

Massimo Carella

List of Publications by Citations

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

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	The gene TFR2 is mutated in a new type of haemochromatosis mapping to 7q22. <i>Nature Genetics</i> , 2000 , 25, 14-5	36.3	649
211	Juvenile hemochromatosis locus maps to chromosome 1q. <i>American Journal of Human Genetics</i> , 1999 , 64, 1388-93	11	209
210	Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization. <i>Nature Genetics</i> , 2014 , 46, 826-36	36.3	199
209	MYO6, the human homologue of the gene responsible for deafness in Snell's waltzer mice, is mutated in autosomal dominant nonsyndromic hearing loss. <i>American Journal of Human Genetics</i> , 2001 , 69, 635-40	11	186
208	Mitochondrial dysregulation and oxidative stress in patients with chronic kidney disease. <i>BMC Genomics</i> , 2009 , 10, 388	4.5	160
207	Gene amplification as double minutes or homogeneously staining regions in solid tumors: origin and structure. <i>Genome Research</i> , 2010 , 20, 1198-206	9.7	132
206	ACE2 gene variants may underlie interindividual variability and susceptibility to COVID-19 in the Italian population. <i>European Journal of Human Genetics</i> , 2020 , 28, 1602-1614	5.3	132
205	Mirna expression profiles identify drivers in colorectal and pancreatic cancers. <i>PLoS ONE</i> , 2012 , 7, e33663	3.7	116
204	NF1 gene mutations represent the major molecular event underlying neurofibromatosis-Noonan syndrome. <i>American Journal of Human Genetics</i> , 2005 , 77, 1092-101	11	115
203	Stomatocytic haemolysis and macrothrombocytopenia (Mediterranean stomatocytosis/macrothrombocytopenia) is the haematological presentation of phytosterolaemia. <i>British Journal of Haematology</i> , 2005 , 130, 297-309	4.5	113
202	The KCNQ1OT1 imprinting control region and non-coding RNA: new properties derived from the study of Beckwith-Wiedemann syndrome and Silver-Russell syndrome cases. <i>Human Molecular Genetics</i> , 2012 , 21, 10-25	5.6	111
201	Disruption of the ASTN2/TRIM32 locus at 9q33.1 is a risk factor in males for autism spectrum disorders, ADHD and other neurodevelopmental phenotypes. <i>Human Molecular Genetics</i> , 2014 , 23, 2752-68	5.6	104
200	Intergenerational instability and marked anticipation in SCA-17. <i>Neurology</i> , 2003 , 61, 1441-3	6.5	104
199	DPM2-CDG: a muscular dystrophy-dystroglycanopathy syndrome with severe epilepsy. <i>Annals of Neurology</i> , 2012 , 72, 550-8	9.4	103
198	Clock gene expression levels and relationship with clinical and pathological features in colorectal cancer patients. <i>Chronobiology International</i> , 2011 , 28, 841-51	3.6	98
197	Mitochondrial DNA mutations in patients with postlingual, nonsyndromic hearing impairment. <i>European Journal of Human Genetics</i> , 2005 , 13, 26-33	5.3	96
196	Loss-of-Function Mutations in APPL1 in Familial Diabetes Mellitus. <i>American Journal of Human Genetics</i> , 2015 , 97, 177-85	11	91

195	Recurrent microdeletion at 17q12 as a cause of Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome: two case reports. <i>Orphanet Journal of Rare Diseases</i> , 2009 , 4, 25	4.2	90
194	7q11.23 dosage-dependent dysregulation in human pluripotent stem cells affects transcriptional programs in disease-relevant lineages. <i>Nature Genetics</i> , 2015 , 47, 132-41	36.3	83
193	Clinical and genetic studies in hereditary spastic paraplegia with thin corpus callosum. <i>Neurology</i> , 2004 , 62, 262-8	6.5	80
