Massimo Carella

List of Publications by Year in Descending Order

212 papers	6,201 citations	42 h-index	71 g-index
222 ext. papers	7,160 ext. citations	5.4 avg, IF	4.89 L-index

#	Paper	IF	Citations
212	Pharmacogenomics of Pediatric Cardiac Arrest: Cisplatin Treatment Worsened by a Ryanodine Receptor 2 Gene Mutation. *Neurology International*, **2022**, 12, 80-88	0	1
211	Contribution of ONECUT1 variants to different forms of non-autoimmune diabetes mellitus in Italian patients.. *Acta Diabetologica*, **2022**, 1	3.9	0
210	Gain of function of Malate Dehydrogenase 2 (MDH2) and familial hyperglycemia. *Journal of Clinical Endocrinology and Metabolism*, **2021**,	5.6	1
209	Electrocardiogram in Friedreich's ataxia: A short-term surrogate endpoint for treatment efficacy. *Annals of Noninvasive Electrocardiology*, **2021**, 26, e12813	1.5	1
208	Biallelic variant in cyclin B3 is associated with failure of maternal meiosis II and recurrent digynic triploidy. *Journal of Medical Genetics*, **2021**, 58, 783-788	5.8	1
207	False-positive results of SARS-CoV-2 IgM/IgG antibody tests in sera stored before the 2020 pandemic in Italy. *International Journal of Infectious Diseases*, **2021**, 104, 159-163	10.5	11
206	Mosaic Segmental and Whole-Chromosome Upd(11)mat in Silver-Russell Syndrome. *Genes*, **2021**, 12,	4.2	0
205	A Novel Genetic Variant in the WFS1 Gene in a Patient with Partial Uniparental Mero-Isodisomy of Chromosome 4. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	1
204	Novel Frameshift Mutation in a Patient Affected by a Syndromic Form of Neurodevelopmental Disorder. *Genes*, **2021**, 12,	4.2	0
203	MitImpact 3: modeling the residue interaction network of the Respiratory Chain subunits. *Nucleic Acids Research*, **2021**, 49, D1282-D1288	20.1	4
202	GDF5 mutation case report and a systematic review of molecular and clinical spectrum: Expanding current knowledge on genotype-phenotype correlations. *Bone*, **2021**, 144, 115803	4.7	2
201	Whole Exome Sequencing Reveals a Novel In-Frame Deletion in a Boy with Global Developmental Delay, Absent Speech, Dysmorphic Features, and Cerebral Anomalies. *Genes*, **2021**, 12,	4.2	3
200	Diagnosis of COVID-19 in Patients with Negative Nasopharyngeal Swabs: Reliability of Radiological and Clinical Diagnosis and Accuracy Versus Serology. *Diagnostics*, **2021**, 11,	3.8	3
199	The recurrent SETBP1 c.2608G > A, p.(Gly870Ser) variant in a patient with Schinzel-Giedion syndrome: an illustrative case of the utility of whole exome sequencing in a critically ill neonate. *Italian Journal of Pediatrics*, **2020**, 46, 74	3.2	3
198	A Link between Genetic Disorders and Cellular Impairment, Using Human Induced Pluripotent Stem Cells to Reveal the Functional Consequences of Copy Number Variations in the Central Nervous System-A Close Look at Chromosome 15. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	2
197	Clinical Significance of Circulating miR-1273g-3p and miR-122-5p in Pancreatic Cancer. *Frontiers in Oncology*, **2020**, 10, 44	5.3	12
196	A Private 16q24.2q24.3 Microduplication in a Boy with Intellectual Disability, Speech Delay and Mild Dysmorphic Features. *Genes*, **2020**, 11,	4.2	5

195	Comparison of the Genomic Profile of Cancer Stem Cells and Their Non-Stem Counterpart: The Case of Ovarian Cancer. *Journal of Clinical Medicine*, **2020**, 9,	5.1	6
194	Dissecting molecular mechanisms of resistance to NOTCH1-targeted therapy in T-cell acute lymphoblastic leukemia xenografts. *Haematologica*, **2020**, 105, 1317-1328	6.6	8
