , 51, 9-25	1
231	Whole-exome sequencing and whole genome re-sequencing for prenatal diagnosis of achondroplasia. <b>2015</b> , 8, 19241-9	4
230	Whole Genome Sequencing as a Genetic Test for Autism Spectrum Disorder: From Bench to Bedside and then Back Again. <b>2016</b> , 25, 116-21	8
229	Copy Number Variation Analysis of IL22 and LCE3C in Different Subtypes of Psoriasis in a Chinese Han Population. <b>2021</b> , 27, e934927	0
228	Whole-genome analysis of a putative rare and complex interchromosomal reciprocal insertion: thorough investigations for a straightforward interpretation.. <b>2021</b> ,	0
227	High Olfactory Receptor-Rich 11q11 Copy Number in Girls and African American Children.. <b>2021</b> , 12,	
226	Association of X Chromosome Aberrations with Male Infertility. <b>2021</b> , 48, 69-72	
225	BreakNet: detecting deletions using long reads and a deep learning approach. <b>2021</b> , 22, 577	0
224	Lung Cancer Subtype Diagnosis by Fusing Image-genomics Data and Hybrid Deep Networks. <b>2021</b> , PP,	0
223	Modeling dopamine dysfunction in autism spectrum disorder: From invertebrates to vertebrates.. <b>2021</b> , 133, 104494	0

222	The therapeutical approaches for rare diseases through the immune processes of IgG Fc Receptors. <b>2020</b> , 070-071	
221	Pediatric Genomic Medicine. <b>2021</b> ,	
220	Multi-view manifold regularized compact low-rank representation for cancer samples clustering on multi-omics data.. <b>2022</b> , 22, 334	
219	An Update Evolving View of Copy Number Variations in Autoimmune Diseases.. <b>2021</b> , 12, 794348	1
218	Comprehensive Pan-Cancer Analyses of Pyroptosis-Related Genes to Predict Survival and Immunotherapeutic Outcome.. <b>2022</b> , 14,	1
217	Recombination, selection and the evolution of tandem gene arrays.	0
216	Identification of Copy Number Variations and Genetic Diversity in Italian Insular Sheep Breeds.. <b>2022</b> , 12,	1
215	ifCNV: a novel isolation-forest-based package to detect copy number variations from various NGS datasets.	
214	Duck Genomics and Biotechnology. <b>2022</b> , 581-615	
213	Genetic Alteration, Prognostic and Immunological Role of Acyl-CoA Synthetase Long-Chain Family Member 4 in a Pan-Cancer Analysis.. <b>2022</b> , 13, 812674	0
212	Role of Genetic Variants and Host Polymorphisms on COVID-19: From Viral Entrance Mechanisms to Immunological Reactions.. <b>2022</b> ,	1
211	Analysis of N6-Methyladenosine Modification Patterns and Tumor Immune Microenvironment in Pancreatic Adenocarcinoma.. <b>2021</b> , 12, 752025	0
210	Editorial: Scalable Bioinformatics: Methods, Software Tools, and Hardware Architectures.. <b>2021</b> , 12, 822986	0
209	CYP2C8, CYP2C9 and CYP2C19 characterization using Next Generation Sequencing and Haplotype Analysis: A GeT-RM Collaborative Project.. <b>2022</b> ,	2
208	Resolving single-cell copy number profiling for large datasets.	
207	Improving Jitter Distribution in the Breakpoints of Genome CNVs. <b>2021</b> , 1, 23-30	
206	Metabolic pathway-based target therapy to hepatocellular carcinoma: a computational approach. <b>2022</b> , 83-103	
205	The Concept of Immunogenetics.. <b>2022</b> , 1367, 1-17	