192	Addendum: Palmieri, O. et al. Functional Implications of MicroRNAs in Crohn's Disease Revealed by Integrating MicroRNA and Messenger RNA Expression Profiling. <i>Int. J. Mol. Sci.</i> 2017, 18, 1580. <i>International Journal of Molecular Sciences</i> , 2017 , 18, 2113	6.3	78
191	Regulation of KEAP1 expression by promoter methylation in malignant gliomas and association with patient's outcome. <i>Epigenetics</i> , 2011 , 6, 317-25	5.7	77
190	Genomewide search for dehydrated hereditary stomatocytosis (hereditary xerocytosis): mapping of locus to chromosome 16 (16q23-qter). <i>American Journal of Human Genetics</i> , 1998 , 63, 810-6	11	70
189	Changes in CpG islands promoter methylation patterns during ductal breast carcinoma progression. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2009 , 18, 2694-700	4	69
188	Genomic organization and evolution of double minutes/homogeneously staining regions with MYC amplification in human cancer. <i>Nucleic Acids Research</i> , 2014 , 42, 9131-45	20.1	67
187	Pathogenetic role of the deafness-related M34T mutation of Cx26. <i>Human Molecular Genetics</i> , 2006 , 15, 2569-87	5.6	66
186	The common -866G/A polymorphism in the promoter region of the UCP-2 gene is associated with reduced risk of type 2 diabetes in Caucasians from Italy. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2005 , 90, 1176-80	5.6	63
185	Molecular and functional analysis of SUMF1 mutations in multiple sulfatase deficiency. <i>Human Mutation</i> , 2004 , 23, 576-81	4.7	57
184	CDC73 mutations and parafibromin immunohistochemistry in parathyroid tumors: clinical correlations in a single-centre patient cohort. <i>Cellular Oncology (Dordrecht)</i> , 2012 , 35, 411-22	7.2	56
183	Assisted Reproductive Techniques and Risk of Beckwith-Wiedemann Syndrome. <i>Pediatrics</i> , 2017 , 140,	7.4	55
182	Wnt5a Drives an Invasive Phenotype in Human Glioblastoma Stem-like Cells. <i>Cancer Research</i> , 2017 , 77, 996-1007	10.1	54
181	A miRNA signature for defining aggressive phenotype and prognosis in gliomas. <i>PLoS ONE</i> , 2014 , 9, e108950	9.7	52
180	A novel CISD2 intragenic deletion, optic neuropathy and platelet aggregation defect in Wolfram syndrome type 2. <i>BMC Medical Genetics</i> , 2014 , 15, 88	2.1	52
179	MYC-containing amplicons in acute myeloid leukemia: genomic structures, evolution, and transcriptional consequences. <i>Leukemia</i> , 2018 , 32, 2152-2166	10.7	50
178	Espin gene (ESPN) mutations associated with autosomal dominant hearing loss cause defects in microvillar elongation or organisation. <i>Journal of Medical Genetics</i> , 2006 , 43, 157-61	5.8	50

177	Altered expression of the clock gene machinery in kidney cancer patients. <i>Biomedicine and Pharmacotherapy</i> , 2012 , 66, 175-9	7.5	48
176	On the statistical assessment of classifiers using DNA microarray data. <i>BMC Bioinformatics</i> , 2006 , 7, 387-396	3.6	48
175	Smaller and larger deletions of the Williams Beuren syndrome region implicate genes involved in mild facial phenotype, epilepsy and autistic traits. <i>European Journal of Human Genetics</i> , 2014 , 22, 64-70	5.3	46
174	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. <i>PLoS ONE</i> , 2013 , 8, e57892	3.7	46
173	Dementia, ataxia, extrapyramidal features, and epilepsy: phenotype spectrum in two Italian families with spinocerebellar ataxia type 17. <i>Neurological Sciences</i> , 2003 , 24, 166-7	3.5	46
172	Genomic instability and increased expression of BUB1B and MAD2L1 genes in ductal breast carcinoma. <i>Cancer Letters</i> , 2007 , 254, 298-307	9.9	45
