Stylianos Antonarakis

List of Publications by Citations

585 papers	82,262 citations	123 h-index	278 g-index
625 ext. papers	93,711 ext. citations	11.2 avg, IF	8.54 L-index

#	Paper	IF	Citations
585	An integrated encyclopedia of DNA elements in the human genome. *Nature*, **2012**, 489, 57-74	50.4	11449
584	Initial sequencing and comparative analysis of the mouse genome. *Nature*, **2002**, 420, 520-62	50.4	5376
583	Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. *Nature*, **2007**, 447, 799-816	50.4	4121
582	Landscape of transcription in human cells. *Nature*, **2012**, 489, 101-8	50.4	3544
581	Sequence and comparative analysis of the chicken genome provide unique perspectives on vertebrate evolution. *Nature*, **2004**, 432, 695-716	50.4	2143
580	The ENCODE (ENCyclopedia Of DNA Elements) Project. *Science*, **2004**, 306, 636-40	33.3	1692
579	Mutation nomenclature extensions and suggestions to describe complex mutations: a discussion. *Human Mutation*, **2000**, 15, 7-12	4.7	1533
578	Transcriptome and genome sequencing uncovers functional variation in humans. *Nature*, **2013**, 501, 506-11	50.4	1323
577	Positional cloning of the APECED gene. *Nature Genetics*, **1997**, 17, 393-8	36.3	1105
576	A user's guide to the encyclopedia of DNA elements (ENCODE). *PLoS Biology*, **2011**, 9, e1001046	9.7	1060
575	A whole-genome association study of major determinants for host control of HIV-1. *Science*, **2007**, 317, 944-7	33.3	999
574	Genome scan meta-analysis of schizophrenia and bipolar disorder, part II: Schizophrenia. *American Journal of Human Genetics*, **2003**, 73, 34-48	11	985
573	The DNA sequence of human chromosome 21. *Nature*, **2000**, 405, 311-9	50.4	911
572	Guidelines for investigating causality of sequence variants in human disease. *Nature*, **2014**, 508, 469-76	50.4	910
571	Linkage of beta-thalassaemia mutations and beta-globin gene polymorphisms with DNA polymorphisms in human beta-globin gene cluster. *Nature*, **1982**, 296, 627-31	50.4	866
570	The genome sequence of taurine cattle: a window to ruminant biology and evolution. *Science*, **2009**, 324, 522-8	33.3	863
569	Recommendations for a nomenclature system for human gene mutations. Nomenclature Working Group. *Human Mutation*, **1998**, 11, 1-3	4.7	839

568	HGVS Recommendations for the Description of Sequence Variants: 2016 Update. *Human Mutation*, **2016**, 37, 564-9	4.7	792
567	Nomenclature for the description of human sequence variations. *Human Genetics*, **2001**, 109, 121-4	6.3	766
566	Inversions disrupting the factor VIII gene are a common cause of severe haemophilia A. *Nature Genetics*, **1993**, 5, 236-41	36.3	709
565	Haemophilia A resulting from de novo insertion of L1 sequences represents a novel mechanism for mutation in man. *Nature*, **1988**, 332, 164-6	50.4	706
564	Presenile dementia and cerebral haemorrhage linked to a mutation at codon 692 of the beta-amyloid precursor protein gene. *Nature Genetics*, **1992**, 1, 218-21	36.3	652
563	Common regulatory variation impacts gene expression in a cell type-dependent manner. *Science*, **2009**, 325, 1246-50	33.3	607
562	Recurrent rearrangements of chromosome 1q21.1 and variable pediatric phenotypes. *New England Journal of Medicine*, **2008**, 359, 1685-99	59.2	587
561	Schizophrenia susceptibility associated with interstitial deletions of chromosome 22q11. *Proceedings of the National Academy of Sciences of the United States of America*, **1995**, 92, 7612-6	11.5	529
560	Targeted disruption of the mouse factor VIII gene produces a model of haemophilia A. *Nature Genetics*, **1995**, 10, 119-21	36.3	523
559	Assessment of transcript reconstruction methods for RNA-seq. *Nature Methods*, **2013**, 10, 1177-84	21.6	477
558	Chromosome 21 and down syndrome: from genomics to pathophysiology. *Nature Reviews Genetics*, **2004**, 5, 725-38	30.1	476
557	A high-resolution anatomical atlas of the transcriptome in the mouse embryo. *PLoS Biology*, **2011**, 9, e1000582	9.7	467
556	Schizophrenia susceptibility loci on chromosomes 13q32 and 8p21. *Nature Genetics*, **1998**, 20, 70-3	36.3	435
555	GENCODE: producing a reference annotation for ENCODE. *Genome Biology*, **2006**, 7 Suppl 1, S4.1-9	18.3	434
554	Genome-wide associations of gene expression variation in humans. *PLoS Genetics*, **2005**, 1, e78	6	431
553	Continuum of overlapping clones spanning the entire human chromosome 21q. *Nature*, **1992**, 359, 380-7	50.4	394
552	A cluster of cystic fibrosis mutations in the first nucleotide-binding fold of the cystic fibrosis conductance regulator protein. *Nature*, **1990**, 346, 366-9	50.4	387
551	Genome scan meta-analysis of schizophrenia and bipolar disorder, part III: Bipolar disorder. *American Journal of Human Genetics*, **2003**, 73, 49-62	11	353

550	Psychotic illness in patients diagnosed with velo-cardio-facial syndrome and their relatives. *Journal of Nervous and Mental Disease*, **1994**, 182, 476-8	1.8	340
549	Genetic Drivers of Epigenetic and Transcriptional Variation in Human Immune Cells. *Cell*, **2016**, 167, 1398-1414.e24	30.1	339
548	Copy number variants and genetic traits: closer to the resolution of phenotypic to genotypic variability. *Nature Reviews Genetics*, **2007**, 8, 639-46	30.1	335
547	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. *Nature*, **2011**, 478, 97-102	50.4	322
546	Sequential strategy to identify a susceptibility gene for schizophrenia: report of potential linkage on chromosome 22q12-q13.1: Part 1. *American Journal of Medical Genetics Part A*, **1994**, 54, 36-43		321
545	Dodecamer repeat expansion in cystatin B gene in progressive myoclonus epilepsy. *Nature*, **1997**, 386, 847-51	50.4	316
544	Exome sequencing identifies recurrent somatic MAP2K1 and MAP2K2 mutations in melanoma. *Nature Genetics*, **2011**, 44, 133-9	36.3	313
543	Endocytic protein intersectin-l regulates actin assembly via Cdc42 and N-WASP. *Nature Cell Biology*, **2001**, 3, 927-32	23.4	312
542	Common genetic variation and the control of HIV-1 in humans. *PLoS Genetics*, **2009**, 5, e1000791	6	310
541	Human microRNA-155 on chromosome 21 differentially interacts with its polymorphic target in the AGTR1 3' untranslated region: a mechanism for functional single-nucleotide polymorphisms related to phenotypes. *American Journal of Human Genetics*, **2007**, 81, 405-13	11	305
540	In vitro analyses of known and novel RUNX1/AML1 mutations in dominant familial platelet disorder with predisposition to acute myelogenous leukemia: implications for mechanisms of pathogenesis. *Blood*, **2002**, 99, 1364-72	2.2	305
539	Passive and active DNA methylation and the interplay with genetic variation in gene regulation. *ELife*, **2013**, 2, e00523	8.9	295
538	HIV-1 Nef promotes infection by excluding SERINC5 from virion incorporation. *Nature*, **2015**, 526, 212-7	50.4	269
537	COMT genotype predicts longitudinal cognitive decline and psychosis in 22q11.2 deletion syndrome. *Nature Neuroscience*, **2005**, 8, 1500-2	25.5	269
536	Mutations in the DNAH11 (axonemal heavy chain dynein type 11) gene cause one form of situs inversus totalis and most likely primary ciliary dyskinesia. *Proceedings of the National Academy of Sciences of the United States of America*, **2002**, 99, 10282-6	11.5	268
535	BLUEPRINT to decode the epigenetic signature written in blood. *Nature Biotechnology*, **2012**, 30, 224-6	44.5	261
534	Abnormal RNA processing due to the exon mutation of beta E-globin gene. *Nature*, **1982**, 300, 768-9	50.4	256
533	Genomic analysis identifies new drivers and progression pathways in skin basal cell carcinoma. *Nature Genetics*, **2016**, 48, 398-406	36.3	242

532	Genetic structure of Europeans: a view from the North-East. *PLoS ONE*, **2009**, 4, e5472	3.7	237
531	Meiotic and epigenetic defects in Dnmt3L-knockout mouse spermatogenesis. *Proceedings of the National Academy of Sciences of the United States of America*, **2005**, 102, 4068-73	11.5	233
530	The International Human Epigenome Consortium: A Blueprint for Scientific Collaboration and Discovery. *Cell*, **2016**, 167, 1145-1149	56.2	232
529	DNAH5 mutations are a common cause of primary ciliary dyskinesia with outer dynein arm defects. *American Journal of Respiratory and Critical Care Medicine*, **2006**, 174, 120-6	10.2	232
528	Recurrent mutations in haemophilia A give evidence for CpG mutation hotspots. *Nature*, **1986**, 324, 380-2	50.4	230
527	Autoimmune regulator is expressed in the cells regulating immune tolerance in thymus medulla. *Biochemical and Biophysical Research Communications*, **1999**, 257, 821-5	3.4	229
526	Mutations in GJB6 cause hidrotic ectodermal dysplasia. *Nature Genetics*, **2000**, 26, 142-4	36.3	227
525	Schizophrenia: a genome scan targets chromosomes 3p and 8p as potential sites of susceptibility genes. *American Journal of Medical Genetics Part A*, **1995**, 60, 252-60		225
524	DNA polymorphism and molecular pathology of the human globin gene clusters. *Human Genetics*, **1985**, 69, 1-14	6.3	223
523	Isolation and initial characterization of a novel zinc finger gene, DNMT3L, on 21q22.3, related to the cytosine-5-methyltransferase 3 gene family. *Genomics*, **2000**, 65, 293-8	4.3	221
522	Parental origin of the extra chromosome in trisomy 21 as indicated by analysis of DNA polymorphisms. Down Syndrome Collaborative Group. *New England Journal of Medicine*, **1991**, 324, 872-6	59.2	216
521	Conserved non-genic sequences - an unexpected feature of mammalian genomes. *Nature Reviews Genetics*, **2005**, 6, 151-7	30.1	208
520	Polymorphisms in the low-density lipoprotein receptor-related protein 5 (LRP5) gene are associated with variation in vertebral bone mass, vertebral bone size, and stature in whites. *American Journal of Human Genetics*, **2004**, 74, 866-75	11	207
519	Genotype-phenotype correlations in Down syndrome identified by array CGH in 30 cases of partial trisomy and partial monosomy chromosome 21. *European Journal of Human Genetics*, **2009**, 17, 454-66	5.3	203
518	Numerous potentially functional but non-genic conserved sequences on human chromosome 21. *Nature*, **2002**, 420, 578-82	50.4	197
517	Somatic Activating KRAS Mutations in Arteriovenous Malformations of the Brain. *New England Journal of Medicine*, **2018**, 378, 250-261	59.2	195
516	Domains of genome-wide gene expression dysregulation in Down's syndrome. *Nature*, **2014**, 508, 345-50	50.4	195
515	Primary ciliary dyskinesia associated with normal axoneme ultrastructure is caused by DNAH11 mutations. *Human Mutation*, **2008**, 29, 289-98	4.7	193