193	Compound Phenotype Due to Recessive Variants in and Genes Disclosed by an Integrated Approach of SNP-Array and Whole Exome Sequencing. *Genes*, **2020**, 11,	4.2	1
192	Pulmonary embolism associated with transfusion after severe post-partum haemorrhage: is less more?. *Blood Transfusion*, **2020**, 18, 13-19	3.6	1
191	Pyntacle: a parallel computing-enabled framework for large-scale network biology analysis. *GigaScience*, **2020**, 9,	7.6	4
190	Double missense mutations in cardiac myosin-binding protein C and myopalladin genes: A case report with diffuse coronary disease, complete atrioventricular block, and progression to dilated cardiomyopathy. *Annals of Noninvasive Electrocardiology*, **2020**, 25, e12687	1.5	4
189	Prenatal whole exome sequencing detects a new homozygous fukutin (FKTN) mutation in a fetus with an ultrasound suspicion of familial Dandy-Walker malformation. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1054	2.3	3
188	microRNA-mRNA network model in patients with achalasia. *Neurogastroenterology and Motility*, **2020**, 32, e13764	4	2
187	Mechanisms of pathogenesis of missense mutations on the KDM6A-H3 interaction in type 2 Kabuki Syndrome. *Computational and Structural Biotechnology Journal*, **2020**, 18, 2033-2042	6.8	3
186	ACE2 gene variants may underlie interindividual variability and susceptibility to COVID-19 in the Italian population. *European Journal of Human Genetics*, **2020**, 28, 1602-1614	5.3	132
185	Heterozygous nonsense ARX mutation in a family highlights the complexity of clinical and molecular diagnosis in case of chromosomal and single gene disorder co-inheritance. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1336	2.3	1
184	Early-Onset Diabetes as Risk Factor for Pancreatic Cancer: miRNA Expression Profiling in Plasma Uncovers a Role for miR-20b-5p, miR-29a, and miR-18a-5p in Diabetes of Recent Diagnosis. *Frontiers in Oncology*, **2020**, 10, 1567	5.3	4
183	The presenting symptoms of Lafora Disease: An electroclinical and genetic study in five Apulian (Southern Italy) families. *Seizure: the Journal of the British Epilepsy Association*, **2020**, 83, 145-153	3.2	2
182	Selective demethylation of two CpG sites causes postnatal activation of the Dao gene and consequent removal of D-serine within the mouse cerebellum. *Clinical Epigenetics*, **2019**, 11, 149	7.7	18
181	The levels of the NMDA receptor co-agonist D-serine are reduced in the substantia nigra of MPTP-lesioned macaques and in the cerebrospinal fluid of Parkinson's disease patients. *Scientific Reports*, **2019**, 9, 8898	4.9	18
180	Stemness underpinning all steps of human colorectal cancer defines the core of effective therapeutic strategies. *EBioMedicine*, **2019**, 44, 346-360	8.8	5
179	Genomic inversions and GOLGA core duplicons underlie disease instability at the 15q25 locus. *PLoS Genetics*, **2019**, 15, e1008075	6	8
178	Free d-aspartate triggers NMDA receptor-dependent cell death in primary cortical neurons and perturbs JNK activation, Tau phosphorylation, and protein SUMOylation in the cerebral cortex of mice lacking d-aspartate oxidase activity. *Experimental Neurology*, **2019**, 317, 51-65	5.7	17

177	Overexpression of the cohesin-core subunit SMC1A contributes to colorectal cancer development. *Journal of Experimental and Clinical Cancer Research*, **2019**, 38, 108	12.8	20
176	1q23.1 homozygous deletion and downregulation of Fc receptor-like family genes confer poor prognosis in chronic lymphocytic leukemia. *Clinical and Experimental Medicine*, **2019**, 19, 261-267	4.9	4
175	A novel dominant-negative FGFR1 variant causes Hartsfield syndrome by deregulating RAS/ERK1/2 pathway. *European Journal of Human Genetics*, **2019**, 27, 1113-1120	5.3	10