171	Testis development in the absence of SRY: chromosomal rearrangements at SOX9 and SOX3. <i>European Journal of Human Genetics</i> , 2015 , 23, 1025-32	5.3	44
170	A novel microdeletion syndrome at 3q13.31 characterised by developmental delay, postnatal overgrowth, hypoplastic male genitals, and characteristic facial features. <i>Journal of Medical Genetics</i> , 2012 , 49, 104-9	5.8	42
169	Genetic Heterogeneity of Congenital Dyserythropoietic Anemia Type II. <i>Blood</i> , 1998 , 92, 2593-2594	2.2	39
168	Decreased free d-aspartate levels are linked to enhanced d-aspartate oxidase activity in the dorsolateral prefrontal cortex of schizophrenia patients. <i>NPJ Schizophrenia</i> , 2017 , 3, 16	5.5	38
167	TBR1 is the candidate gene for intellectual disability in patients with a 2q24.2 interstitial deletion. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 828-33	2.5	38
166	High RAD51 mRNA expression characterize estrogen receptor-positive/progesteron receptor-negative breast cancer and is associated with patient's outcome. <i>International Journal of Cancer</i> , 2011 , 129, 536-45	7.5	37
165	GOAL: automated Gene Ontology analysis of expression profiles. <i>Nucleic Acids Research</i> , 2004 , 32, W492-9	20.1	36
164	Hearing loss features in GJB2 biallelic mutations and GJB2/GJB6 digenic inheritance in a large Italian cohort. <i>International Journal of Audiology</i> , 2009 , 48, 12-7	2.6	34
163	3p14.1 de novo microdeletion involving the FOXP1 gene in an adult patient with autism, severe speech delay and deficit of motor coordination. <i>Gene</i> , 2013 , 516, 107-13	3.8	32
162	Systematic analysis of circadian genes using genome-wide cDNA microarrays in the inflammatory bowel disease transcriptome. <i>Chronobiology International</i> , 2015 , 32, 903-16	3.6	31
161	Promoter methylation correlates with reduced NDRG2 expression in advanced colon tumour. <i>BMC Medical Genomics</i> , 2009 , 2, 11	3.7	31
160	GABA (gamma-amino-butyric acid) neurotransmission: identification and fine mapping of the human GABAB receptor gene. <i>Biochemical and Biophysical Research Communications</i> , 1998 , 250, 240-5	3.4	31

159	High-confidence assessment of functional impact of human mitochondrial non-synonymous genome variations by APOGEE. <i>PLoS Computational Biology</i> , 2017 , 13, e1005628	5	31
158	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. <i>Molecular Genetics and Metabolism</i> , 2010 , 99, 42-52	3.7	28
157	A splicing mutation of the HMGA2 gene is associated with Silver-Russell syndrome phenotype. <i>Journal of Human Genetics</i> , 2015 , 60, 287-93	4.3	27
156	Juvenile hemochromatosis locus maps to chromosome 1q in a French Canadian population. <i>European Journal of Human Genetics</i> , 2003 , 11, 585-9	5.3	27
155	Microdeletion of 12q24.31: report of a girl with intellectual disability, stereotypies, seizures and facial dysmorphisms. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 438-44	2.5	26
154	Genetic heterogeneity of FG syndrome: a fourth locus (FGS4) maps to Xp11.4-p11.3 in an Italian family. <i>Human Genetics</i> , 2003 , 112, 124-30	6.3	26
153	Establishment of stable iPS-derived human neural stem cell lines suitable for cell therapies. <i>Cell Death and Disease</i> , 2018 , 9, 937	9.8	26
152	Incomplete penetrance and phenotypic variability of 6q16 deletions including SIM1. <i>European Journal of Human Genetics</i> , 2015 , 23, 1010-8	5.3	25