204	Genomic characterization of the world's longest selection experiment in mouse reveals the complexity of polygenic traits.. <b>2022</b> , 20, 52	1
203	Progress in Methods for Copy Number Variation Profiling.. <b>2022</b> , 23,	1
202	Genomic variations and epigenomic landscape of the Medaka Inbred Kiyosu-Karlsruhe (MIKK) panel.. <b>2022</b> , 23, 58	1
201	The Psychoemotional Stress-Induced Changes in the Abundance of SatIII (1q12) and Telomere Repeats, but Not Ribosomal DNA, in Human Leukocytes.. <b>2022</b> , 13,	0
200	Emerging Glycation-Based Therapeutics-Glyoxalase 1 Inducers and Glyoxalase 1 Inhibitors.. <b>2022</b> , 23,	4
199	Assessment of linkage disequilibrium patterns between structural variants and single nucleotide polymorphisms in three commercial chicken populations.. <b>2022</b> , 23, 193	1
198	Structural deviations of the posterior fossa and the cerebellum and their cognitive links in a neurodevelopmental deletion syndrome.	0
197	Population Genomic Sequencing Delineates Global Landscape of Copy Number Variations that Drive Domestication and Breed Formation of in Chicken.. <b>2022</b> , 13, 830393	1
196	Dynamic evolution of recently duplicated genes in <i>Caenorhabditis elegans</i> .	0
195	CRIA: An Interactive Gene Selection Algorithm for Cancers Prediction Based on Copy Number Variations.. <b>2022</b> , 13, 839044	1
194	Copy Number Variation Analysis of Euploid Pregnancy Loss.. <b>2022</b> , 13, 766492	0
193	Towards the detection of copy number variation from single sperm sequencing in cattle.. <b>2022</b> , 23, 215	
192	Sparse modelling of cancer patients' survival based on genomic copy number alterations.. <b>2022</b> , 128, 104025	
191	Copy number alteration signatures as biomarkers in cancer: a review.. <b>2022</b> ,	0
190	Detection of copy number variations from NGS data by using an adaptive kernel density estimation-based outlier factor. <b>2022</b> , 126, 103524	
189	Dosage sensitivity and exon shuffling shape the landscape of polymorphic duplicates in <i>Drosophila</i> and humans.. <b>2021</b> ,	0
188	Fine-Mapping of the Major Histocompatibility Complex Region Linked to Leprosy in Northern China.. <b>2021</b> , 12, 768259	0
187	Haplotype-resolved inversion landscape reveals hotspots of mutational recurrence associated with genomic disorders.	2

186 Copy number variation of the putative speciation genes in the European house mouse hybrid zone.

185 VarGenius-HZD Allows Accurate Detection of Rare Homozygous or Hemizygous Deletions in Targeted Sequencing Leveraging Breadth of Coverage.. **2021**, 12, 0

184 KNNCNV: A K-Nearest Neighbor Based Method for Detection of Copy Number Variations Using NGS Data.. **2021**, 9, 796249 1

183 Social behavior in 16p11.2 and 22q11.2 copy number variations: Insights from mice and humans. **2021**, e12787 0

182 Rare dosage abnormalities flanking the SHOX gene. **2021**, 22,

181 Two Different Copy Number Variations of the Gene in Chinese Cattle and Their Association with Growth Traits.. **2021**, 12, 1