174	Are Gaming-Enabled Graphic Processing Unit Cards Convenient for Molecular Dynamics Simulation?. *Evolutionary Bioinformatics*, **2019**, 15, 1176934319850144	1.9	9
173	Concurrent chromothripsis events in a case of TP53 depleted acute myeloid leukemia with myelodysplasia-related changes. *Cancer Genetics*, **2019**, 237, 63-68	2.3	2
172	Long QT syndrome in chromosome 7q35q36.3 deletion involving KCNH2 gene: Warning for chlorpheniramine prescription. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e855	2.3	4
171	Novel Variants in Two Italian Patients with Classical-Like Ehlers-Danlos Syndrome. *Genes*, **2019**, 10,	4.2	6
170	The phenotypic variations of multi-locus imprinting disturbances associated with maternal-effect variants of NLRP5 range from overt imprinting disorder to apparently healthy phenotype. *Clinical Epigenetics*, **2019**, 11, 190	7.7	13
169	Sudden death in mild hypertrophic cardiomyopathy with compound DSG2/DSC2/MYH6 mutations: Revisiting phenotype after genetic assessment in a master runner athlete. *Journal of Electrocardiology*, **2019**, 53, 95-99	1.4	7
168	Spontaneous remission in a Diamond-Blackfan anaemia patient due to a revertant uniparental disomy ablating a de novo RPS19 mutation. *British Journal of Haematology*, **2019**, 185, 994-998	4.5	12
167	MYC-containing amplicons in acute myeloid leukemia: genomic structures, evolution, and transcriptional consequences. *Leukemia*, **2018**, 32, 2152-2166	10.7	50
166	Refinement of the critical 7p22.1 deletion region: Haploinsufficiency of ACTB is the cause of the 7p22.1 microdeletion-related developmental disorders. *European Journal of Medical Genetics*, **2018**, 61, 248-252	2.6	6
165	The Hidden Genomic and Transcriptomic Plasticity of Giant Marker Chromosomes in Cancer. *Genetics*, **2018**, 208, 951-961	4	10
164	Insights From Molecular Characterization of Adult Patients of Families With Multigenerational Diabetes. *Diabetes*, **2018**, 67, 137-145	0.9	18
163	Sudden cardiac death in J wave syndrome with short QT associated to a novel mutation in Na 1.8 coding gene SCN10A: First case report for a possible pharmacogenomic role. *Journal of Electrocardiology*, **2018**, 51, 809-813	1.4	6
162	The Emerging Role of Altered d-Aspartate Metabolism in Schizophrenia: New Insights From Preclinical Models and Human Studies. *Frontiers in Psychiatry*, **2018**, 9, 559	5	18
161	Establishment of stable iPS-derived human neural stem cell lines suitable for cell therapies. *Cell Death and Disease*, **2018**, 9, 937	9.8	26
160	A single-center study on 140 patients with cerebral cavernous malformations: 28 new pathogenic variants and functional characterization of a PDCD10 large deletion. *Human Mutation*, **2018**, 39, 1885-1900	4.7	11

159	DNA methylation landscape of the genes regulating D-serine and D-aspartate metabolism in post-mortem brain from controls and subjects with schizophrenia. *Scientific Reports*, **2018**, 8, 10163	4.9	23
158	Clinical and molecular characterization of an emerging chromosome 22q13.31 microdeletion syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 391-398	2.5	9
157	Putative TMPRSS3/GJB2 digenic inheritance of hearing loss detected by targeted resequencing. *Molecular and Cellular Probes*, **2017**, 33, 24-27	3.3	4
156	Clinical and molecular characterization of a second family with the 12q14 microdeletion syndrome and review of the literature. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1922-1930	2.5	6