514	A common mutation in Sardinian autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy patients. *Human Genetics*, **1998**, 103, 428-34	6.3	192
513	Reproducibility of high-throughput mRNA and small RNA sequencing across laboratories. *Nature Biotechnology*, **2013**, 31, 1015-22	44.5	187
512	EGASP: the human ENCODE Genome Annotation Assessment Project. *Genome Biology*, **2006**, 7 Suppl 1, S2.1-31	18.3	187
511	DNA methylation profiles of human active and inactive X chromosomes. *Genome Research*, **2011**, 21, 1592-600	9.7	186
510	Conserved noncoding sequences are selectively constrained and not mutation cold spots. *Nature Genetics*, **2006**, 38, 223-7	36.3	184
509	Novel triplet repeat containing genes in human brain: cloning, expression, and length polymorphisms. *Genomics*, **1993**, 16, 572-9	4.3	183
508	Multicenter linkage study of schizophrenia candidate regions on chromosomes 5q, 6q, 10p, and 13q: schizophrenia linkage collaborative group III. *American Journal of Human Genetics*, **2000**, 67, 652-63	11	182
507	A combined analysis of D22S278 marker alleles in affected sib-pairs: support for a susceptibility locus for schizophrenia at chromosome 22q12. Schizophrenia Collaborative Linkage Group (Chromosome 22). *American Journal of Medical Genetics Part A*, **1996**, 67, 40-5		180
506	Molecular genetic approach to the characterization of the "Down syndrome region" of chromosome 21. *Genomics*, **1989**, 5, 325-31	4.3	178
505	The implications of alternative splicing in the ENCODE protein complement. *Proceedings of the National Academy of Sciences of the United States of America*, **2007**, 104, 5495-500	11.5	177
504	Human chromosome 21 gene expression atlas in the mouse. *Nature*, **2002**, 420, 582-6	50.4	177
503	Hemophilia A. Detection of molecular defects and of carriers by DNA analysis. *New England Journal of Medicine*, **1985**, 313, 842-8	59.2	175
502	Gene duplication: a drive for phenotypic diversity and cause of human disease. *Annual Review of Genomics and Human Genetics*, **2007**, 8, 17-35	9.7	172
501	Human genomics. Effect of predicted protein-truncating genetic variants on the human transcriptome. *Science*, **2015**, 348, 666-9	33.3	170
500	Cloning of the TMPRSS2 gene, which encodes a novel serine protease with transmembrane, LDLRA, and SRCR domains and maps to 21q22.3. *Genomics*, **1997**, 44, 309-20	4.3	168
499	Mendelian disorders deserve more attention. *Nature Reviews Genetics*, **2006**, 7, 277-82	30.1	168
498	The autoimmune regulator protein has transcriptional transactivating properties and interacts with the common coactivator CREB-binding protein. *Journal of Biological Chemistry*, **2000**, 275, 16802-9	5.4	168
497	beta-Thalassemia in American Blacks: novel mutations in the "TATA" box and an acceptor splice site. *Proceedings of the National Academy of Sciences of the United States of America*, **1984**, 81, 1154-8	11.5	168

496	Meis1 and pKnox1 bind DNA cooperatively with Pbx1 utilizing an interaction surface disrupted in oncoprotein E2a-Pbx1. *Proceedings of the National Academy of Sciences of the United States of America*, **1997**, 94, 14553-8	11.5	166
495	Loss of LKB1 kinase activity in Peutz-Jeghers syndrome, and evidence for allelic and locus heterogeneity. *American Journal of Human Genetics*, **1998**, 63, 1641-50	11	166
494	Evolutionary discrimination of mammalian conserved non-genic sequences (CNGs). *Science*, **2003**, 302, 1033-5	33.3	164
493	Binding of PTEN to specific PDZ domains contributes to PTEN protein stability and phosphorylation by microtubule-associated serine/threonine kinases. *Journal of Biological Chemistry*, **2005**, 280, 28936-43	5.4	164
492	Analyses of deep mammalian sequence alignments and constraint predictions for 1% of the human genome. *Genome Research*, **2007**, 17, 760-74	9.7	163
491	Insertion of beta-satellite repeats identifies a transmembrane protease causing both congenital and childhood onset autosomal recessive deafness. *Nature Genetics*, **2001**, 27, 59-63	36.3	159
490	Pseudogenes in the ENCODE regions: consensus annotation, analysis of transcription, and evolution. *Genome Research*, **2007**, 17, 839-51	9.7	158
489	Tandem chimerism as a means to increase protein complexity in the human genome. *Genome Research*, **2006**, 16, 37-44	9.7	157
488	Prominent use of distal 5' transcription start sites and discovery of a large number of additional exons in ENCODE regions. *Genome Research*, **2007**, 17, 746-59	9.7	156
487	RNA and protein expression of the murine autoimmune regulator gene (Aire) in normal, RelB-deficient and in NOD mouse. *European Journal of Immunology*, **2000**, 30, 1884-93	6.1	155
486	Natural gene-expression variation in Down syndrome modulates the outcome of gene-dosage imbalance. *American Journal of Human Genetics*, **2007**, 81, 252-63	11	152
485	Gene expression from the aneuploid chromosome in a trisomy mouse model of down syndrome. *Genome Research*, **2004**, 14, 1268-74	9.7	151
484	Submicroscopic deletion in patients with Williams-Beuren syndrome influences expression levels of the nonhemizygous flanking genes. *American Journal of Human Genetics*, **2006**, 79, 332-41	11	149
483	The phenotypic spectrum of GLI3 morphopathies includes autosomal dominant preaxial polydactyly type-IV and postaxial polydactyly type-A/B; No phenotype prediction from the position of GLI3 mutations. *American Journal of Human Genetics*, **1999**, 65, 645-55	11	149
482	Mutation in GLI3 in postaxial polydactyly type A. *Nature Genetics*, **1997**, 17, 269-71	36.3	148
481	Additional support for schizophrenia linkage on chromosomes 6 and 8: a multicenter study. Schizophrenia Linkage Collaborative Group for Chromosomes 3, 6 and 8. *American Journal of Medical Genetics Part A*, **1996**, 67, 580-94		147
480	Two patients with cystic fibrosis, nonsense mutations in each cystic fibrosis gene, and mild pulmonary disease. *New England Journal of Medicine*, **1990**, 323, 1685-9	59.2	146
479	Factor VIII gene inversions causing severe hemophilia A originate almost exclusively in male germ cells. *Human Molecular Genetics*, **1994**, 3, 1035-9	5.6	144

478	Down syndrome and the complexity of genome dosage imbalance. *Nature Reviews Genetics*, **2017**, 18, 147-163	30.1	143
477	Tissue-specific effects of genetic and epigenetic variation on gene regulation and splicing. *PLoS Genetics*, **2015**, 11, e1004958	6	140
476	Cell-type, allelic, and genetic signatures in the human pancreatic beta cell transcriptome. *Genome Research*, **2013**, 23, 1554-62	9.7	140
475	Identification and characterization of two putative human arginine methyltransferases (HRMT1L1 and HRMT1L2). *Genomics*, **1998**, 48, 330-40	4.3	138
474	Systems medicine and integrated care to combat chronic noncommunicable diseases. *Genome Medicine*, **2011**, 3, 43	14.4	137
473	10 years of Genomics, chromosome 21, and Down syndrome. *Genomics*, **1998**, 51, 1-16	4.3	136
472	Synaptojanin 1-linked phosphoinositide dyshomeostasis and cognitive deficits in mouse models of Down's syndrome. *Proceedings of the National Academy of Sciences of the United States of America*, **2008**, 105, 9415-20	11.5	133
471	Common mutations in autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy patients of different origins. *Molecular Endocrinology*, **1998**, 12, 1112-9		133
470	Structured RNAs in the ENCODE selected regions of the human genome. *Genome Research*, **2007**, 17, 852-64	9.7	131
469	Multi-omic measurements of heterogeneity in HeLa cells across laboratories. *Nature Biotechnology*, **2019**, 37, 314-322	44.5	129
468	Mutations in ZMYND10, a gene essential for proper axonemal assembly of inner and outer dynein arms in humans and flies, cause primary ciliary dyskinesia. *American Journal of Human Genetics*, **2013**, 93, 346-56	11	126
467	Prenatal diagnosis using DNA polymorphisms. Report on 95 pregnancies at risk for sickle-cell disease or beta-thalassemia. *New England Journal of Medicine*, **1983**, 308, 1054-8	59.2	126
466	The transmembrane serine protease (TMPRSS3) mutated in deafness DFNB8/10 activates the epithelial sodium channel (ENaC) in vitro. *Human Molecular Genetics*, **2002**, 11, 2829-36	5.6	124
465	Follow-up of a report of a potential linkage for schizophrenia on chromosome 22q12-q13.1: Part 2. *American Journal of Medical Genetics Part A*, **1994**, 54, 44-50		124
464	Peutz-Jeghers syndrome: confirmation of linkage to chromosome 19p13.3 and identification of a potential second locus, on 19q13.4. *American Journal of Human Genetics*, **1997**, 61, 1327-34	11	123
463	DYRK1A-dosage imbalance perturbs NRSF/REST levels, deregulating pluripotency and embryonic stem cell fate in Down syndrome. *American Journal of Human Genetics*, **2008**, 83, 388-400	11	123
462	Primary ciliary dyskinesia: a genome-wide linkage analysis reveals extensive locus heterogeneity. *European Journal of Human Genetics*, **2000**, 8, 109-18	5.3	123
461	Molecular characterization of mild-to-moderate hemophilia A: detection of the mutation in 25 of 29 patients by denaturing gradient gel electrophoresis. *Proceedings of the National Academy of Sciences of the United States of America*, **1991**, 88, 8307-11	11.5	121

460	Down syndrome. *Nature Reviews Disease Primers*, **2020**, 6, 9	51.1	120
459	Mitotic errors in somatic cells cause trisomy 21 in about 4.5% of cases and are not associated with advanced maternal age. *Nature Genetics*, **1993**, 3, 146-50	36.3	120
458	Modelling and rescuing neurodevelopmental defect of Down syndrome using induced pluripotent stem cells from monozygotic twins discordant for trisomy 21. *EMBO Molecular Medicine*, **2014**, 6, 259-77	12	117
457	Schizophrenia susceptibility and chromosome 6p24-22. *Nature Genetics*, **1995**, 11, 235-6	36.3	113
456	APOBEC-induced mutations in human cancers are strongly enriched on the lagging DNA strand during replication. *Genome Research*, **2016**, 26, 174-82	9.7	110
455	Molecular etiology of factor VIII deficiency in hemophilia A. *Human Mutation*, **1995**, 5, 1-22	4.7	110
454	Evidence for multiple origins of the beta E-globin gene in Southeast Asia. *Proceedings of the National Academy of Sciences of the United States of America*, **1982**, 79, 6608-11	11.5	109
453	The complete sequence of a human genome.. *Science*, **2022**, 376, 44-53	33.3	107
452	APECED mutations in the autoimmune regulator (AIRE) gene. *Human Mutation*, **2001**, 18, 205-11	4.7	106
451	TNPO3 protects HIV-1 replication from CPSF6-mediated capsid stabilization in the host cell cytoplasm. *Retrovirology*, **2013**, 10, 20	3.6	105
450	Molecular heterogeneity of inherited antithrombin III deficiency. *New England Journal of Medicine*, **1983**, 308, 1549-52	59.2	105
449	Aire-deficient C57BL/6 mice mimicking the common human 13-base pair deletion mutation present with only a mild autoimmune phenotype. *Journal of Immunology*, **2009**, 182, 3902-18	5.3	103
448	DEPDC5 mutations in families presenting as autosomal dominant nocturnal frontal lobe epilepsy. *Neurology*, **2014**, 82, 2101-6	6.5	98
447	Comparison of mouse and human genomes followed by experimental verification yields an estimated 1,019 additional genes. *Proceedings of the National Academy of Sciences of the United States of America*, **2003**, 100, 1140-5	11.5	95
446	LKB1 interacts with and phosphorylates PTEN: a functional link between two proteins involved in cancer predisposing syndromes. *Human Molecular Genetics*, **2005**, 14, 2209-19	5.6	95
445	The challenge of Down syndrome. *Trends in Molecular Medicine*, **2006**, 12, 473-9	11.5	94
444	A large genomic deletion leads to enhancer adoption by the lamin B1 gene: a second path to autosomal dominant adult-onset demyelinating leukodystrophy (ADLD). *Human Molecular Genetics*, **2015**, 24, 3143-54	5.6	93
443	Identification of sequence variants and analysis of the role of the catechol-O-methyl-transferase gene in schizophrenia susceptibility. *Biological Psychiatry*, **1998**, 43, 425-31	7.9	93