180 CNVs with adaptive potential in : genome architecture and new annotated assembly.. **2022**, 5, 0

179 Data\_Sheet\_1.ZIP. **2020**,

178 Data\_Sheet\_1.PDF. **2020**,

177 Table\_1.XLSX. **2018**,

176 Table\_2.XLSX. **2018**,

175 Table\_3.XLSX. **2018**,

174 Table\_4.XLSX. **2018**,

173 Table\_5.XLSX. **2018**,

172 Table\_6.XLSX. **2018**,

171 Table\_7.XLSX. **2018**,

170 Table\_8.XLSX. **2018**,

169 Table\_9.XLSX. **2018**,

168 Data\_Sheet\_1.XLSX. 2018,

167 Data\_Sheet\_2.DOCX. 2018,

166 Data\_Sheet\_3.XLSX. 2018,

165 Data\_Sheet\_4.XLSX. 2018,

164 Data\_Sheet\_5.XLSX. 2018,

163 Data\_Sheet\_6.XLSX. 2018,

162 Data\_Sheet\_7.XLSX. 2018,

161 Data\_Sheet\_8.XLSX. 2018,

160 Data\_Sheet\_9.XLSX. 2018,

159 DataSheet\_1.docx. 2020,

158 Table\_1.xls. 2020,

157 Table\_10.xls. 2020,

156 Table\_11.xls. 2020,

155 Table\_2.xls. 2020,

154 Table\_3.xls. 2020,

153 Table\_4.xls. 2020,

152 Table\_5.xls. 2020,

151 Table\_6.xls. 2020,

150 Table\_7.xls. 2020,

149 Table\_8.xls. 2020,

148 Table\_9.xls. 2020,

147 Table\_1.xlsx. 2019,

146 Table\_2.xlsx. 2019,

145 Table\_3.xlsx. 2019,

144 Table\_4.xlsx. 2019,

143 Table\_5.xlsx. 2019,

142 Table\_6.xlsx. 2019,

141 Table\_7.xlsx. 2019,

140 Table\_8.xlsx. 2019,

139 Table\_1.XLSX. 2019,

138 Table\_10.XLSX. 2019,

137 Table\_11.XLSX. 2019,

136 Table\_12.XLSX. 2019,

135 Table\_13.XLSX. 2019,

134 Table\_14.XLSX. 2019,

133 Table\_15.XLSX. 2019,

132 Table\_2.XLSX. 2019,

131 Table\_3.XLSX. 2019,

130 Table\_4.XLSX. 2019,

129 Table\_5.XLSX. 2019,

128 Table\_6.XLSX. 2019,

127 Table\_7.XLSX. 2019,

126 Table\_8.XLSX. 2019,

125 Table\_9.XLSX. 2019,

124 Image\_1.jpeg. 2019,

123 Image\_2.jpeg. 2019,

122 Table\_1.xlsx. 2019,

121 Table\_2.xlsx. 2019,

120 Table\_3.xlsx. 2019,

119 Table\_4.xlsx. 2019,

118 Table\_5.xlsx. 2019,

117 Table\_6.xlsx. 2019,

116 Table\_1.DOCX. 2020,

115 Table\_1.docx. 2020,



114	DataSheet_1.xlsx. <b>2020,</b>		
113	Table_1.XLS. <b>2019,</b>		
112	Table_2.XLS. <b>2019,</b>		
111	Table_3.xls. <b>2019,</b>		
110	DataSheet_1.zip. <b>2020,</b>		
109	DataSheet_2.docx. <b>2020,</b>		
108	Table_1.xlsx. <b>2020,</b>		
107	Recombination, selection and the evolution of tandem gene arrays.. <b>2022,</b>		
106	Recurrent inversion polymorphisms in humans associate with genetic instability and genomic disorders.. <b>2022,</b>		2
105	Genetic and chemotherapeutic influences on germline hypermutation.. <i>Nature</i> , <b>2022</b> , 605, 503-508	50.4	0
104	Combining nucleotide variations and structure variations for improving astaxanthin biosynthesis.. <b>2022</b> , 21, 79		
103	Breast Cancer and Next-Generation Sequencing: Towards Clinical Relevance and Future. <b>2022</b> , 477-518		
102	Directed yeast genome evolution by controlled introduction of trans-chromosomal structural variations.		1
101	DNA sequence features underlying large-scale duplications and deletions in human.. <b>2022,</b>		
100	Copy Number Variant (CNV). <b>2022</b> , 1714-1717		
99	The evolving role of diagnostic genomics in kidney transplantation. <b>2022,</b>		0
98	Genome-wide identification of copy number variation and association with fat deposition in thin and fat-tailed sheep breeds. <b>2022</b> , 12,		0
97	Genome-Wide Association Studies, Runs of Homozygosity Analysis, and Copy Number Variation Detection to Identify Reproduction-Related Genes in Bama Xiang Pigs. 9,		0