155	Epigenetically induced ectopic expression of UNCX impairs the proliferation and differentiation of myeloid cells. *Haematologica*, **2017**, 102, 1204-1214	6.6	6
154	Assisted Reproductive Techniques and Risk of Beckwith-Wiedemann Syndrome. *Pediatrics*, **2017**, 140,	7.4	55
153	Wnt5a Drives an Invasive Phenotype in Human Glioblastoma Stem-like Cells. *Cancer Research*, **2017**, 77, 996-1007	10.1	54
152	MYC-containing amplicons in acute myeloid leukemia: genomic structures, evolution, and transcriptional consequences. *Leukemia*, **2017**,	10.7	1
151	The epilepsy phenotype in adult patients with intellectual disability and pathogenic copy number variants. *Seizure: the Journal of the British Epilepsy Association*, **2017**, 53, 86-93	3.2	3
150	Decreased free d-aspartate levels are linked to enhanced d-aspartate oxidase activity in the dorsolateral prefrontal cortex of schizophrenia patients. *NPJ Schizophrenia*, **2017**, 3, 16	5.5	38
149	Addendum: Palmieri, O. et al. Functional Implications of MicroRNAs in Crohn's Disease Revealed by Integrating MicroRNA and Messenger RNA Expression Profiling. Int. J. Mol. Sci. 2017, 18, 1580. *International Journal of Molecular Sciences*, **2017**, 18, 2113	6.3	78
148	Functional Implications of MicroRNAs in Crohn's Disease Revealed by Integrating MicroRNA and Messenger RNA Expression Profiling. *International Journal of Molecular Sciences*, **2017**, 18,	6.3	14
147	Developmental Coordination Disorder in a Patient with Mental Disability and a Mild Phenotype Carrying Terminal 6q26-qter Deletion. *Frontiers in Genetics*, **2017**, 8, 206	4.5	3
146	High-confidence assessment of functional impact of human mitochondrial non-synonymous genome variations by APOGEE. *PLoS Computational Biology*, **2017**, 13, e1005628	5	31
145	TGFbeta and miRNA regulation in familial and sporadic breast cancer. *Oncotarget*, **2017**, 8, 50715-50723	3.3	17
144	A primary tumor gene expression signature identifies a crucial role played by tumor stroma myofibroblasts in lymph node involvement in oral squamous cell carcinoma. *Oncotarget*, **2017**, 8, 104913-104927	3.3	8
143	MicroRNA co-expression networks exhibit increased complexity in pancreatic ductal compared to Vater's papilla adenocarcinoma. *Oncotarget*, **2017**, 8, 105320-105339	3.3	4
142	Microduplication: Clinical and Molecular Characterization of a Further Case and Review of the Literature. *Molecular Syndromology*, **2016**, 7, 282-286	1.5	4

141	Whole exome sequencing and single nucleotide polymorphism array analyses to identify germline alterations in genes associated with testosterone metabolism in a patient with androgen insensitivity syndrome and early-onset colorectal cancer. *Chinese Journal of Cancer*, **2016**, 35, 51		2
140	miRNA profiling in serum and tissue samples to assess noninvasive biomarkers for NSCLC clinical outcome. *Tumor Biology*, **2016**, 37, 5503-13	2.9	14
139	A rare but recurrent t(8;13)(q24;q14) translocation in B-cell chronic lymphocytic leukaemia causing MYC up-regulation and concomitant loss of PVT1, miR-15/16 and DLEU7. *British Journal of Haematology*, **2016**, 172, 296-9	4.5	4
138	Analysis of clock gene-miRNA correlation networks reveals candidate drivers in colorectal cancer. *Oncotarget*, **2016**, 7, 45444-45461	3.3	15
137	Support Vector Machine Based on microRNA Expression Profiles to Predict Histological Origin of Ampullary Carcinoma: Case Report of a Patient Affected From Adenocarcinoma of the Papilla of Vater With Lynch Syndrome. *Pancreas*, **2016**, 45, 626-9	2.6	1
136	Gene expression of muscular and neuronal pathways is cooperatively dysregulated in patients with idiopathic achalasia. *Scientific Reports*, **2016**, 6, 31549	4.9	19