442	Gene expression variation and expression quantitative trait mapping of human chromosome 21 genes. *Human Molecular Genetics*, **2005**, 14, 3741-9	5.6	93
441	Guidelines for human gene nomenclature (1997). HUGO Nomenclature Committee. *Genomics*, **1997**, 45, 468-71	4.3	91
440	Early history of mammals is elucidated with the ENCODE multiple species sequencing data. *PLoS Genetics*, **2007**, 3, e2	6	91
439	Mutation analyses of North American APS-1 patients. *Human Mutation*, **1999**, 13, 69-74	4.7	90
438	Down syndrome: from understanding the neurobiology to therapy. *Journal of Neuroscience*, **2010**, 30, 14943-5	6.6	89
437	Diagnosis of genetic disorders at the DNA level. *New England Journal of Medicine*, **1989**, 320, 153-63	59.2	89
436	Biased allelic expression in human primary fibroblast single cells. *American Journal of Human Genetics*, **2015**, 96, 70-80	11	88
435	X-linked primary ciliary dyskinesia due to mutations in the cytoplasmic axonemal dynein assembly factor PIH1D3. *Nature Communications*, **2017**, 8, 14279	17.4	87
434	A new mouse model for the trisomy of the Abcg1-U2af1 region reveals the complexity of the combinatorial genetic code of down syndrome. *Human Molecular Genetics*, **2009**, 18, 4756-69	5.6	86
433	Genetic heterogeneity in schizophrenia: stratification of genome scan data using co-segregating related phenotypes. *Molecular Psychiatry*, **2000**, 5, 650-3	15.1	86
432	Two isoforms of a human intersectin (ITSN) protein are produced by brain-specific alternative splicing in a stop codon. *Genomics*, **1998**, 53, 369-76	4.3	86
431	Characterization of five partial deletions of the factor VIII gene. *Proceedings of the National Academy of Sciences of the United States of America*, **1987**, 84, 3772-6	11.5	85
430	New class of gene-termini-associated human RNAs suggests a novel RNA copying mechanism. *Nature*, **2010**, 466, 642-6	50.4	84
429	Life-history traits drive the evolutionary rates of mammalian coding and noncoding genomic elements. *Proceedings of the National Academy of Sciences of the United States of America*, **2007**, 104, 20443-8	11.5	84
428	APECED: a monogenic autoimmune disease providing new clues to self-tolerance. *Trends in Immunology*, **1998**, 19, 384-6		83
427	Regulation of fibrinogen production by microRNAs. *Blood*, **2010**, 116, 2608-15	2.2	81
426	Deciphering Cell Lineage Specification during Male Sex Determination with Single-Cell RNA Sequencing. *Cell Reports*, **2018**, 22, 1589-1599	10.6	80
425	Characterization of mutations in the factor VIII gene by direct sequencing of amplified genomic DNA. *Genomics*, **1990**, 6, 65-71	4.3	80

424	Linkage map of the short arm of human chromosome 11: location of the genes for catalase, calcitonin, and insulin-like growth factor II. *Proceedings of the National Academy of Sciences of the United States of America*, **1985**, 82, 5064-7	11.5	80
423	Knobloch syndrome: novel mutations in COL18A1, evidence for genetic heterogeneity, and a functionally impaired polymorphism in endostatin. *Human Mutation*, **2004**, 23, 77-84	4.7	79
422	Mice trisomic for a bacterial artificial chromosome with the single-minded 2 gene (Sim2) show phenotypes similar to some of those present in the partial trisomy 16 mouse models of Down syndrome. *Human Molecular Genetics*, **2000**, 9, 1853-64	5.6	79
421	Haemophilia A: database of nucleotide substitutions, deletions, insertions and rearrangements of the factor VIII gene, second edition. *Nucleic Acids Research*, **1994**, 22, 3511-33	20.1	79
420	The cerebellar transcriptome during postnatal development of the Ts1Cje mouse, a segmental trisomy model for Down syndrome. *Human Molecular Genetics*, **2005**, 14, 373-84	5.6	78
419	A genetic linkage map of 27 markers on human chromosome 21. *Genomics*, **1991**, 9, 407-19	4.3	78
418	Nonhomologous recombination in the human genome: deletions in the human factor VIII gene. *Genomics*, **1991**, 10, 94-101	4.3	78
417	Molecular and clinical characterization of 25 individuals with exonic deletions of NRXN1 and comprehensive review of the literature. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2013**, 162B, 388-403	3.5	76
416	Trisomy for synaptojanin1 in Down syndrome is functionally linked to the enlargement of early endosomes. *Human Molecular Genetics*, **2012**, 21, 3156-72	5.6	76
415	Disease-causing 7.4 kb cis-regulatory deletion disrupting conserved non-coding sequences and their interaction with the FOXL2 promotor: implications for mutation screening. *PLoS Genetics*, **2009**, 5, e1000522	6	76
414	A genomic rearrangement resulting in a tandem duplication is associated with split hand-split foot malformation 3 (SHFM3) at 10q24. *Human Molecular Genetics*, **2003**, 12, 1959-71	5.6	75
413	A genetic linkage map of 17 markers on human chromosome 21. *Genomics*, **1989**, 4, 579-91	4.3	75
412	The TPTE gene family: cellular expression, subcellular localization and alternative splicing. *Gene*, **2003**, 323, 189-99	3.8	74
411	Follow-up report of potential linkage for schizophrenia on chromosome 22q: Part 3. *American Journal of Medical Genetics Part A*, **1995**, 60, 172-3		74
410	Single-minded and Down syndrome?. *Nature Genetics*, **1995**, 10, 9-10	36.3	73
409	Clinical, biochemical, and neuropsychiatric evaluation of a patient with a contiguous gene syndrome due to a microdeletion Xp11.3 including the Norrie disease locus and monoamine oxidase (MAOA and MAOB) genes. *American Journal of Medical Genetics Part A*, **1992**, 42, 127-34		73
408	Construction of human chromosome 21-specific yeast artificial chromosomes. *Proceedings of the National Academy of Sciences of the United States of America*, **1989**, 86, 9991-5	11.5	73
407	Use of denaturing gradient gel electrophoresis to detect point mutations in the factor VIII gene. *Genomics*, **1990**, 6, 293-301	4.3	73

406	Epistatic interactions with a common hypomorphic RET allele in syndromic Hirschsprung disease. *Human Mutation*, **2007**, 28, 790-6	4.7	71
405	Functional genetic variation of human miRNAs and phenotypic consequences. *Mammalian Genome*, **2008**, 19, 503-9	3.2	70
404	Duplications of BHLHA9 are associated with ectrodactyly and tibia hemimelia inherited in non-Mendelian fashion. *Journal of Medical Genetics*, **2012**, 49, 119-25	5.8	68
403	Molecular genetic analysis in autosomal dominant keratoconus. *Cornea*, **1992**, 11, 302-8	3.1	68
402	Human chromosome 21: genome mapping and exploration, circa 1993. *Trends in Genetics*, **1993**, 9, 142-8	8.5	68
401	The mouse brain transcriptome by SAGE: differences in gene expression between P30 brains of the partial trisomy 16 mouse model of Down syndrome (Ts65Dn) and normals. *Genome Research*, **2000**, 10, 2006-21	9.7	68
400	Diagnostic exome sequencing to elucidate the genetic basis of likely recessive disorders in consanguineous families. *Human Mutation*, **2014**, 35, 1203-10	4.7	67
399	Novel missense mutations of TMPRSS3 in two consanguineous Tunisian families with non-syndromic autosomal recessive deafness. *Human Mutation*, **2001**, 18, 101-8	4.7	67
398	MLL2 mutation detection in 86 patients with Kabuki syndrome: a genotype-phenotype study. *Clinical Genetics*, **2013**, 84, 539-45	4	66
397	Paternal nondisjunction in trisomy 21: excess of male patients. *Human Molecular Genetics*, **1993**, 2, 1691-5	5.6	66
396	Cloning of a novel human neural cell adhesion molecule gene (NCAM2) that maps to chromosome region 21q21 and is potentially involved in Down syndrome. *Genomics*, **1997**, 43, 43-51	4.3	65
395	Language skills in children with velocardiofacial syndrome (deletion 22q11.2). *Journal of Pediatrics*, **2002**, 140, 753-8	3.6	65
394	DNA polymorphism haplotypes of the human apolipoprotein APOA1-APOC3-APOA4 gene cluster. *Human Genetics*, **1988**, 80, 265-73	6.3	65
393	Deletion of the fibrinogen [correction of fibrogen] alpha-chain gene (FGA) causes congenital afibrogenemia. *Journal of Clinical Investigation*, **1999**, 103, 215-8	15.9	64
392	The human gene encoding insulin-like growth factor I is located on chromosome 12. *Human Genetics*, **1985**, 69, 157-60	6.3	63
391	LRP5 gene polymorphisms and idiopathic osteoporosis in men. *Bone*, **2005**, 37, 770-5	4.7	62
390	Association of the connexin36 gene with juvenile myoclonic epilepsy. *Journal of Medical Genetics*, **2004**, 41, e93	5.8	62
389	A gene which causes severe ocular alterations and occipital encephalocele (Knobloch syndrome) is mapped to 21q22.3. *Human Molecular Genetics*, **1996**, 5, 843-7	5.6	62

388	A linkage map of human chromosome 21:43 PCR markers at average intervals of 2.5 cM. *Genomics*, **1993**, 16, 562-71	4.3	62
387	Exome sequencing identifies putative drivers of progression of transient myeloproliferative disorder to AMKL in infants with Down syndrome. *Blood*, **2013**, 122, 554-61	2.2	60
386	Mendelian disorders and multifactorial traits: the big divide or one for all?. *Nature Reviews Genetics*, **2010**, 11, 380-4	30.1	60
385	Identification and characterization of a novel cyclic nucleotide phosphodiesterase gene (PDE9A) that maps to 21q22.3: alternative splicing of mRNA transcripts, genomic structure and sequence. *Human Genetics*, **1998**, 103, 386-92	6.3	60
384	Phylogeny of human beta-globin haplotypes and its implications for recent human evolution. *American Journal of Physical Anthropology*, **1990**, 81, 113-30	2.5	60
383	The molecular basis of hemophilia A in man. *Trends in Genetics*, **1988**, 4, 233-7	8.5	60
382	Systematic proteome and proteostasis profiling in human Trisomy 21 fibroblast cells. *Nature Communications*, **2017**, 8, 1212	17.4	59
381	Pathogenic Variants in PIGG Cause Intellectual Disability with Seizures and Hypotonia. *American Journal of Human Genetics*, **2016**, 98, 615-26	11	59
380	Exome sequencing in 53 sporadic cases of schizophrenia identifies 18 putative candidate genes. *PLoS ONE*, **2014**, 9, e112745	3.7	59
379	In vitro whole-genome analysis identifies a susceptibility locus for HIV-1. *PLoS Biology*, **2008**, 6, e32	9.7	59
378	A DNA resequencing array for pathogenic mutation detection in hypertrophic cardiomyopathy. *Human Mutation*, **2008**, 29, 879-85	4.7	59
377	Linkage analysis of the human HMG14 gene on chromosome 21 using a GT dinucleotide repeat as polymorphic marker. *Genomics*, **1990**, 7, 136-8	4.3	59
376	Identifying protein-coding genes in genomic sequences. *Genome Biology*, **2009**, 10, 201	18.3	58
375	The subcellular localization of the ChoRE-binding protein, encoded by the Williams-Beuren syndrome critical region gene 14, is regulated by 14-3-3. *Human Molecular Genetics*, **2004**, 13, 1505-14	5.6	58
374	Molecular analysis of the fibrinogen gene cluster in 16 patients with congenital afibrinogenemia: novel truncating mutations in the FGA and FGG genes. *Human Genetics*, **2001**, 108, 237-40	6.3	58
373	A testis-specific gene, TPTE, encodes a putative transmembrane tyrosine phosphatase and maps to the pericentromeric region of human chromosomes 21 and 13, and to chromosomes 15, 22, and Y. *Human Genetics*, **1999**, 105, 399-409	6.3	58
372	Characterization of a nondeleterious L1 insertion in an intron of the human factor VIII gene and further evidence of open reading frames in functional L1 elements. *Genomics*, **1989**, 4, 290-6	4.3	58
371	The complete sequence of a human genome		58