- 96 Identification of Three Prognosis-Related Differentially Expressed lncRNAs Driven by Copy Number Variation in Thyroid Cancer. **2022**, 2022, 1-18 ○
- 95 Copy Number Variation (CNV): A New Genomic Insight in Horses. **2022**, 12, 1435 ○
- 94 Targeted copy number variant identification across the neurodegenerative disease spectrum. ○
- 93 Copy Number Analyses Identified a Novel Gene: APOBEC3A Related to Lipid Metabolism in the Pathogenesis of Preeclampsia. 9, ○
- 92 Human Leukocyte Antigen Fine-Mapping and Correlation Analysis of Han and Minority Leprosy Patients in Southern China. 13, ○
- 91 WAVECNV: A New Approach for Detecting Copy Number Variation by Wavelet Clustering. **2022**, 10, 2151 ○
- 90 Genomic imbalance in euploid pregnancy loss. ○
- 89 Behavioral genetics and animal science. **2022**, 1-47 1
- 88 Genome-Wide Detection of Copy Number Variations and Evaluation of Candidate Copy Number Polymorphism Genes Associated With Complex Traits of Pigs. 9, 1
- 87 Resolving single-cell copy number profiling for large datasets. **2022**, 23, ○
- 86 Role of the Glyoxalase System in Breast Cancer and Gynecological Cancer-Implications for Therapeutic Intervention: a Review. 12, 1
- 85 Not functional yet a difference maker: junk DNA as a case study. **2022**, 37, ○
- 84 Down syndrome: a model for chromosome abnormalities. **2023**, 45-68 ○
- 83 Potentials and challenges of chromosomal microarray analysis in prenatal diagnosis. 13, 1
- 82 Genome-wide evaluation of copy gain and loss variations in three Afghan sheep breeds. **2022**, 12, ○
- 81 CNest: A novel copy number association discovery method uncovers 862 new associations from 200,629 whole-exome sequence datasets in the UK Biobank. **2022**, 2, 100167 ○
- 80 Assembly of a pangenome for global cattle reveals missing sequences and novel structural variations, providing new insights into their diversity and evolutionary history. **2022**, 32, 1585-1601 ○
- 79 Genome-wide identification, evolutionary analysis, and antimicrobial activity prediction of CC chemokines in allotetraploid common carp, *Cyprinus carpio*. **2022**, 130, 114-131 ○

78	Evolving Approaches to Identifying Genetic Risk Variants for Sleep Disorders. <b>2022</b> , 3-20	0
77	The genetic basis of Gilles de la Tourette syndrome. <b>2022</b> , 3-38	0
76	Introduction of medical genomics and clinical informatics integration for p-Health care. <b>2022</b> , 1-37	1
75	Gene-dosage imbalance due to trisomic HSA21 and genotype-phenotype association in Down syndrome. <b>2022</b> , 93-134	0
74	A second unveiling: haplotig masking of the eastern oyster genome improves population-level inference.	0
73	An evolutionary look into the history of lentil reveals unexpected diversity. <b>2022</b> , 15, 1313-1325	0
72	CNVs Associated with Different Clinical Phenotypes of Psoriasis and Anti-TNF-Induced Palmoplantar Pustulosis. <b>2022</b> , 12, 1452	0
71	PEcnv: accurate and efficient detection of copy number variations of various lengths. <b>2022</b> , 23,	1
70	Genome-Wide Copy Number Variant and High-Throughput Transcriptomics Analyses of Placental Tissues Underscore Persisting Child Susceptibility in At-Risk Pregnancies Cleared in Standard Genetic Testing. <b>2022</b> , 23, 11448	1
69	Genome-wide detection of copy number variation in American mink using whole-genome sequencing. <b>2022</b> , 23,	0
68	Copy number variations of LRRFIP1 gene and the relationship with growth traits in four Chinese sheep. 1-8	0
67	Genome-wide analysis of CNVs in three populations of Tibetan sheep using whole-genome resequencing. 13,	0
66	The accuracy and feasibility of noninvasive prenatal testing in a consecutive series of 20,626 pregnancies with different clinical characteristics.	1
65	Nucleotide and structural polymorphisms of the eastern oyster genome paint a mosaic of divergence, selection, and human impacts.	0
64	Copy number variation of EIF4A2 loci related to phenotypic traits in Chinese cattle. <b>2022</b> , 8, 2147-2156	0
63	Smart breeding approaches in post-genomics era for developing climate-resilient food crops. 13,	0
62	Investigation of Copy Number Variation in South African Patients With Congenital Heart Defects.	0
61	ifCNV: A novel isolation-forest-based package to detect copy-number variations from various targeted NGS datasets. <b>2022</b> , 30, 174-183	1