135	Two maternal duplications involving the CDKN1C gene are associated with contrasting growth phenotypes. *Clinical Epigenetics*, **2016**, 8, 69	7.7	6
134	Clinical and molecular characterization of a de novo 19p13.3 microdeletion. *Molecular Cytogenetics*, **2016**, 9, 40	2	4
133	Multifaceted enrichment analysis of RNA-RNA crosstalk reveals cooperating micro-societies in human colorectal cancer. *Nucleic Acids Research*, **2016**, 44, 4025-36	20.1	9
132	Systematic analysis of circadian genes using genome-wide cDNA microarrays in the inflammatory bowel disease transcriptome. *Chronobiology International*, **2015**, 32, 903-16	3.6	31
131	A splicing mutation of the HMGA2 gene is associated with Silver-Russell syndrome phenotype. *Journal of Human Genetics*, **2015**, 60, 287-93	4.3	27
130	Report of a patient and further clinical and molecular characterization of interstitial 4p16.3 microduplication. *Molecular Cytogenetics*, **2015**, 8, 15	2	8
129	Target sequencing approach intended to discover new mutations in non-syndromic intellectual disability. *Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis*, **2015**, 781, 32-6	3.3	6
128	A new case of de novo 6q24.2-q25.2 deletion on paternal chromosome 6 with growth hormone deficiency: a twelve-year follow-up and literature review. *BMC Medical Genetics*, **2015**, 16, 69	2.1	4
127	7q11.23 dosage-dependent dysregulation in human pluripotent stem cells affects transcriptional programs in disease-relevant lineages. *Nature Genetics*, **2015**, 47, 132-41	36.3	83
126	Incomplete penetrance and phenotypic variability of 6q16 deletions including SIM1. *European Journal of Human Genetics*, **2015**, 23, 1010-8	5.3	25
125	Testis development in the absence of SRY: chromosomal rearrangements at SOX9 and SOX3. *European Journal of Human Genetics*, **2015**, 23, 1025-32	5.3	44
124	t(15;21) translocations leading to the concurrent downregulation of RUNX1 and its transcription factor partner genes SIN3A and TCF12 in myeloid disorders. *Molecular Cancer*, **2015**, 14, 211	42.1	9

123	De novo microduplication of CHL1 in a patient with non-syndromic developmental phenotypes. *Molecular Cytogenetics*, **2015**, 8, 66	2	12
122	Neurological features of 14q24-q32 interstitial deletion: report of a new case. *Molecular Cytogenetics*, **2015**, 8, 93	2	4
121	Maternal uniparental isodisomy (iUPD) of chromosome 4 in a subject with mild intellectual disability and speech delay. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 2219-22	2.5	4
120	Genome-wide Pathway Analysis Using Gene Expression Data of Colonic Mucosa in Patients with Inflammatory Bowel Disease. *Inflammatory Bowel Diseases*, **2015**, 21, 1260-8	4.5	14
119	Paternal uniparental disomy chromosome 14-like syndrome due a maternal de novo 160 kb deletion at the 14q32.2 region not encompassing the IG- and the MEG3-DMRs: Patient report and genotype-phenotype correlation. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 3130-8	2.5	12
118	Identification and Clinical Characterization of Adult Patients with Multigenerational Diabetes Mellitus. *PLoS ONE*, **2015**, 10, e0135855	3.7	11
117	Loss-of-Function Mutations in APPL1 in Familial Diabetes Mellitus. *American Journal of Human Genetics*, **2015**, 97, 177-85	11	91
116	Excess of runs of homozygosity is associated with severe cognitive impairment in intellectual disability. *Genetics in Medicine*, **2015**, 17, 396-9	8.1	16
115	Microdeletion of 12q24.31: report of a girl with intellectual disability, stereotypies, seizures and facial dysmorphisms. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 438-44	2.5	26