370	Identification of cis- and trans-regulatory variation modulating microRNA expression levels in human fibroblasts. *Genome Research*, **2011**, 21, 68-73	9.7	57
369	Cloning of two human homologs of the Drosophila single-minded gene SIM1 on chromosome 6q and SIM2 on 21q within the Down syndrome chromosomal region. *Genome Research*, **1997**, 7, 615-24	9.7	57
368	Parental origin of the deletion 22q11.2 and brain development in velocardiofacial syndrome: a preliminary study. *Archives of General Psychiatry*, **2001**, 58, 64-8		57
367	Isolation and initial characterization of the mouse Dnmt3l gene. *Cytogenetic and Genome Research*, **2001**, 92, 122-6	1.9	57
366	OMIM passes the 1,000-disease-gene mark. *Nature Genetics*, **2000**, 25, 11	36.3	57
365	A PCR amplification method reveals instability of the dodecamer repeat in progressive myoclonus epilepsy (EPM1) and no correlation between the size of the repeat and age at onset. *American Journal of Human Genetics*, **1998**, 62, 842-7	11	56
364	Genetic and epigenetic regulation of human lincRNA gene expression. *American Journal of Human Genetics*, **2013**, 93, 1015-26	11	55
363	Preliminary structure and predictive value of attenuated negative symptoms in 22q11.2 deletion syndrome. *Psychiatry Research*, **2012**, 196, 277-84	9.9	55
362	Comparison of human chromosome 21 conserved nongenic sequences (CNGs) with the mouse and dog genomes shows that their selective constraint is independent of their genic environment. *Genome Research*, **2004**, 14, 852-9	9.7	55
361	Loss of function mutation in the palmitoyl-transferase HHAT leads to syndromic 46,XY disorder of sex development by impeding Hedgehog protein palmitoylation and signaling. *PLoS Genetics*, **2014**, 10, e1004340	6	54
360	Tmprss3, a transmembrane serine protease deficient in human DFNB8/10 deafness, is critical for cochlear hair cell survival at the onset of hearing. *Journal of Biological Chemistry*, **2011**, 286, 17383-97	5.4	54
359	Two high throughput technologies to detect segmental aneuploidies identify new Williams-Beuren syndrome patients with atypical deletions. *Journal of Medical Genetics*, **2006**, 43, 266-73	5.8	54
358	Isolation and characterization of the mouse Aire gene. *Biochemical and Biophysical Research Communications*, **1999**, 255, 483-90	3.4	54
357	The tetranucleotide repeat polymorphism D21S1245 demonstrates hypermutability in germline and somatic cells. *Human Molecular Genetics*, **1995**, 4, 1193-9	5.6	54
356	Uniparental isodisomy due to duplication of chromosome 21 occurring in somatic cells monosomic for chromosome 21. *Genomics*, **1992**, 13, 269-74	4.3	54
355	Carney complex, Peutz-Jeghers syndrome, Cowden disease, and Bannayan-Zonana syndrome share cutaneous and endocrine manifestations, but not genetic loci. *Journal of Clinical Endocrinology and Metabolism*, **1998**, 83, 2972-6	5.6	53
354	Haemophilia A: database of nucleotide substitutions, deletions, insertions and rearrangements of the factor VIII gene, second edition. *Nucleic Acids Research*, **1994**, 22, 4851-68	20.1	52
353	Phenotypic variability in siblings with Farber disease. *Journal of Pediatrics*, **1984**, 104, 406-9	3.6	52

352	A novel homozygous mutation in FGFR3 causes tall stature, severe lateral tibial deviation, scoliosis, hearing impairment, camptodactyly, and arachnodactyly. *Human Mutation*, **2014**, 35, 959-63	4.7	51
351	Evidence for transcript networks composed of chimeric RNAs in human cells. *PLoS ONE*, **2012**, 7, e28213	3.7	51
350	Human nocturnal frontal lobe epilepsy: pharmocogenomic profiles of pathogenic nicotinic acetylcholine receptor beta-subunit mutations outside the ion channel pore. *Molecular Pharmacology*, **2008**, 74, 379-91	4.3	51
349	No evidence for linkage between schizophrenia and markers at chromosome 15q13-14. *American Journal of Medical Genetics Part A*, **1999**, 88, 109-12		51
348	Familial isolated aniridia associated with a translocation involving chromosomes 11 and 22 [t(11;22)(p13;q12.2)]. *Human Genetics*, **1986**, 72, 297-302	6.3	51
347	Genetic diseases: diagnosis by restriction endonuclease analysis. *Journal of Pediatrics*, **1982**, 100, 845-56	3.6	51
346	Genetic characterization of northeastern Italian population isolates in the context of broader European genetic diversity. *European Journal of Human Genetics*, **2013**, 21, 659-65	5.3	50
345	A single-nucleotide substitution mutator phenotype revealed by exome sequencing of human colon adenomas. *Cancer Research*, **2012**, 72, 6279-89	10.1	50
344	Cloning of a human RNA editing deaminase (ADARB1) of glutamate receptors that maps to chromosome 21q22.3. *Genomics*, **1997**, 41, 210-7	4.3	50
343	Laryngeal atresia type III (glottic web) with 22q11.2 microdeletion: Report of three patients **1997**, 70, 130-133		50
342	Abnormal cortical activation during response inhibition in 22q11.2 deletion syndrome. *Human Brain Mapping*, **2007**, 28, 533-42	5.9	50
341	Mutations in the TMPRSS3 gene are a rare cause of childhood nonsyndromic deafness in Caucasian patients. *Journal of Molecular Medicine*, **2002**, 80, 124-31	5.5	50
340	Disease-causing mutations in the human genome. *European Journal of Pediatrics*, **2000**, 159 Suppl 3, S173-8	4.8	50
339	Cloning of a human homolog of the Drosophila enhancer of zeste gene (EZH2) that maps to chromosome 21q22.2. *Genomics*, **1996**, 38, 30-7	4.3	50
338	RFLP for the human lipoprotein lipase (LPL) gene: HindIII. *Nucleic Acids Research*, **1987**, 15, 6763	20.1	50
337	Carrier screening for recessive disorders. *Nature Reviews Genetics*, **2019**, 20, 549-561	30.1	49
336	Cardiomyogenesis is controlled by the miR-99a/let-7c cluster and epigenetic modifications. *Stem Cell Research*, **2014**, 12, 323-37	1.6	49
335	Spectrum of mutations in CRM-positive and CRM-reduced hemophilia A. *Genomics*, **1993**, 15, 392-8	4.3	49

334	The complex SNP and CNV genetic architecture of the increased risk of congenital heart defects in Down syndrome. *Genome Research*, **2013**, 23, 1410-21	9.7	48
333	Evolutionary comparison provides evidence for pathogenicity of RMRP mutations. *PLoS Genetics*, **2005**, 1, e47	6	48
332	Hemophilia A due to mutations that create new N-glycosylation sites. *Proceedings of the National Academy of Sciences of the United States of America*, **1992**, 89, 4933-7	11.5	46
331	Extrachromosomal driver mutations in glioblastoma and low-grade glioma. *Nature Communications*, **2014**, 5, 5690	17.4	45
330	Three common polymorphisms in the CYBA gene form a haplotype associated with decreased ROS generation. *Human Mutation*, **2009**, 30, 1123-33	4.7	45
329	Mouse models for Down syndrome-associated developmental cognitive disabilities. *Developmental Neuroscience*, **2011**, 33, 404-13	2.2	44
328	A narcolepsy susceptibility locus maps to a 5 Mb region of chromosome 21q. *Annals of Neurology*, **2004**, 56, 382-8	9.4	44
327	Determination of gene dosage by a quantitative adaptation of the polymerase chain reaction (gd-PCR): rapid detection of deletions and duplications of gene sequences. *Genomics*, **1994**, 21, 304-10	4.3	44
326	Search for the second Peutz-Jeghers syndrome locus: exclusion of the STK13, PRKCG, KLK10, and PSCD2 genes on chromosome 19 and the STK11IP gene on chromosome 2. *Cytogenetic and Genome Research*, **2002**, 97, 171-8	1.9	43
325	Nineteen additional unpredicted transcripts from human chromosome 21. *Genomics*, **2002**, 79, 824-32	4.3	43
324	Brief report: isogenic induced pluripotent stem cell lines from an adult with mosaic down syndrome model accelerated neuronal ageing and neurodegeneration. *Stem Cells*, **2015**, 33, 2077-84	5.8	42
323	Pathogenic mutations and polymorphisms in the lipoprotein receptor-related protein 5 reveal a new biological pathway for the control of bone mass. *Current Opinion in Lipidology*, **2005**, 16, 207-14	4.4	42
322	Clouston hidrotic ectodermal dysplasia (HED): genetic homogeneity, presence of a founder effect in the French Canadian population and fine genetic mapping. *European Journal of Human Genetics*, **2000**, 8, 372-80	5.3	42
321	A teratocarcinoma-like human embryonic stem cell (hESC) line and four hESC lines reveal potentially oncogenic genomic changes. *PLoS ONE*, **2010**, 5, e10263	3.7	42
320	DNA Methylation Profiling of Uniparental Disomy Subjects Provides a Map of Parental Epigenetic Bias in the Human Genome. *American Journal of Human Genetics*, **2016**, 99, 555-566	11	41
319	Purifying selection in mammalian mitochondrial protein-coding genes is highly effective and congruent with evolution of nuclear genes. *Molecular Biology and Evolution*, **2013**, 30, 347-55	8.3	41
318	Frequent cases of RAS-mutated Down syndrome acute lymphoblastic leukaemia lack JAK2 mutations. *Nature Communications*, **2014**, 5, 4654	17.4	41
317	Asp1424Asn MYH9 mutation results in an unstable protein responsible for the phenotypes in May-Hegglin anomaly/Fechtner syndrome. *Blood*, **2003**, 102, 529-34	2.2	41

316	Genetic variability of mu-opioid receptor in an obstetric population. *Anesthesiology*, **2004**, 100, 1030-3	4.3	41
315	Axonemal beta heavy chain dynein DNAH9: cDNA sequence, genomic structure, and investigation of its role in primary ciliary dyskinesia. *Genomics*, **2001**, 72, 21-33	4.3	41
314	A transmission disequilibrium and linkage analysis of D22S278 marker alleles in 574 families: further support for a susceptibility locus for schizophrenia at 22q12. Schizophrenia Collaborative Linkage Group for Chromosome 22. *Schizophrenia Research*, **1998**, 32, 115-21	3.6	41
313	Localization of 102 exons to a 2.5 Mb region involved in Down syndrome. *Human Molecular Genetics* , **1995**, 4, 1305-11	5.6	41
312	Analysis of DNA haplotypes suggests a genetic predisposition to trisomy 21 associated with DNA sequences on chromosome 21. *Proceedings of the National Academy of Sciences of the United States of America*, **1985**, 82, 3360-4	11.5	41
311	Network-guided analysis of genes with altered somatic copy number and gene expression reveals pathways commonly perturbed in metastatic melanoma. *PLoS ONE*, **2011**, 6, e18369	3.7	40
310	Cloning of 559 potential exons of genes of human chromosome 21 by exon trapping. *Genome Research*, **1996**, 6, 747-60	9.7	40
309	Analysis of the human Sonic Hedgehog coding and promoter regions in sacral agenesis, triphalangeal thumb, and mirror polydactyly. *Human Genetics*, **1998**, 102, 387-92	6.3	40
308	No evidence for an effect of COMT Val158Met genotype on executive function in patients with 22q11 deletion syndrome. *American Journal of Psychiatry*, **2006**, 163, 537-9	11.9	40
307	Different mechanisms preclude mutant CLDN14 proteins from forming tight junctions in vitro. *Human Mutation*, **2005**, 25, 543-9	4.7	40
306	Peutz-Jeghers LKB1 mutants fail to activate GSK-3beta, preventing it from inhibiting Wnt signaling. *Molecular Genetics and Genomics*, **2005**, 273, 184-96	3.1	40
305	A t(2;8) balanced translocation with breakpoints near the human HOXD complex causes mesomelic dysplasia and vertebral defects. *Genomics*, **2002**, 79, 493-8	4.3	40
304	Altered spacing of promoter elements due to the dodecamer repeat expansion contributes to reduced expression of the cystatin B gene in EPM1. *Human Molecular Genetics*, **1999**, 8, 1791-8	5.6	40
303	Microdeletion in the X-chromosome and prenatal diagnosis in a family with Norrie disease. *American Journal of Medical Genetics Part A*, **1989**, 33, 485-8		40
302	Yeast artificial chromosome vectors for efficient clone manipulation and mapping. *Genomics*, **1991**, 10, 505-8	4.3	40
301	Cloning of a novel homeobox-containing gene, PKNOX1, and mapping to human chromosome 21q22.3. *Genomics*, **1997**, 41, 193-200	4.3	39
300	Detection of aneuploidies by paralogous sequence quantification. *Journal of Medical Genetics*, **2004** , 41, 908-15	5.8	39
299	Isolation and characterization of the UBASH3A gene on 21q22.3 encoding a potential nuclear protein with a novel combination of domains. *Human Genetics*, **2001**, 108, 140-7	6.3	39