- 60 Genetic Susceptibility in Tourette Syndrome. **2022**, 125-136 ○
- 59 Benign-Ex: Delineating Regions of the Human Genome Benign to Copy Number Variation. ○
- 58 Genomic Disorders in Chronic Kidney Disease Across the Lifespan. ASN.2022060725 ○
- 57 Genetic variation in placental insufficiency: What have we learned over time?. 10, ○
- 56 Copy Number Variants Are Ovarian Cancer Risk Alleles at Known and Novel Risk Loci. ○
- 55 A new framework for detecting copy number variants from single nucleotide polymorphism data: CNVq a versatile R package for paralogs and CNVs detection. ○
- 54 j5p13 microduplication in a malformed fetus and his unaffected father. ○
- 53 Identification of prognostic factors in classic Hodgkin lymphoma by integrating whole slide imaging and next generation sequencing. ○
- 52 Eugenics and the Theory of Inheritability. **2022**, 59-85 ○
- 51 Genetics of Schizophrenia Spectrum Disorders: Looking Back and Peering Ahead. **2009**, 38, 436-439 ○
- 50 Copy Number Variation among Resistance Genes Analogues in Brassica napus. **2022**, 13, 2037 ○
- 49 Genome Organization and Copy-Number Variation Reveal Clues to Virulence Evolution in Coccidioides posadasii. **2022**, 8, 1235 ○
- 48 Association of Fc Gamma Receptor 3B Gene Copy Number Variation with Rheumatoid Arthritis Susceptibility. **2022**, 13, 2238 ○
- 47 CNV-PCC: An efficient method for detecting copy number variations from next-generation sequencing data. 10, ○
- 46 Identification of Copy Number Variations in Four Horse Breed Populations in South Korea. **2022**, 12, 3501 ○
- 45 Comprehensive Analysis of Copy Number Variations on Glycoside Hydrolase 45 Genes among Different Bursaphelenchus xylophilus Strains. **2022**, 23, 15323 ○
- 44 Inferencing Bulk Tumor and Single-Cell Multi-Omics Regulatory Networks for Discovery of Biomarkers and Therapeutic Targets. **2023**, 12, 101 ○
- 43 Analysis of Copy Number Variation in the Whole Genome of Normal-Haired and Long-Haired Tianzhu White Yaks. **2022**, 13, 2405 ○

- 42 Association of Inherited Copy Number Variation in ADAM3A and ADAM5 Pseudogenes with Oropharynx Cancer Risk and Outcome. **2022**, 13, 2408 ○
- 41 Comparative epigenomics reveals the impact of ruminant-specific regulatory elements on complex traits. **2022**, 20, ○
- 40 Remnants of SIRE1 retrotransposons in human genome?. **2023**, 102, ○
- 39 Deepening the understanding of CNVs on chromosome 15q11-13 by using hiPSCs: An overview. 10, ○
- 38 ParseCNV2: a versatile and integrated tool for copy number variation association studies. ○
- 37 Comparative genomic diversity analysis of copy number variations (CNV) in indicine and taurine cattle thriving in Europe and Indian subcontinent. 1-12 ○
- 36 Deciphering the exact breakpoints of structural variations using long sequencing reads with DeBreak. **2023**, 14, ○
- 35 Understanding the Contribution of Lactate Metabolism in Cancer Progress: A Perspective from Isomers. **2023**, 15, 87 ○
- 34 Evolutionary dynamics of glue gene copy number in Drosophila species. ○
- 33 Multiallelic Copy Number Variation in ORM1 is Associated with Plasma Cell-Free DNA Levels as an Intermediate Phenotype for Venous Thromboembolism. ○
- 32 Effects of Noise and Light. **2023**, 357-378 10
- 31 Population differentiated copy number variation between Eurasian wild boar and domesticated pig populations. **2023**, 13, ○
- 30 Exosome-Associated Gene Signature for Predicting the Prognosis of Ovarian Cancer Patients. **2023**, 2023, 1-17 ○
- 29 Copy number variations on chromosome 2: impact on human phenotype, a cross-sectional study. **2023**, 8, e198 ○
- 28 Reduction, evolutionary pattern and positive selection of genes encoding formate dehydrogenase in WoodWjungdahl pathway of gastrointestinal acetogens suggests their adaptation to formate-rich habitats. **2023**, 15, 129-141 ○
- 27 Copy number variations in a Chinese series of patients with DiGeorge syndrome-related hypoparathyroidism. ○
- 26 Correlation of mutated gene and signalling pathways in ASD. **2023**, 14, 384-392 ○
- 25 A comparison of algorithms for identifying copy number variants in family-based whole-exome sequencing data and its implications in inheritance pattern analysis. **2023**, 861, 147237 ○