114	Patient affected by neurofibromatosis type 1 and thyroid C-cell hyperplasia harboring pathogenic germ-line mutations in both NF1 and RET genes. *Gene*, **2014**, 536, 332-5	3.8	16
113	Disruption of the ASTN2/TRIM32 locus at 9q33.1 is a risk factor in males for autism spectrum disorders, ADHD and other neurodevelopmental phenotypes. *Human Molecular Genetics*, **2014**, 23, 2752-68	5.6	104
112	MicroRNA expression profiling in male and female familial breast cancer. *British Journal of Cancer*, **2014**, 111, 2361-8	8.7	14
111	Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization. *Nature Genetics*, **2014**, 46, 826-36	36.3	199
110	Rhodobacter sphaeroides adaptation to high concentrations of cobalt ions requires energetic metabolism changes. *FEMS Microbiology Ecology*, **2014**, 88, 345-57	4.3	20
109	Establishment and genetic characterization of ANGM-CSS, a novel, immortal cell line derived from a human glioblastoma multiforme. *International Journal of Oncology*, **2014**, 44, 717-24	4.4	7
108	Evaluation of genome-wide expression profiles of blood and sputum neutrophils in cystic fibrosis patients before and after antibiotic therapy. *PLoS ONE*, **2014**, 9, e104080	3.7	12
107	A miRNA signature for defining aggressive phenotype and prognosis in gliomas. *PLoS ONE*, **2014**, 9, e108950	3.7	52
106	Low prevalence of HNF1A mutations after molecular screening of multiple MODY genes in 58 Italian families recruited in the pediatric or adult diabetes clinic from a single Italian hospital. *Diabetes Care*, **2014**, 37, e258-60	14.6	19

105	TBR1 is the candidate gene for intellectual disability in patients with a 2q24.2 interstitial deletion. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 828-33	2.5	38
104	Giant breast tumors in a patient with Beckwith-Wiedemann syndrome. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 182-5	2.5	5
103	A novel CISD2 intragenic deletion, optic neuropathy and platelet aggregation defect in Wolfram syndrome type 2. *BMC Medical Genetics*, **2014**, 15, 88	2.1	52
102	Combined microRNA and ER expression: a new classifier for familial and sporadic breast cancer patients. *Journal of Translational Medicine*, **2014**, 12, 319	8.5	8
101	Genomic organization and evolution of double minutes/homogeneously staining regions with MYC amplification in human cancer. *Nucleic Acids Research*, **2014**, 42, 9131-45	20.1	67
100	Smaller and larger deletions of the Williams Beuren syndrome region implicate genes involved in mild facial phenotype, epilepsy and autistic traits. *European Journal of Human Genetics*, **2014**, 22, 64-70	5.3	46
99	Variable phenotype in 17q12 microdeletions: clinical and molecular characterization of a new case. *Gene*, **2014**, 538, 373-8	3.8	21
98	EYA1-related disorders: two clinical cases and a literature review. *International Journal of Pediatric Otorhinolaryngology*, **2014**, 78, 1201-10	1.7	8
97	A de novo 11p13 Microduplication in a Patient with Some Features Invoking Silver-Russell Syndrome. *Molecular Syndromology*, **2014**, 5, 11-8	1.5	6
96	Multiple tumor types including leiomyoma and Wilms tumor in a patient with Gorlin syndrome due to 9q22.3 microdeletion encompassing the PTCH1 and FANC-C loci. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2894-901	2.5	15
95	MODY type 2 P59S GCK mutant: founder effect in South of Italy. *Clinical Genetics*, **2013**, 83, 83-7	4	4
94	8q12.1q12.3 de novo microdeletion involving the CHD7 gene in a patient without the major features of CHARGE syndrome: case report and critical review of the literature. *Gene*, **2013**, 513, 209-13	3.8	8