298	Transcriptional map of the 2.5-Mb CBR-ERG region of chromosome 21 involved in Down syndrome. *Genomics*, **1998**, 48, 12-23	4.3	39
297	The same "TATA" box beta-thalassemia mutation in Chinese and US blacks: another example of independent origins of mutation. *Human Genetics*, **1986**, 74, 162-4	6.3	39
296	Novel mutations of TMPRSS3 in four DFNB8/B10 families segregating congenital autosomal recessive deafness. *Journal of Medical Genetics*, **2001**, 38, 396-400	5.8	39
295	DNA-Methylation Patterns in Trisomy 21 Using Cells from Monozygotic Twins. *PLoS ONE*, **2015**, 10, e0135555	3.7	39
294	Exome sequencing reveals pathogenic mutations in 91 strains of mice with Mendelian disorders. *Genome Research*, **2015**, 25, 948-57	9.7	38
293	A novel TMPRSS3 missense mutation in a DFNB8/10 family prevents proteolytic activation of the protein. *Human Genetics*, **2005**, 117, 528-35	6.3	38
292	Extensive cellular heterogeneity of X inactivation revealed by single-cell allele-specific expression in human fibroblasts. *Proceedings of the National Academy of Sciences of the United States of America*, **2018**, 115, 13015-13020	11.5	38
291	Biallelic variants in KIF14 cause intellectual disability with microcephaly. *European Journal of Human Genetics*, **2018**, 26, 330-339	5.3	37
290	The locus for combined factor V-factor VIII deficiency (F5F8D) maps to 18q21, between D18S849 and D18S1103. *American Journal of Human Genetics*, **1997**, 61, 143-50	11	37
289	Patterns of meiotic recombination on the long arm of human chromosome 21. *Genome Research*, **2000**, 10, 1319-32	9.7	37
288	Methylation profiling in individuals with uniparental disomy identifies novel differentially methylated regions on chromosome 15. *Genome Research*, **2010**, 20, 1271-8	9.7	36
287	Mitochondrial DNA mutation and heteroplasmy in type I Leber hereditary optic neuropathy. *American Journal of Medical Genetics Part A*, **1992**, 42, 173-9		36
286	Efficient targeted transcript discovery via array-based normalization of RACE libraries. *Nature Methods*, **2008**, 5, 629-35	21.6	35
285	MECP2 mutant allele in a boy with Rett syndrome and his unaffected heterozygous mother. *Brain and Development*, **2007**, 29, 47-50	2.2	35
284	Detection of Imprinted Genes by Single-Cell Allele-Specific Gene Expression. *American Journal of Human Genetics*, **2017**, 100, 444-453	11	34
283	An apparently dominant bipolar affective disorder (BPAD) locus on chromosome 20p11.2-q11.2 in a large Turkish pedigree. *European Journal of Human Genetics*, **2001**, 9, 39-44	5.3	34
282	Homologous loci DXYS156X and DXYS156Y contain a polymorphic pentanucleotide repeat (TAAAA)n and map to human X and Y chromosomes. *Human Mutation*, **1994**, 4, 208-11	4.7	34
281	A cSNP Map and Database for Human Chromosome 21. *Genome Research*, **2001**, 11, 300-307	9.7	34

280	Rapid detection of genetic variants in hypertrophic cardiomyopathy by custom DNA resequencing array in clinical practice. *Journal of Medical Genetics*, **2011**, 48, 572-6	5.8	33
279	Specific BACE1 genotypes provide additional risk for late-onset Alzheimer disease in APOE epsilon 4 carriers **2003**, 119B, 44-7		33
278	Isolation and characterization of a human chromosome 21q22.3 gene (WDR4) and its mouse homologue that code for a WD-repeat protein. *Genomics*, **2000**, 68, 71-9	4.3	33
277	From PREDs and open reading frames to cDNA isolation: revisiting the human chromosome 21 transcription map. *Genomics*, **2001**, 78, 46-54	4.3	33
276	Autosomal-Recessive Mutations in AP3B2, Adaptor-Related Protein Complex 3 Beta 2 Subunit, Cause an Early-Onset Epileptic Encephalopathy with Optic Atrophy. *American Journal of Human Genetics*, **2016**, 99, 1368-1376	11	32
275	A recurrent 14q32.2 microdeletion mediated by expanded TGG repeats. *Human Molecular Genetics*, **2010**, 19, 1967-73	5.6	32
274	Proliferation deficits and gene expression dysregulation in Down's syndrome (Ts1Cje) neural progenitor cells cultured from neurospheres. *Journal of Neuroscience Research*, **2009**, 87, 3143-52	4.4	32
273	Alphoid DNA polymorphisms for chromosome 21 can be distinguished from those of chromosome 13 using probes homologous to both. *Genomics*, **1991**, 9, 141-6	4.3	32
272	Farber disease: pathologic diagnosis in sibs with phenotypic variability. *American Journal of Medical Genetics Part A*, **1987**, 3, 233-41		32
271	Simultaneous identification and prioritization of variants in familial, de novo, and somatic genetic disorders with VariantMaster. *Genome Research*, **2014**, 24, 349-55	9.7	31
270	Activation of multiple cryptic donor splice sites by the common congenital afibrinogenemia mutation, FGA IVS4 + 1 G-->T. *Blood*, **2001**, 97, 1879-81	2.2	31
269	Eye gaze during face processing in children and adolescents with 22q11.2 deletion syndrome. *Journal of the American Academy of Child and Adolescent Psychiatry*, **2010**, 49, 665-74	7.2	30
268	Linkage mapping of highly informative DNA polymorphisms within the human interferon-alpha receptor gene on chromosome 21. *Genomics*, **1991**, 11, 573-6	4.3	30
267	TMPRSS3, a type II transmembrane serine protease mutated in non-syndromic autosomal recessive deafness. *Frontiers in Bioscience - Landmark*, **2008**, 13, 1557-67	2.8	30
266	Monozygotic twins discordant for trisomy 21 and maternal 21q inheritance: a complex series of events. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 2086-93	2.5	29
265	"Compensatory" uniparental disomy of chromosome 21 in two cases. *Journal of Medical Genetics*, **1994**, 31, 534-40	5.8	29
264	Pathogenic variants in non-protein-coding sequences. *Clinical Genetics*, **2013**, 84, 422-8	4	28
263	Genomewide linkage scan for split-hand/foot malformation with long-bone deficiency in a large Arab family identifies two novel susceptibility loci on chromosomes 1q42.2-q43 and 6q14.1. *American Journal of Human Genetics*, **2007**, 80, 105-11	11	28

262	The murine orthologue of the Golgi-localized TPTE protein provides clues to the evolutionary history of the human TPTE gene family. *Human Genetics*, **2001**, 109, 569-75	6.3	28
261	Cloning and characterization of a putative human glycerol 3-phosphate permease gene (SLC37A1 or G3PP) on 21q22.3: mutation analysis in two candidate phenotypes, DFNB10 and a glycerol kinase deficiency. *Genomics*, **2000**, 70, 190-200	4.3	28
260	Lack of linkage or association between schizophrenia and the polymorphic trinucleotide repeat within the KCNN3 gene on chromosome 1q21 **1999**, 88, 348-351		28
259	Linkage mapping of the highly informative DNA marker D21S156 to human chromosome 21 using a polymorphic GT dinucleotide repeat. *Genomics*, **1990**, 8, 400-2	4.3	28
258	The gene for autosomal dominant hidrotic ectodermal dysplasia (Clouston syndrome) in a large Indian family maps to the 13q11-q12.1 pericentromeric region. *American Journal of Medical Genetics Part A*, **1997**, 71, 80-6		27
257	Differential gene expression studies to explore the molecular pathophysiology of Down syndrome. *Brain Research Reviews*, **2001**, 36, 265-74		27
256	Chromosome 21: from sequence to applications. *Current Opinion in Genetics and Development*, **2001** , 11, 241-6	4.9	27
255	Slightly deleterious genomic variants and transcriptome perturbations in Down syndrome embryonic selection. *Genome Research*, **2018**, 28, 1-10	9.7	27
254	The effect of genetic variation on promoter usage and enhancer activity. *Nature Communications*, **2017**, 8, 1358	17.4	26
253	Gene age predicts the strength of purifying selection acting on gene expression variation in humans. *American Journal of Human Genetics*, **2014**, 95, 660-74	11	26
252	Subtelomeric 6p deletion: clinical and array-CGH characterization in two patients. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 2094-102	2.5	26
251	Split-hand/split-foot malformation 3 (SHFM3) at 10q24, development of rapid diagnostic methods and gene expression from the region. *American Journal of Medical Genetics, Part A*, **2006**, 140, 1384-95	2.5	26
250	Genomewide scan for nonsyndromic cleft lip and palate in multigenerational Indian families reveals significant evidence of linkage at 13q33.1-34. *American Journal of Human Genetics*, **2006**, 79, 580-5	11	26
249	Arg16 homozygosity of the beta2-adrenergic receptor improves the outcome after beta2-agonist tocolysis for preterm labor. *Clinical Pharmacology and Therapeutics*, **2005**, 78, 656-63	6.1	26
248	Gene finding in the chicken genome. *BMC Bioinformatics*, **2005**, 6, 131	3.6	26
247	The COL6A1 and COL6A2 genes exist as a gene cluster and detect highly informative DNA polymorphisms in the telomeric region of human chromosome 21q. *Human Genetics*, **1991**, 87, 162-6	6.3	26
246	Excess Synaptojanin 1 Contributes to Place Cell Dysfunction and Memory Deficits in the Aging Hippocampus in Three Types of Alzheimer's Disease. *Cell Reports*, **2018**, 23, 2967-2975	10.6	25
245	Mutation nomenclature. *Current Protocols in Human Genetics*, **2003**, Chapter 7, Unit 7.13	3.2	25

244	No deleterious mutations in the FOXJ1 (alias HFH-4) gene in patients with primary ciliary dyskinesia (PCD). *Cytogenetic and Genome Research*, **2000**, 90, 119-22	1.9	25
243	Refined localization of autosomal recessive nonsyndromic deafness DFNB10 locus using 34 novel microsatellite markers, genomic structure, and exclusion of six known genes in the region. *Genomics*, **2000**, 68, 22-9	4.3	25
242	Tandem repeat sequence variation as causative cis-eQTLs for protein-coding gene expression variation: the case of CSTB. *Human Mutation*, **2012**, 33, 1302-9	4.7	24
241	Mapping of small RNAs in the human ENCODE regions. *American Journal of Human Genetics*, **2008**, 82, 971-81	11	24
240	Frequency of replication/transcription errors in (A)/(T) runs of human genes. *Human Genetics*, **2001**, 109, 40-7	6.3	24
239	An autosomal dominant triphalangeal thumb: polysyndactyly syndrome with variable expression in a large Indian family maps to 7q36. *American Journal of Medical Genetics Part A*, **1996**, 66, 209-15		24
238	Complete Sequence of the 22q11.2 Allele in 1,053 Subjects with 22q11.2 Deletion Syndrome Reveals Modifiers of Conotruncal Heart Defects. *American Journal of Human Genetics*, **2020**, 106, 26-40	11	24
237	Opposite phenotypes of muscle strength and locomotor function in mouse models of partial trisomy and monosomy 21 for the proximal Hspa13-App region. *PLoS Genetics*, **2015**, 11, e1005062	6	23
236	Isolation of a human gene (HES1) with homology to an Escherichia coli and a zebrafish protein that maps to chromosome 21q22.3. *Human Genetics*, **1997**, 99, 616-23	6.3	23
235	FOXL2-mutations in blepharophimosis-ptosis-epicanthus inversus syndrome (BPES); challenges for genetic counseling in female patients **2003**, 117A, 143-6		23
234	Genomic structure of a copy of the human TPTE gene which encompasses 87 kb on the short arm of chromosome 21. *Human Genetics*, **2000**, 107, 127-31	6.3	23
233	Young children with Velo-Cardio-Facial syndrome (CATCH-22). Psychological and language phenotypes. *European Child and Adolescent Psychiatry*, **2000**, 9, 109-14	5.5	23
232	Report from the Maryland Epidemiology Schizophrenia Linkage Study: no evidence for linkage between schizophrenia and a number of candidate and other genomic regions using a complex dominant model. *American Journal of Medical Genetics Part A*, **1994**, 54, 345-53		23
231	A 48,XXY,+21 Down syndrome patient with additional paternal X and maternal 21. *Human Genetics*, **1991**, 87, 54-6	6.3	23
230	Restriction site polymorphism in the phosphoglycerate kinase gene on the X chromosome. *Human Genetics*, **1984**, 66, 217-9	6.3	23
229	A systematic enhancer screen using lentivector transgenesis identifies conserved and non-conserved functional elements at the Olig1 and Olig2 locus. *PLoS ONE*, **2010**, 5, e15741	3.7	23
228	The role of biobanking in rare diseases: European consensus expert group report. *Biopreservation and Biobanking*, **2009**, 7, 155-6	2.1	22
227	Evolutionary forces shape the human RFPL1,2,3 genes toward a role in neocortex development. *American Journal of Human Genetics*, **2008**, 83, 208-18	11	22