151	Are MYO1C and MYO1F associated with hearing loss?. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2009 , 1792, 27-32	6.9	24
150	A second locus mapping to 2q35-36 for familial pseudohyperkalaemia. <i>European Journal of Human Genetics</i> , 2004 , 12, 1073-6	5.3	24
149	DHPLC analysis of the MECP2 gene in Italian Rett patients. <i>Human Mutation</i> , 2001 , 18, 132-40	4.7	24
148	Charcot-Marie-Tooth disease type 2C: a distinct genetic entity. Clinical and molecular characterization of the first European family. <i>Neuromuscular Disorders</i> , 2002 , 12, 399-404	2.9	24
147	Paternal deletion of the 11p15.5 centromeric-imprinting control region is associated with alteration of imprinted gene expression and recurrent severe intrauterine growth restriction. <i>Journal of Medical Genetics</i> , 2013 , 50, 99-103	5.8	23
146	Molecular analysis of the HuD gene in neuroendocrine lung cancers. <i>Lung Cancer</i> , 2010 , 67, 69-75	5.9	23
145	DNA methylation landscape of the genes regulating D-serine and D-aspartate metabolism in post-mortem brain from controls and subjects with schizophrenia. <i>Scientific Reports</i> , 2018 , 8, 10163	4.9	23
144	Functional characterization of a novel Cx26 (T55N) mutation associated to non-syndromic hearing loss. <i>Biochemical and Biophysical Research Communications</i> , 2005 , 337, 799-805	3.4	22
143	Variable phenotype in 17q12 microdeletions: clinical and molecular characterization of a new case. <i>Gene</i> , 2014 , 538, 373-8	3.8	21
142	Hereditary hemochromatosis: generation of a transcription map within a refined and extended map of the HLA class I region. <i>Genomics</i> , 1996 , 31, 319-26	4.3	21

141	Overexpression of the cohesin-core subunit SMC1A contributes to colorectal cancer development. <i>Journal of Experimental and Clinical Cancer Research</i> , 2019 , 38, 108	12.8	20
140	Rhodobacter sphaeroides adaptation to high concentrations of cobalt ions requires energetic metabolism changes. <i>FEMS Microbiology Ecology</i> , 2014 , 88, 345-57	4.3	20
139	High specificity of quantitative methylation-specific PCR analysis for MGMT promoter hypermethylation detection in gliomas. <i>Journal of Biomedicine and Biotechnology</i> , 2009 , 2009, 531692		20
138	Statistical assessment of functional categories of genes deregulated in pathological conditions by using microarray data. <i>Bioinformatics</i> , 2007 , 23, 2063-72	7.2	20
137	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. <i>Diabetes Care</i> , 2014 , 37, e258-60	14.6	19
136	Gene expression of muscular and neuronal pathways is cooperatively dysregulated in patients with idiopathic achalasia. <i>Scientific Reports</i> , 2016 , 6, 31549	4.9	19
135	Selective demethylation of two CpG sites causes postnatal activation of the Dao gene and consequent removal of D-serine within the mouse cerebellum. <i>Clinical Epigenetics</i> , 2019 , 11, 149	7.7	18
134	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. <i>Scientific Reports</i> , 2019 , 9, 8898	4.9	18
133	Insights From Molecular Characterization of Adult Patients of Families With Multigenerational Diabetes. <i>Diabetes</i> , 2018 , 67, 137-145	0.9	18
132	Construction of a YAC contig covering human chromosome 6p22. <i>Genomics</i> , 1996 , 36, 399-407	4.3	18
131	The Emerging Role of Altered d-Aspartate Metabolism in Schizophrenia: New Insights From Preclinical Models and Human Studies. <i>Frontiers in Psychiatry</i> , 2018 , 9, 559	5	18
130	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. <i>Experimental Neurology</i> , 2019 , 317, 51-65	5.7	17