93	3p14.1 de novo microdeletion involving the FOXP1 gene in an adult patient with autism, severe speech delay and deficit of motor coordination. *Gene*, **2013**, 516, 107-13	3.8	32
92	Paternal deletion of the 11p15.5 centromeric-imprinting control region is associated with alteration of imprinted gene expression and recurrent severe intrauterine growth restriction. *Journal of Medical Genetics*, **2013**, 50, 99-103	5.8	23
91	VHL gene alterations in Italian patients with isolated renal cell carcinomas. *International Journal of Biological Markers*, **2013**, 28, 208-15	2.8	2
90	Differences in gene expression and cytokine release profiles highlight the heterogeneity of distinct subsets of adipose tissue-derived stem cells in the subcutaneous and visceral adipose tissue in humans. *PLoS ONE*, **2013**, 8, e57892	3.7	46
89	Genome-wide analysis of differentially expressed genes and splicing isoforms in clear cell renal cell carcinoma. *PLoS ONE*, **2013**, 8, e78452	3.7	14
88	Identification and functional characterization of three NoLS (nucleolar localisation signals) mutations of the CDC73 gene. *PLoS ONE*, **2013**, 8, e82292	3.7	16

87	A rare S33C mutation of CTNNB1 encoding -catenin in a parathyroid adenoma found in an Italian primary hyperparathyroid cohort. *Endocrine*, **2012**, 41, 152-5	4	13
86	An emerging phenotype of interstitial 15q25.2 microdeletions: clinical report and review. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 3182-9	2.5	12
85	DPM2-CDG: a muscular dystrophy-dystroglycanopathy syndrome with severe epilepsy. *Annals of Neurology*, **2012**, 72, 550-8	9.4	103
84	CASR gene activating mutations in two families with autosomal dominant hypocalcemia. *Molecular Genetics and Metabolism*, **2012**, 107, 548-52	3.7	8
83	CDC73 mutations and parafibromin immunohistochemistry in parathyroid tumors: clinical correlations in a single-centre patient cohort. *Cellular Oncology (Dordrecht)*, **2012**, 35, 411-22	7.2	56
82	A further contribution to the delineation of the 17q21.31 microdeletion syndrome: central nervous involvement in two Italian patients. *European Journal of Medical Genetics*, **2012**, 55, 466-71	2.6	12
81	The expression of leucine-rich repeat gene family members in colorectal cancer. *Experimental Biology and Medicine*, **2012**, 237, 1123-8	3.7	16
80	Altered expression of the clock gene machinery in kidney cancer patients. *Biomedicine and Pharmacotherapy*, **2012**, 66, 175-9	7.5	48
79	Thrombocytopenia-absent-radius syndrome in a child showing a larger 1q21.1 deletion than the one in his healthy mother, and a significant downregulation of the commonly deleted genes. *European Journal of Medical Genetics*, **2012**, 55, 120-3	2.6	8
78	BEAT: Bioinformatics Exon Array Tool to store, analyze and visualize Affymetrix GeneChip Human Exon Array data from disease experiments. *BMC Bioinformatics*, **2012**, 13 Suppl 4, S21	3.6	4
77	Molecular pathways undergoing dramatic transcriptomic changes during tumor development in the human colon. *BMC Cancer*, **2012**, 12, 608	4.8	13
76	Interstitial 16p13.3 microduplication: case report and critical review of genotype-phenotype correlation. *European Journal of Medical Genetics*, **2012**, 55, 747-52	2.6	14
75	Mirna expression profiles identify drivers in colorectal and pancreatic cancers. *PLoS ONE*, **2012**, 7, e33663	3.7	116
74	A novel deletion in 2q24.1q24.2 in a girl with mental retardation and generalized hypotonia: a case report. *Molecular Cytogenetics*, **2012**, 5, 1	2	16