- 24 Assessment of copy number variants in three Brazilian locally adapted cattle breeds using whole-genome re-sequencing data. ○
- 23 Detection rates of abnormalities in over 10,000 amniotic fluid samples at a single laboratory. **2023**, 23, ○
- 22 Copy Number Variation Analysis Revealed the Evolutionary Difference between Chinese Indigenous Pigs and Asian Wild Boars. **2023**, 14, 472 ○
- 21 Genomic structural variation: A complex but important driver of human evolution. ○
- 20 Copy Number Variation and Osteoporosis. **2023**, 21, 167-172 ○
- 19 The idiosyncratic genome of Korean long-tailed chicken as a valuable genetic resource. **2023**, 26, 106236 ○
- 18 Xeno-free generation of human induced pluripotent stem cells from donor-matched fibroblasts isolated from dermal and oral tissues. ○
- 17 Genetic evaluation of living kidney donor candidates: a review and recommendations for best practices. **2023**, ○
- 16 Genome-Wide Detection and Analysis of Copy Number Variation in Anhui Indigenous and Western Commercial Pig Breeds Using Porcine 80K SNP BeadChip. **2023**, 14, 654 ○
- 15 Molecular Cytogenetics in Domestic Bovids: A Review. **2023**, 13, 944 ○
- 14 Genetic mechanisms in generalized epilepsies. **2023**, 5, ○
- 13 Optical Genome Mapping for Cytogenetic Diagnostics in AML. **2023**, 15, 1684 ○
- 12 Recommendations, guidelines, and best practice for the use of human induced pluripotent stem cells for neuropharmacological studies of neuropsychiatric disorders. **2023**, 2, 101125 ○
- 11 Genetics and Kidney Disease (APOL1). **2018**, 356-359.e1 ○
- 10 Prognostic value of modified-Gustave-Roussy Immunity Score in resectable proximal gastric cancer. **2023**, 102, e33334 ○
- 9 Noninvasive Prenatal Screening for 22q11.2 Deletion/Duplication Syndrome Using multiplex dPCR. ○
- 8 Identification of Extremely Rare Pathogenic CNVs by Array CGH in Saudi Children with Developmental Delay, Congenital Malformations, and Intellectual Disability. **2023**, 10, 662 ○
- 7 Sequence Motif Analysis of PRDM9 and Short Inverted Repeats Suggests Their Contribution to Human Microdeletion and Microduplication Syndromes. **2023**, 3, 267-279 ○

- 6 Molecular Markers for Mutant Characterization. **2023**, 205-232
- 5 Identification of copy number variations in the genome of Dairy Gir cattle. **2023**, 18, e0284085
- 4 From genetic laboratory to the genetic clinic Opportunities and challenges: An illustrated review. **2023**, 20, 123
- 3 Chromosomal microarrays and next-generation sequencing for diagnosis of fetal abnormalities. **2023**, 767-787
- 2 Precision medicine: Overview and challenges to clinical implementation. **2023**, 513-529
- 1 A second unveiling: Haplotig masking of the eastern oyster genome improves population-level inference.