129	Multiple spinal ganglioneuromas in a patient harboring a pathogenic NF1 mutation. <i>Clinical Genetics</i> , 2010 , 77, 293-7	4	17
128	Congenital dyserythropoietic anemia type II: exclusion of seven candidate genes. <i>Blood Cells, Molecules, and Diseases</i> , 2003 , 30, 22-9	2.1	17
127	TGFbeta and miRNA regulation in familial and sporadic breast cancer. <i>Oncotarget</i> , 2017 , 8, 50715-50723	3.3	17
126	Patient affected by neurofibromatosis type 1 and thyroid C-cell hyperplasia harboring pathogenic germ-line mutations in both NF1 and RET genes. <i>Gene</i> , 2014 , 536, 332-5	3.8	16
125	Excess of runs of homozygosity is associated with severe cognitive impairment in intellectual disability. <i>Genetics in Medicine</i> , 2015 , 17, 396-9	8.1	16
124	The expression of leucine-rich repeat gene family members in colorectal cancer. <i>Experimental Biology and Medicine</i> , 2012 , 237, 1123-8	3.7	16

123	A novel deletion in 2q24.1q24.2 in a girl with mental retardation and generalized hypotonia: a case report. <i>Molecular Cytogenetics</i> , 2012 , 5, 1	2	16
122	Identification and functional characterization of three NoLS (nucleolar localisation signals) mutations of the CDC73 gene. <i>PLoS ONE</i> , 2013 , 8, e82292	3.7	16
121	22q11.2 Distal Deletion Syndrome: Description of a New Case with Truncus Arteriosus Type 2 and Review. <i>Molecular Syndromology</i> , 2011 , 2, 35-44	1.5	16
120	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. <i>American Journal of Medical Genetics, Part A</i> , 2013 , 161A, 2894-901	2.5	15
119	Analysis of clock gene-miRNA correlation networks reveals candidate drivers in colorectal cancer. <i>Oncotarget</i> , 2016 , 7, 45444-45461	3.3	15
118	miRNA profiling in serum and tissue samples to assess noninvasive biomarkers for NSCLC clinical outcome. <i>Tumor Biology</i> , 2016 , 37, 5503-13	2.9	14
117	MicroRNA expression profiling in male and female familial breast cancer. <i>British Journal of Cancer</i> , 2014 , 111, 2361-8	8.7	14
116	Functional Implications of MicroRNAs in Crohn's Disease Revealed by Integrating MicroRNA and Messenger RNA Expression Profiling. <i>International Journal of Molecular Sciences</i> , 2017 , 18,	6.3	14
115	Genome-wide Pathway Analysis Using Gene Expression Data of Colonic Mucosa in Patients with Inflammatory Bowel Disease. <i>Inflammatory Bowel Diseases</i> , 2015 , 21, 1260-8	4.5	14
114	Interstitial 16p13.3 microduplication: case report and critical review of genotype-phenotype correlation. <i>European Journal of Medical Genetics</i> , 2012 , 55, 747-52	2.6	14
113	Genome-wide analysis of differentially expressed genes and splicing isoforms in clear cell renal cell carcinoma. <i>PLoS ONE</i> , 2013 , 8, e78452	3.7	14
112	Mental retardation, congenital heart malformation, and myelodysplasia in a patient with a complex chromosomal rearrangement involving the critical region 21q22. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 1697-705	2.5	14
111	A novel autosomal dominant non-syndromic deafness locus (DFNA48) maps to 12q13-q14 in a large Italian family. <i>Human Genetics</i> , 2003 , 112, 319-20	6.3	14
110	A rare S33C mutation of CTNNB1 encoding βcatenin in a parathyroid adenoma found in an Italian primary hyperparathyroid cohort. <i>Endocrine</i> , 2012 , 41, 152-5	4	13
109	Molecular pathways undergoing dramatic transcriptomic changes during tumor development in the human colon. <i>BMC Cancer</i> , 2012 , 12, 608	4.8	13