226	Promoter polymorphisms and allelic imbalance in ABCB1 expression. *Pharmacogenetics and Genomics*, **2007**, 17, 951-9	1.9	22
225	Complex genetic predisposition to cancer in an extended HNPCC family with an ancestral hMLH1 mutation. *Journal of Medical Genetics*, **1996**, 33, 636-40	5.8	22
224	The haemophilia A mutation search test and resource site, home page of the factor VIII mutation database: HAMSTeRS. *Nucleic Acids Research*, **1996**, 24, 100-2	20.1	22
223	Gene defects in beta-thalassemia and their prenatal diagnosis. *Annals of the New York Academy of Sciences*, **1990**, 612, 1-14	6.5	22
222	MspI polymorphism in the 3' flanking region of the human factor VIII gene. *Nucleic Acids Research*, **1987**, 15, 6312	20.1	22
221	De novo duplication of MECP2 in a girl with mental retardation and no obvious dysmorphic features. *Clinical Genetics*, **2010**, 78, 175-80	4	21
220	A spectrum of LMX1B mutations in Nail-Patella syndrome: new point mutations, deletion, and evidence of mosaicism in unaffected parents. *Genetics in Medicine*, **2010**, 12, 431-9	8.1	21
219	Pericentromeric instability and spontaneous emergence of human neoacrocentric and minute chromosomes in the alternative pathway of telomere lengthening. *Cancer Research*, **2008**, 68, 8146-55	10.1	21
218	Islands of euchromatin-like sequence and expressed polymorphic sequences within the short arm of human chromosome 21. *Genome Research*, **2007**, 17, 1690-6	9.7	21
217	Identification of a novel member of the CLIC family, CLIC6, mapping to 21q22.12. *Gene*, **2003**, 320, 31-40	3.8	21
216	The Caenorhabditis elegans ortholog of C21orf80, a potential new protein O-fucosyltransferase, is required for normal development. *Genomics*, **2004**, 84, 320-30	4.3	21
215	The 11 kb FGA deletion responsible for congenital afibrinogenaemia is mediated by a short direct repeat in the fibrinogen gene cluster. *European Journal of Human Genetics*, **1999**, 7, 897-902	5.3	21
214	Severe hemophilia A in a female by cryptic translocation: order and orientation of factor VIII within Xq28. *Genomics*, **1993**, 16, 20-5	4.3	21
213	A method for the extraction of genomic DNA from human brain tissue fixed and stored in formalin for many years. *Acta Neuropathologica*, **1997**, 93, 408-13	14.3	20
212	Analysis of mutations and chromosomal localisation of the gene encoding RFX5, a novel transcription factor affected in major histocompatibility complex class II deficiency. *Human Mutation*, **1997**, 10, 430-5	4.7	20
211	Characterization of a novel gene, C21orf2, on human chromosome 21q22.3 and its exclusion as the APECED gene by mutation analysis. *Genomics*, **1998**, 47, 64-70	4.3	20
210	Restriction endonuclease mapping of six novel deletions of the factor VIII gene in hemophilia A. *Human Genetics*, **1988**, 80, 143-8	6.3	20
209	Macrorestriction mapping of COL4A1 and COL4A2 collagen genes on human chromosome 13q34. *Genomics*, **1988**, 3, 256-63	4.3	20

208	Bi-allelic Loss-of-Function Variants in DNMBP Cause Infantile Cataracts. *American Journal of Human Genetics*, **2018**, 103, 568-578	11	20
207	Perturbations of heart development and function in cardiomyocytes from human embryonic stem cells with trisomy 21. *Stem Cells*, **2015**, 33, 1434-46	5.8	19
206	Recessive thrombocytopenia likely due to a homozygous pathogenic variant in the FYB gene: case report. *BMC Medical Genetics*, **2014**, 15, 135	2.1	19
205	Genomic determinants in the phenotypic variability of Down syndrome. *Progress in Brain Research*, **2012**, 197, 15-28	2.9	19
204	Refinement of the X-linked nonsyndromic high-grade myopia locus MYP1 on Xq28 and exclusion of 13 known positional candidate genes by direct sequencing **2011**, 52, 6814-9		19
203	Frequency of genomic rearrangements involving the SHFM3 locus at chromosome 10q24 in syndromic and non-syndromic split-hand/foot malformation. *American Journal of Medical Genetics, Part A*, **2006**, 140, 1375-83	2.5	19
202	Linearization and purification of BAC DNA for the development of transgenic mice. *Transgenic Research*, **1999**, 8, 147-50	3.3	19
201	Early-onset dementias: clinical, neuropathological and genetic characteristics. *Acta Neuropathologica*, **1996**, 91, 451-65	14.3	19
200	Biparental inheritance of chromosome 21 polymorphic markers indicates that some Robertsonian translocations t(21;21) occur postzygotically. *American Journal of Medical Genetics Part A*, **1994**, 49, 363-8		19
199	Detection of mutations in the factor VIII gene using single-stranded conformational polymorphism (SSCP). *Genomics*, **1992**, 13, 909-11	4.3	19
198	Linkage mapping of the cystathionine beta-synthase (CBS) gene on human chromosome 21 using a DNA polymorphism in the 3' untranslated region. *Human Genetics*, **1993**, 90, 566-8	6.3	19
197	beta-Amyloid gene is not present in three copies in autopsy-validated Alzheimer's disease. *Genomics*, **1987**, 1, 307-12	4.3	19
196	NANOG priming before full reprogramming may generate germ cell tumours. *European Cells and Materials*, **2011**, 22, 258-74; discussio 274	4.3	19
195	Assaying the regulatory potential of mammalian conserved non-coding sequences in human cells. *Genome Biology*, **2008**, 9, R168	18.3	18
194	The human sugar-phosphate/phosphate exchanger family SLC37. *Pflugers Archiv European Journal of Physiology*, **2004**, 447, 780-3	4.6	18
193	A high-resolution physical map of human chromosome 21p using yeast artificial chromosomes. *Genome Research*, **1999**, 9, 1059-73	9.7	18
192	Cloning of the cDNA for the human ATP synthase OSCP subunit (ATP5O) by exon trapping and mapping to chromosome 21q22.1-q22.2. *Genomics*, **1995**, 28, 470-6	4.3	18
191	Cloning of the cDNA for a human homolog of the rat PEP-19 gene and mapping to chromosome 21q22.2-q22.3. *Human Genetics*, **1996**, 98, 672-7	6.3	18

190	Mild hemophilia A resulting from Arg-to-Leu substitution in exon 26 of the factor VIII gene. *Human Genetics*, **1989**, 81, 335-8	6.3	18
189	Mild hemophilia A associated with a cryptic donor splice site mutation in intron 4 of the factor VIII gene. *Genomics*, **1988**, 2, 32-6	4.3	18
188	Galanin pathogenic mutations in temporal lobe epilepsy. *Human Molecular Genetics*, **2015**, 24, 3082-91	5.6	17
187	Localisation of a human homologue of the Drosophila mnb and rat Dyrk genes to chromosome 21q22.2. *Human Genetics*, **1997**, 99, 262-5	6.3	17
186	Characterization of mouse Dactylaplasia mutations: a model for human ectrodactyly SHFM3. *Mammalian Genome*, **2008**, 19, 272-8	3.2	17
185	The epilepsy, the protease inhibitor and the dodecamer: progressive myoclonus epilepsy, cystatin b and a 12-mer repeat expansion. *Cytogenetic and Genome Research*, **2003**, 100, 213-23	1.9	17
184	C21orf5, a novel human chromosome 21 gene, has a Caenorhabditis elegans ortholog (pad-1) required for embryonic patterning. *Genomics*, **2000**, 68, 30-40	4.3	17
183	YAC and cosmid FISH mapping of an unbalanced chromosomal translocation causing partial trisomy 21 and Down syndrome. *Human Genetics*, **1996**, 98, 460-6	6.3	17
182	Duplication and loss of chromosome 21 in two children with Down syndrome and acute leukemia. *American Journal of Medical Genetics Part A*, **1995**, 59, 174-81		17
181	A highly polymorphic locus cloned from the breakpoint of a chromosome 11p13 deletion associated with the WAGR syndrome. *Genomics*, **1989**, 5, 880-93	4.3	17
180	Physical mapping by PFGE localizes the COL3A1 and COL5A2 genes to a 35-kb region on human chromosome 2. *Genomics*, **1990**, 8, 407-10	4.3	17
179	Single cell transcriptome in aneuploidies reveals mechanisms of gene dosage imbalance. *Nature Communications*, **2019**, 10, 4495	17.4	16
178	Biallelic variants in LINGO1 are associated with autosomal recessive intellectual disability, microcephaly, speech and motor delay. *Genetics in Medicine*, **2018**, 20, 778-784	8.1	16
177	Fortuitous detection of uniparental isodisomy of chromosome 6. *Journal of Medical Genetics*, **1997**, 34, 77-8	5.8	16
176	A large family with subtelomeric translocation t(18;21)(q23;q22.1) and molecular breakpoint in the Down syndrome critical region. *Human Genetics*, **1997**, 100, 669-75	6.3	16
175	CNVs and genetic medicine (excitement and consequences of a rediscovery). *Cytogenetic and Genome Research*, **2008**, 123, 7-16	1.9	16
174	Ectrodactyly with aplasia of long bones (OMIM; 119100) in a large inbred Arab family with an apparent autosomal dominant inheritance and reduced penetrance: clinical and genetic analysis. *American Journal of Medical Genetics, Part A*, **2006**, 140, 1440-6	2.5	16
173	Role of the pleckstrin homology domain in intersectin-L Dbl homology domain activation of Cdc42 and signaling. *Biochimica Et Biophysica Acta - Molecular Cell Research*, **2003**, 1640, 61-8	4.9	16

172	Cytogenetic and molecular analysis of a ring (21) in a patient with partial trisomy 21 and megakaryocytic leukemia. *American Journal of Medical Genetics Part A*, **1995**, 57, 527-36		16
171	PvuII polymorphic site upstream to the human ApoCIII gene. *Nucleic Acids Research*, **1986**, 14, 5571	20.1	16
170	Molecular characterization of beta-globin gene mutations in patients with beta-thalassaemia intermedia in south China. *British Journal of Haematology*, **1988**, 70, 357-61	4.5	16
169	Germ-line chromosomal localization of genes in chromosome 11p linkage: parathyroid hormone, beta-globin, c-Ha-ras-1, and insulin. *Somatic Cell and Molecular Genetics*, **1985**, 11, 197-202		16
168	Chromosome conformation capture uncovers potential genome-wide interactions between human conserved non-coding sequences. *PLoS ONE*, **2011**, 6, e17634	3.7	16
167	MBV: a method to solve sample mislabeling and detect technical bias in large combined genotype and sequencing assay datasets. *Bioinformatics*, **2017**, 33, 1895-1897	7.2	15
166	Association between Variants at BCL11A Erythroid-Specific Enhancer and Fetal Hemoglobin Levels among Sickle Cell Disease Patients in Cameroon: Implications for Future Therapeutic Interventions. *OMICS A Journal of Integrative Biology*, **2015**, 19, 627-31	3.8	15
165	The effect of heterogeneous Transcription Start Sites (TSS) on the translatome: implications for the mammalian cellular phenotype. *BMC Genomics*, **2015**, 16, 986	4.5	15
164	What is expanded in progressive myoclonus epilepsy?. *Nature Genetics*, **1997**, 17, 17	36.3	15
163	Localization of a novel human RNA-editing deaminase (hRED2 or ADARB2) to chromosome 10p15. *Human Genetics*, **1997**, 100, 398-400	6.3	15
162	Increased levels of a chromosome 21-encoded tumour invasion and metastasis factor (TIAM1) mRNA in bone marrow of down syndrome children during the acute phase of AML(M7) **1998**, 23, 61-66		15
161	Transcriptional activation by bidirectional RNA polymerase II elongation over a silent promoter. *EMBO Reports*, **2005**, 6, 956-60	6.5	15
160	An alpha satellite DNA polymorphism specific for the centromeric region of chromosome 13. *Genomics*, **1990**, 7, 110-4	4.3	15
159	DNA polymorphic sites in the human ApoAI-CIII-AIV cluster: Taq I and Ava I. *Nucleic Acids Research*, **1986**, 14, 1924	20.1	15
158	Perturbations of genes essential for Müllerian duct and Wolffian duct development in Mayer-Rokitansky-Küster-Hauser syndrome. *American Journal of Human Genetics*, **2021**, 108, 337-345	11	15
157	Visual impairment and progressive phthisis bulbi caused by recessive pathogenic variant in MARK3. *Human Molecular Genetics*, **2018**, 27, 2703-2711	5.6	15
156	Genome-Wide Association Study to Find Modifiers for Tetralogy of Fallot in the 22q11.2 Deletion Syndrome Identifies Variants in the Locus on 5q14.3. *Circulation: Cardiovascular Genetics*, **2017**, 10,		14
155	Cognition and hippocampal plasticity in the mouse is altered by monosomy of a genomic region implicated in Down syndrome. *Genetics*, **2014**, 197, 899-912	4	14