73	A novel microdeletion syndrome at 3q13.31 characterised by developmental delay, postnatal overgrowth, hypoplastic male genitals, and characteristic facial features. *Journal of Medical Genetics*, **2012**, 49, 104-9	5.8	42
72	The KCNQ1OT1 imprinting control region and non-coding RNA: new properties derived from the study of Beckwith-Wiedemann syndrome and Silver-Russell syndrome cases. *Human Molecular Genetics*, **2012**, 21, 10-25	5.6	111
71	Chromosomal 17p13.3 microdeletion unmasking recessive Canavan disease mutation. *Molecular Genetics and Metabolism*, **2011**, 104, 706-7	3.7	3
70	22q11.2 Distal Deletion Syndrome: Description of a New Case with Truncus Arteriosus Type 2 and Review. *Molecular Syndromology*, **2011**, 2, 35-44	1.5	16

69	Mental retardation, congenital heart malformation, and myelodysplasia in a patient with a complex chromosomal rearrangement involving the critical region 21q22. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1697-705	2.5	14
68	Dissecting the mucosal expression of human leucine-rich repeat family genes in inflammatory bowel disease patients. *Inflammatory Bowel Diseases*, **2011**, 17, 1834-5	4.5	1
67	High RAD51 mRNA expression characterize estrogen receptor-positive/progesteron receptor-negative breast cancer and is associated with patient's outcome. *International Journal of Cancer*, **2011**, 129, 536-45	7.5	37
66	Regulation of KEAP1 expression by promoter methylation in malignant gliomas and association with patient's outcome. *Epigenetics*, **2011**, 6, 317-25	5.7	77
65	Clock gene expression levels and relationship with clinical and pathological features in colorectal cancer patients. *Chronobiology International*, **2011**, 28, 841-51	3.6	98
64	Multiple spinal ganglioneuromas in a patient harboring a pathogenic NF1 mutation. *Clinical Genetics*, **2010**, 77, 293-7	4	17
63	Gene amplification as double minutes or homogeneously staining regions in solid tumors: origin and structure. *Genome Research*, **2010**, 20, 1198-206	9.7	132
62	Molecular analysis of the HuD gene in neuroendocrine lung cancers. *Lung Cancer*, **2010**, 67, 69-75	5.9	23
61	Large rearrangements detected by MLPA, point mutations, and survey of the frequency of mutations within the SLC3A1 and SLC7A9 genes in a cohort of 172 cystinuric Italian patients. *Molecular Genetics and Metabolism*, **2010**, 99, 42-52	3.7	28
60	Overlapping genes may control reprogramming of mouse somatic cells into induced pluripotent stem cells (iPSCs) and breast cancer stem cells. *In Silico Biology*, **2010**, 10, 207-21	2	6
59	On the reproducibility of results of pathway analysis in genome-wide expression studies of colorectal cancers. *Journal of Biomedical Informatics*, **2010**, 43, 397-406	10.2	10
58	Constitutional ring chromosome 11 mosaicism in a Wilms tumor patient: Cytogenetic, molecular and clinico-pathological studies. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1756-63	2.5	8
57	Changes in CpG islands promoter methylation patterns during ductal breast carcinoma progression. *Cancer Epidemiology Biomarkers and Prevention*, **2009**, 18, 2694-700	4	69
56	High specificity of quantitative methylation-specific PCR analysis for MGMT promoter hypermethylation detection in gliomas. *Journal of Biomedicine and Biotechnology*, **2009**, 2009, 531692		20
55	Mitochondrial dysregulation and oxidative stress in patients with chronic kidney disease. *BMC Genomics*, **2009**, 10, 388	4.5	160
54	Promoter methylation correlates with reduced NDRG2 expression in advanced colon tumour. *BMC Medical Genomics*, **2009**, 2, 11	3.7	31
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