108	Hereditary hemochromatosis: a HpaI polymorphism within the HLA-H gene. <i>Molecular and Cellular Probes</i> , 1997 , 11, 229-30	3.3	13
107	The phenotypic variations of multi-locus imprinting disturbances associated with maternal-effect variants of NLRP5 range from overt imprinting disorder to apparently healthy phenotype. <i>Clinical Epigenetics</i> , 2019 , 11, 190	7.7	13
106	Clinical Significance of Circulating miR-1273g-3p and miR-122-5p in Pancreatic Cancer. <i>Frontiers in Oncology</i> , 2020 , 10, 44	5.3	12

105	De novo microduplication of CHL1 in a patient with non-syndromic developmental phenotypes. <i>Molecular Cytogenetics</i> , 2015 , 8, 66	2	12
104	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. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 3130-8	2.5	12
103	Evaluation of genome-wide expression profiles of blood and sputum neutrophils in cystic fibrosis patients before and after antibiotic therapy. <i>PLoS ONE</i> , 2014 , 9, e104080	3.7	12
102	An emerging phenotype of interstitial 15q25.2 microdeletions: clinical report and review. <i>American Journal of Medical Genetics, Part A</i> , 2012 , 158A, 3182-9	2.5	12
101	A further contribution to the delineation of the 17q21.31 microdeletion syndrome: central nervous involvement in two Italian patients. <i>European Journal of Medical Genetics</i> , 2012 , 55, 466-71	2.6	12
100	Audiometric evaluation of carriers of the connexin 26 mutation 35delG. <i>European Archives of Oto-Rhino-Laryngology</i> , 2005 , 262, 921-4	3.5	12
99	Spontaneous remission in a Diamond-Blackfan anaemia patient due to a revertant uniparental disomy ablating a de novo RPS19 mutation. <i>British Journal of Haematology</i> , 2019 , 185, 994-998	4.5	12
98	Identification and Clinical Characterization of Adult Patients with Multigenerational Diabetes Mellitus. <i>PLoS ONE</i> , 2015 , 10, e0135855	3.7	11
97	Molecular basis of hypoxanthine-guanine phosphoribosyltransferase deficiency in Italian Lesch-Nyhan patients: identification of nine novel mutations. <i>Journal of Inherited Metabolic Disease</i> , 2004 , 27, 767-73	5.4	11
96	False-positive results of SARS-CoV-2 IgM/IgG antibody tests in sera stored before the 2020 pandemic in Italy. <i>International Journal of Infectious Diseases</i> , 2021 , 104, 159-163	10.5	11
95	A single-center study on 140 patients with cerebral cavernous malformations: 28 new pathogenic variants and functional characterization of a PDCD10 large deletion. <i>Human Mutation</i> , 2018 , 39, 1885-1900	4.7	11
94	A novel dominant-negative FGFR1 variant causes Hartsfield syndrome by deregulating RAS/ERK1/2 pathway. <i>European Journal of Human Genetics</i> , 2019 , 27, 1113-1120	5.3	10
93	The Hidden Genomic and Transcriptomic Plasticity of Giant Marker Chromosomes in Cancer. <i>Genetics</i> , 2018 , 208, 951-961	4	10
92	Identification of a novel mutation in the SLC26A4 gene in an Italian with fluctuating sensorineural hearing loss. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2009 , 73, 1458-63	1.7	10
91	On the reproducibility of results of pathway analysis in genome-wide expression studies of colorectal cancers. <i>Journal of Biomedical Informatics</i> , 2010 , 43, 397-406	10.2	10
90	MtDNA mutation associated with mitochondrial dysfunction in megakaryoblastic leukaemic cells. <i>Leukemia</i> , 2008 , 22, 1938-41	10.7	10
89	Molecular detection of neuron-specific ELAV-like-positive cells in the peripheral blood of patients with small-cell lung cancer. <i>Cellular Oncology</i> , 2008 , 30, 291-7		10