154	Novel homozygous, heterozygous and hemizygous FRMD7 gene mutations segregated in the same consanguineous family with congenital X-linked nystagmus. *European Journal of Human Genetics*, **2012**, 20, 1032-6	5.3	14
153	Autosomal dominant nonsyndromic cleft lip and palate: significant evidence of linkage at 18q21.1. *American Journal of Human Genetics*, **2007**, 81, 180-8	11	14
152	D21S215 is a (GT)n polymorphic marker close to centromeric alphoid sequences on chromosome 21. *Genomics*, **1992**, 13, 1365-7	4.3	14
151	Taurine treatment of retinal degeneration and cardiomyopathy in a consanguineous family with SLC6A6 taurine transporter deficiency. *Human Molecular Genetics*, **2020**, 29, 618-623	5.6	14
150	Congenital Neuronal Ceroid Lipofuscinosis with a Novel CTSD Gene Mutation: A Rare Cause of Neonatal-Onset Neurodegenerative Disorder. *Neuropediatrics*, **2018**, 49, 150-153	1.6	13
149	Pure Progressive Ataxia and Palatal Tremor (PAPT) Associated with a New Polymerase Gamma (POLG) Mutation. *Cerebellum*, **2016**, 15, 829-831	4.3	13
148	BDNF and DYRK1A are variable and inversely correlated in lymphoblastoid cell lines from Down syndrome patients. *Molecular Neurobiology*, **2012**, 46, 297-303	6.2	13
147	Isolation of the human BACH1 transcription regulator gene, which maps to chromosome 21q22.1. *Human Genetics*, **1998**, 102, 282-8	6.3	13
146	Genomic determinants of the efficiency of internal ribosomal entry sites of viral and cellular origin. *Nucleic Acids Research*, **2008**, 36, 6918-25	20.1	13
145	No association between DUP25 and anxiety disorders. *American Journal of Medical Genetics Part A*, **2004**, 128B, 80-3		13
144	Construction of a 2.5-Mb integrated physical and gene map of distal 21q22.3. *Genomics*, **1998**, 49, 1-13	4.3	13
143	Structure of the human CRFB4 gene: Comparison with its IFNAR neighbor. *Journal of Molecular Evolution*, **1995**, 41, 338-344	3.1	13
142	The human lanosterol synthase gene maps to chromosome 21q22.3. *Human Genetics*, **1996**, 97, 620-4	6.3	13
141	Two novel mutations affecting mRNA splicing of the neurofibromatosis type 1 (NF1) gene. *Human Mutation*, **1996**, 7, 172-5	4.7	13
140	Exome sequencing discloses KALRN homozygous variant as likely cause of intellectual disability and short stature in a consanguineous pedigree. *Human Genomics*, **2016**, 10, 26	6.8	12
139	CATCHing putative causative variants in consanguineous families. *BMC Bioinformatics*, **2015**, 16, 310	3.6	12
138	Exome sequencing reveals a mutation in DMP1 in a family with familial sclerosing bone dysplasia. *Bone*, **2014**, 68, 142-5	4.7	12
137	A mouse embryonic stem cell bank for inducible overexpression of human chromosome 21 genes. *Genome Biology*, **2010**, 11, R64	18.3	12

136	Differential rates of frameshift alterations in four repeat sequences of hereditary nonpolyposis colorectal cancer tumors. *Human Genetics*, **2002**, 111, 284-9	6.3	12
135	Genomic structure, sequence, and refined mapping of the human intersectin gene (ITSN), which encompasses 250 kb on chromosome 21q22.1-->q22.2. *Cytogenetic and Genome Research*, **1998**, 83, 218-20	1.9	12
134	No uniparental disomy for chromosome 3 in Brachmann-De Lange syndrome. *American Journal of Medical Genetics Part A*, **1994**, 49, 133-5		12
133	Linkage mapping of D21S171 to the distal long arm of human chromosome 21 using a polymorphic (AC)n dinucleotide repeat. *Human Genetics*, **1991**, 87, 401-4	6.3	12
132	Detection of genomic variation by selection of a 9 mb DNA region and high throughput sequencing. *PLoS ONE*, **2009**, 4, e6659	3.7	11
131	DNA deamination enables direct PCR amplification of the cystatin B (CSTB) gene-associated dodecamer repeat expansion in myoclonus epilepsy type Unverricht-Lundborg. *Human Mutation*, **2003**, 22, 404-8	4.7	11
130	Characterization of cDNA clones containing CCA trinucleotide repeats derived from human brain. *Somatic Cell and Molecular Genetics*, **1995**, 21, 279-84		11
129	Mapping of the gene for the p60 subunit of the human chromatin assembly factor (CAF1A) to the Down syndrome region of chromosome 21. *Genomics*, **1996**, 33, 309-12	4.3	11
128	Cloning the cDNA of human PWP2, which encodes a protein with WD repeats and maps to 21q22.3. *Genomics*, **1996**, 35, 321-7	4.3	11
127	Apolipoprotein A1 Baltimore (Arg10----Leu), a new ApoA1 variant. *Human Genetics*, **1990**, 84, 439-45	6.3	11
126	Molecular heterogeneity of beta-thalassemia in mestizo Mexicans. *Genomics*, **1991**, 11, 474	4.3	11
125	A Case of Wiedemann-Steiner Syndrome Associated with a 46,XY Disorder of Sexual Development and Gonadal Dysgenesis. *Sexual Development*, **2015**, 9, 289-95	1.6	10
124	Human Hemoglobin **2010**, 365-401		10
123	Characterization of an interstitial deletion 6q13-q14.1 in a female with mild mental retardation, language delay and minor dysmorphisms. *European Journal of Medical Genetics*, **2009**, 52, 49-52	2.6	10
122	Immunochemical characterization of a novel mitochondrially located protein encoded by a nuclear gene within the DFNB8/10 critical region on 21q22.3. *Biochemical and Biophysical Research Communications*, **1997**, 238, 806-10	3.4	10
121	Familial translocation t(Y;15)(q12;p11) and de novo deletion of the Prader-Willi syndrome (PWS) critical region on 15q11-q13. *American Journal of Medical Genetics Part A*, **1997**, 70, 222-8		10
120	Comparative gene finding in chicken indicates that we are closing in on the set of multi-exonic widely expressed human genes. *Nucleic Acids Research*, **2005**, 33, 1935-9	20.1	10
119	Localization of a human homolog of the mouse Tiam-1 gene to chromosome 21q22.1. *Genomics*, **1995**, 30, 123-7	4.3	10

118	The gene for human U2 snRNP auxiliary factor small 35-kDa subunit (U2AF1) maps to the progressive myoclonus epilepsy (EPM1) critical region on chromosome 21q22.3. *Genomics*, **1996**, 33, 298-300	4.3	10
117	Dinucleotide repeat (GT)n markers on chromosome 21. *Genomics*, **1992**, 14, 818-9	4.3	10
116	Molecular etiology of factor VIII deficiency in hemophilia A. *Advances in Experimental Medicine and Biology*, **1995**, 386, 19-34	3.6	10
115	The molecular genetics of hemophilia A and B in man. Factor VIII and factor IX deficiency. *Advances in Human Genetics*, **1988**, 17, 27-59		10
114	No evidence for the presence of genetic variants predisposing to psychotic disorders on the non-deleted 22q11.2 allele of VCFS patients. *Translational Psychiatry*, **2017**, 7, e1039	8.6	9
113	Biallelic variants in PSMB1 encoding the proteasome subunit β6 cause impairment of proteasome function, microcephaly, intellectual disability, developmental delay and short stature. *Human Molecular Genetics*, **2020**, 29, 1132-1143	5.6	9
112	Data in brief: Transcriptome analysis of induced pluripotent stem cells from monozygotic twins discordant for trisomy 21. *Genomics Data*, **2014**, 2, 226-9		9
111	Germline PMS2 and somatic POLE exonuclease mutations cause hypermutability of the leading DNA strand in biallelic mismatch repair deficiency syndrome brain tumours. *Journal of Pathology*, **2017**, 243, 331-341	9.4	9
110	Structure of the human Lanosterol synthase gene and its analysis as a candidate for holoprosencephaly (HPE1). *Human Genetics*, **1999**, 105, 489-95	6.3	9
109	A contiguous physical map of the pericentromeric region of chromosome 21q between D21Z1 and D21S13E. *Genomics*, **1993**, 15, 626-30	4.3	9
108	Exon skipping associated with A-->G transition at +4 of the IVS33 splice donor site of the neurofibromatosis type 1 (NF1) gene. *Human Molecular Genetics*, **1994**, 3, 663-5	5.6	9
107	Discussion on mutation nomenclature. *Human Mutation*, **1994**, 4, 166	4.7	9
106	The Mouse Brain Transcriptome by SAGE: Differences in Gene Expression between P30 Brains of the Partial Trisomy 16 Mouse Model of Down Syndrome (Ts65Dn) and Normals. *Genome Research*, **2000**, 10, 2006-2021	9.7	9
105	Biallelic variants in FBXL3 cause intellectual disability, delayed motor development and short stature. *Human Molecular Genetics*, **2019**, 28, 972-979	5.6	9
104	Biallelic loss of function variants in PPP1R21 cause a neurodevelopmental syndrome with impaired endocytic function. *Human Mutation*, **2019**, 40, 267-280	4.7	9
103	Chromosome 21: a small land of fascinating disorders with unknown pathophysiology. *International Journal of Developmental Biology*, **2002**, 46, 89-96	1.9	9
102	Bi-allelic Variants in DYNC1I2 Cause Syndromic Microcephaly with Intellectual Disability, Cerebral Malformations, and Dysmorphic Facial Features. *American Journal of Human Genetics*, **2019**, 104, 1073-1087	11	8
101	Bi-allelic Variants in IQSEC1 Cause Intellectual Disability, Developmental Delay, and Short Stature. *American Journal of Human Genetics*, **2019**, 105, 907-920	11	8

100	Multiplex targeted high-throughput sequencing for Mendelian cardiac disorders. *Clinical Genetics*, **2014**, 85, 365-70	4	8
99	Genome-wide linkage and copy number variation analysis reveals 710 kb duplication on chromosome 1p31.3 responsible for autosomal dominant omphalocele. *Journal of Medical Genetics*, **2012**, 49, 270-6	5.8	8
98	An Xba I polymorphism 3' to the human erythropoietin (EPO) gene. *Nucleic Acids Research*, **1987**, 15, 6768	20.1	8
97	Correction: Passive and active DNA methylation and the interplay with genetic variation in gene regulation. *ELife*,2,	8.9	8
96	De Novo KAT5 Variants Cause a Syndrome with Recognizable Facial Dysmorphisms, Cerebellar Atrophy, Sleep Disturbance, and Epilepsy. *American Journal of Human Genetics*, **2020**, 107, 564-574	11	8
95	Novel NEXMIF pathogenic variant in a boy with severe autistic features, intellectual disability, and epilepsy, and his mildly affected mother. *Journal of Human Genetics*, **2018**, 63, 847-850	4.3	7
94	Translating molecular advances in Down syndrome and Fragile X syndrome into therapies. *European Neuropsychopharmacology*, **2018**, 28, 675-690	1.2	7
93	SERPINI1 pathogenic variants: An emerging cause of childhood-onset progressive myoclonic epilepsy. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2456-2460	2.5	7
92	A de novo 12q13.11 microdeletion in a patient with severe mental retardation, cleft palate, and high myopia. *European Journal of Medical Genetics*, **2011**, 54, 94-6	2.6	7
91	Mapping of the human transcription factor GABPA (E4TF1-60) gene to chromosome 21. *Genomics*, **1995**, 28, 119-22	4.3	7
90	The CEPH consortium linkage map of human chromosome 13. *Genomics*, **1993**, 16, 486-96	4.3	7
89	Localization of the human gene for mu-crystallin to chromosome 16p. *Genomics*, **1992**, 14, 1115-6	4.3	7
88	Linkage mapping of the carbonyl reductase (CBR) gene on human chromosome 21 using a DNA polymorphism in the 3' untranslated region. *Genomics*, **1992**, 13, 447-8	4.3	7
87	Multipoint mapping studies of six loci on chromosome 11. *Human Heredity*, **1987**, 37, 94-101	1.1	7
86	Specific Susceptibility to COVID-19 in Adults with Down Syndrome. *NeuroMolecular Medicine*, **2021**, 23, 561-571	4.6	7
85	Familial epilepsy in Algeria: Clinical features and inheritance profiles. *Seizure: the Journal of the British Epilepsy Association*, **2015**, 31, 12-8	3.2	6
84	Clinical, cytogenetic, and molecular genetic characterization of two unrelated patients with different duplications of 21q. *American Journal of Medical Genetics Part A*, **1990**, 7, 104-9		6
83	Gene for the human transmembrane-type protein tyrosine phosphatase H (PTPRH): genomic structure, fine-mapping and its exclusion as a candidate for Peutz-Jeghers syndrome. *Cytogenetic and Genome Research*, **2001**, 92, 213-6	1.9	6

82	DNA polymorphisms in the 3' untranslated region of genes on human chromosome 21. *Genomics*, **1993**, 15, 98-102	4.3	6
81	D21S210: a highly polymorphic (GT)n marker closely linked to the beta-amyloid protein precursor (APP) gene. *Human Genetics*, **1993**, 91, 87-8	6.3	6
80	Beta-thalassemia in China: a systematic molecular characterization of beta-thalassemia mutations. *Hemoglobin*, **1988**, 12, 621-8	0.6	6
79	The genomic landscape of human cellular circadian variation points to a novel role for the signalosome. *ELife*, **2017**, 6,	8.9	6
78	De Novo and Bi-allelic Pathogenic Variants in NARS1 Cause Neurodevelopmental Delay Due to Toxic Gain-of-Function and Partial Loss-of-Function Effects. *American Journal of Human Genetics*, **2020**, 107, 311-324	11	6
77	Immune Dysregulation and the Increased Risk of Complications and Mortality Following Respiratory Tract Infections in Adults With Down Syndrome. *Frontiers in Immunology*, **2021**, 12, 621440	8.4	6
76	Mutation nomenclature extensions and suggestions to describe complex mutations: A discussion **2000**, 15, 7		6
75	Viva Europa, a Land of Excellence in Research and Innovation for Health and Wellbeing. *Progress in Preventive Medicine (New York, N Y)*, **2017**, 2, e006	0.7	5
74	HSA21 Single-Minded 2 (Sim2) Binding Sites Co-Localize with Super-Enhancers and Pioneer Transcription Factors in Pluripotent Mouse ES Cells. *PLoS ONE*, **2015**, 10, e0126475	3.7	5
73	Human Gene Mutation: Mechanisms and Consequences **2010**, 319-363		5
72	High-throughput sequencing and rare genetic diseases. *Molecular Syndromology*, **2012**, 3, 197-203	1.5	5
71	Clinical, cytogenetic, and molecular evaluation of a patient with partial trisomy 21 (21q11-q22) lacking the classical Down syndrome phenotype. *American Journal of Medical Genetics Part A*, **1990**, 7, 110-4		5
70	Localization of a human homolog of the mouse pericentrin gene (PCNT) to chromosome 21qter. *Genomics*, **1996**, 35, 620-4	4.3	5
69	A novel zinc finger cDNA with a polymorphic pentanucleotide repeat (ATTTT)n maps on human chromosome 19p. *Genomics*, **1993**, 15, 621-5	4.3	5
68	Dinucleotide repeat polymorphism within ERCC5 gene. *Human Molecular Genetics*, **1994**, 3, 214	5.6	5
67	Cloning and linkage mapping of three polymorphic tetranucleotide (TAAA)n repeats on human chromosome 21. *Genomics*, **1992**, 14, 1071-5	4.3	5
66	MspI polymorphic site in intron 22 of the factor VIII gene in the Japanese population. *Human Genetics*, **1990**, 84, 214-5	6.3	5
65	Linkage map on chromosome 21q and the association of a DNA haplotype with a propensity to nondisjunction and trisomy 21. *Annals of the New York Academy of Sciences*, **1985**, 450, 95-107	6.5	5

64	SCN8A heterozygous variants are associated with anoxic-epileptic seizures. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1209-1216	2.5	4
63	Array-CGH analysis in a patient with WAGR syndrome and a reciprocal translocation t(2;11) inherited from the normal father with double translocation. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 2130-3	2.5	4
62	The SH3D1A gene maps to human chromosome 21q22.1-->q22.2. *Cytogenetic and Genome Research* , **1997**, 78, 213-5	1.9	4
61	Welcome to PathoGenetics. *PathoGenetics*, **2008**, 1, 1		4
60	**2001**,		4
59	The 21q22.1 STS marker, VN02 (EST00541 cDNA), is part of the 3' sequence of the human Na+/myo-inositol cotransporter (SLC5A3) gene. *Cytogenetic and Genome Research*, **1996**, 73, 77-8	1.9	4
58	Linkage mapping of the AML1 gene on human chromosome 21 using a DNA polymorphism in the 3' untranslated region. *Genomics*, **1992**, 14, 506-7	4.3	4
57	Extensive natural variation for cellular hydrogen peroxide release is genetically controlled. *PLoS ONE*, **2012**, 7, e43566	3.7	4
56	Genomic, Proteomic and Phenotypic Heterogeneity in HeLa Cells across Laboratories: Implications for Reproducibility of Research Results		4
55	De novo variants in CELF2 that disrupt the nuclear localization signal cause developmental and epileptic encephalopathy. *Human Mutation*, **2021**, 42, 66-76	4.7	4
54	Genomic databases: A WHO affair. *Science*, **2017**, 356, 812-813	33.3	3
53	Kirrel3-Mediated Synapse Formation Is Attenuated by Disease-Associated Missense Variants. *Journal of Neuroscience*, **2020**, 40, 5376-5388	6.6	3
52	Homozygous deletion of a gene-free region of 4p15 in a child with multiple anomalies: could biallelic loss of conserved, non-coding elements lead to a phenotype?. *European Journal of Medical Genetics*, **2012**, 55, 63-6	2.6	3
51	Human Genome Sequence and Variation **2010**, 31-53		3
50	Lessons from the Genome-Wide Association Studies for Complex Multifactorial Disorders and Traits **2010**, 287-297		3
49	Sequence variation in ultraconserved and highly conserved elements does not cause X-linked mental retardation. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 888-90	2.5	3
48	Cytogenetic, Molecular and FISH Analysis of an Isodicentric Chromosome 21 idic(21)(q22.3) in a Mildly-Affected Patient with Down Syndrome. *International Journal of Human Genetics*, **2007**, 7, 215-218	1	3
47	Unbalanced translocation, t(18;21), detected by fluorescence in situ hybridization (FISH) in a child with 18q- syndrome and a ring chromosome 21. *American Journal of Medical Genetics Part A*, **1993**, 46, 647-51		3

46	Two PstI DNA polymorphisms adjacent to the human gene for the interferon-induced p78 protein (MX1 gene). *Nucleic Acids Research*, **1989**, 17, 7546	20.1	3
45	YAC cloning of DNA embedded in an agarose matrix. *Gene Analysis Techniques*, **1990**, 7, 114-8		3
44	History of the methodology of disease gene identification. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3266-3275	2.5	3
43	Karyotypic Flexibility of the Complex Cancer Genome and the Role of Polyploidization in Maintenance of Structural Integrity of Cancer Chromosomes. *Cancers*, **2020**, 12,	6.6	2
42	Transcriptional and post-transcriptional profile of human chromosome 21. *Genome Research*, **2009**, 19, 1471-9	9.7	2
41	Mutations in Human Genetic Disease **2006**,		2
40	CpG Dinucleotides and Human Disorders **2006**,		2
39	A response to Suzuki et al. How pathogenic is the p.D104N/endostatin polymorphic allele of COL18A1 in Knobloch syndrome? *Human Mutation*, **2005**, 25, 316-316	4.7	2
38	Highly polymorphic repeat marker within the beta-amyloid precursor protein gene. *Human Genetics* , **1994**, 93, 85-6	6.3	2
37	A novel form of human polymorphism involving the hDHFR-psi 1 pseudogene identifies three RFLPs. *Nucleic Acids Research*, **1987**, 15, 5501	20.1	2
36	A DNA polymorphism with KpnI of the human liver-type phosphofructokinase (PFKL) gene. *Nucleic Acids Research*, **1988**, 16, 9060	20.1	2
35	Biallelic truncation variants in ATP9A are associated with a novel autosomal recessive neurodevelopmental disorder. *Npj Genomic Medicine*, **2021**, 6, 94	6.2	2
34	Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations. *Genetics in Medicine*, **2021**, 23, 1922-1932	8.1	2
33	Three decades of the Human Genome Organization. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3314-3321	2.5	2
32	Exome sequencing in the diagnosis of an atypical phenotype of infantile hyalinosis. *Middle East Journal of Medical Genetics*, **2015**, 4, 18-23		1
31	Human Gene Mutation in Inherited Disease **2013**, 1-48		1
30	Analysis of the Born in Bradford birth cohort. *Lancet, The*, **2014**, 383, 123	40	1
29	From sequence to functional understanding: the difficult road ahead. *Genome Medicine*, **2011**, 3, 21	14.4	1

28	Variation in novel exons (RACEfrags) of the MECP2 gene in Rett syndrome patients and controls. *Human Mutation*, **2009**, 30, E866-79	4.7	1
27	Eye Gaze During Face Processing in Children and Adolescents With 22q11.2 Deletion Syndrome. *Journal of the American Academy of Child and Adolescent Psychiatry*, **2010**, 49, 665-674	7.2	1
26	Trapping and sequence analysis of 1138 putative exons from human chromosome 18. *Molecular Psychiatry*, **2003**, 8, 619-23	15.1	1
25	An MspI polymorphism at the MX1 locus in 21q22.3. *Nucleic Acids Research*, **1991**, 19, 5451	20.1	1
24	Chromosome 21 genetic linkage data set based on CEPH pedigrees. *Cytogenetic and Genome Research*, **1992**, 59, 86-7	1.9	1
23	The mapping and sequencing of the human genome. *Southern Medical Journal*, **1990**, 83, 876-8	0.6	1
22	PvuII and XhoI/EcoRV polymorphisms adjacent to the alpha A-crystallin (CRYA1) gene on human chromosome 21. *Nucleic Acids Research*, **1990**, 18, 4300	20.1	1
21	Hemophilia A persistence and gene mutational vulnerability. *Hospital Practice (1995)*, **1987**, 22, 93-5, 99-102	2.2	1
20	Mapping by sequence homology. *European Journal of Human Genetics*, **1996**, 4, 247-9	5.3	1
19	Chromosome 21 and Down syndrome: the post-sequence era. *Cold Spring Harbor Symposia on Quantitative Biology*, **2003**, 68, 425-30	3.9	1
18	Trisomic Phase Inference. *Lecture Notes in Computer Science*, **2004**, 1-8	0.9	1
17	PIGG variant pathogenicity assessment reveals characteristic features within 19 families. *Genetics in Medicine*, **2021**, 23, 1873-1881	8.1	1
16	Taurine newborn screening to prevent one form of retinal degeneration and cardiomyopathy. *European Journal of Human Genetics*, **2020**, 28, 1479-1480	5.3	0
15	William Allan Award Introduction: Haig H. Kazazian, Jr.. *American Journal of Human Genetics*, **2009**, 84, 103-104	11	0
14	Two-dimensional electrophoresis southern transfer method for detecting human genome variability using a LINE-1 sequence probe. *Analytical Biochemistry*, **1995**, 227, 319-27	3.1	0
13	2019 William Allan Award. *American Journal of Human Genetics*, **2020**, 106, 289-294	11	0
12	Biallelic variants in TMEM222 cause a new autosomal recessive neurodevelopmental disorder. *Genetics in Medicine*, **2021**, 23, 1246-1254	8.1	0
11	Human Genomic Variants and Inherited Disease: Molecular Mechanisms and Clinical Consequences **2019**, 125-200		0

10	Next generation diagnostics on cardiomyopathy. *Molecular Cytogenetics*, **2014**, 7, I4	2
9	Independent occurrence of the novel Arg2163 to His mutation in the factor VIII gene in three unrelated families with haemophilia A with different phenotypes. *Human Mutation*, **1998**, 11, 334-334	4.7
8	Uniparental Disomy for Individual Human Chromosomes: Review of Cases49-132	
7	Mechanisms Generating Uniparental Disomy and Genomic Imprinting Disorders25-48	
6	Detection of Uniparental Disomy and Imprinting by DNA Analysis13-24	
5	The human genome project and its impact in medicine. *European Review*, **1996**, 4, 415	0.3
4	Polymorphic dinucleotide repeats at the D3S1417, D3S1418 and D12S271 loci. *Human Molecular Genetics*, **1993**, 2, 1325	5.6
3	Approaches to the Diagnosis of Renal Genetic Disorders Using DNA Analysis **1990**, 53-63	
2	Dodecamer Repeat Expansion in Progressive Myoclonus Epilepsy 1 **2006**, 121-141	
1	The human lanosterol synthase gene maps to chromosome 21q22.3. *Human Genetics*, **1996**, 97, 620-624	6.3