88	Are Gaming-Enabled Graphic Processing Unit Cards Convenient for Molecular Dynamics Simulation?. <i>Evolutionary Bioinformatics</i> , 2019 , 15, 1176934319850144	1.9	9

87	t(15;21) translocations leading to the concurrent downregulation of RUNX1 and its transcription factor partner genes SIN3A and TCF12 in myeloid disorders. <i>Molecular Cancer</i> , 2015 , 14, 211	42.1	9
86	Comparison between real-time quantitative PCR detection of HER2 mRNA copy number in peripheral blood and ELISA of serum HER2 protein for determining HER2 status in breast cancer patients. <i>Cellular Oncology</i> , 2009 , 31, 203-11		9
85	Multifaceted enrichment analysis of RNA-RNA crosstalk reveals cooperating micro-societies in human colorectal cancer. <i>Nucleic Acids Research</i> , 2016 , 44, 4025-36	20.1	9
84	Clinical and molecular characterization of an emerging chromosome 22q13.31 microdeletion syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018 , 176, 391-398	2.5	9
83	Genomic inversions and GOLGA core duplicons underlie disease instability at the 15q25 locus. <i>PLoS Genetics</i> , 2019 , 15, e1008075	6	8
82	Report of a patient and further clinical and molecular characterization of interstitial 4p16.3 microduplication. <i>Molecular Cytogenetics</i> , 2015 , 8, 15	2	8
81	Dissecting molecular mechanisms of resistance to NOTCH1-targeted therapy in T-cell acute lymphoblastic leukemia xenografts. <i>Haematologica</i> , 2020 , 105, 1317-1328	6.6	8
80	Combined microRNA and ER expression: a new classifier for familial and sporadic breast cancer patients. <i>Journal of Translational Medicine</i> , 2014 , 12, 319	8.5	8
79	EYA1-related disorders: two clinical cases and a literature review. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2014 , 78, 1201-10	1.7	8
78	CASR gene activating mutations in two families with autosomal dominant hypocalcemia. <i>Molecular Genetics and Metabolism</i> , 2012 , 107, 548-52	3.7	8
77	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. <i>European Journal of Medical Genetics</i> , 2012 , 55, 120-3	2.6	8
76	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. <i>Gene</i> , 2013 , 513, 209-13	3.8	8
75	Constitutional ring chromosome 11 mosaicism in a Wilms tumor patient: Cytogenetic, molecular and clinico-pathological studies. <i>American Journal of Medical Genetics, Part A</i> , 2010 , 152A, 1756-63	2.5	8
74	Development of real-time quantitative reverse transcription-PCR for Her2 detection in peripheral blood from patients with breast cancer. <i>Clinica Chimica Acta</i> , 2007 , 384, 52-6	6.2	8
73	Detection of C282Y and H63D in the HFE gene. <i>Genetic Testing and Molecular Biomarkers</i> , 2000 , 4, 115-20		8
72	A primary tumor gene expression signature identifies a crucial role played by tumor stroma myofibroblasts in lymph node involvement in oral squamous cell carcinoma. <i>Oncotarget</i> , 2017 , 8, 104913-104927	3.3	8
71	Establishment and genetic characterization of ANGM-CSS, a novel, immortal cell line derived from a human glioblastoma multiforme. <i>International Journal of Oncology</i> , 2014 , 44, 717-24	4.4	7
70	Cloning of a new gene (FB19) within HLA class I region. <i>Biochemical and Biophysical Research Communications</i> , 1998 , 250, 555-7	3.4	7

69	A YAC Contig Spanning the Blepharophimosis-Ptoxis- Epicanthus inversus Syndrome and Propionic Acidemia Loci. <i>European Journal of Human Genetics</i> , 1997 , 5, 171-174	5.3	7
68	Sudden death in mild hypertrophic cardiomyopathy with compound DSG2/DSC2/MYH6 mutations: Revisiting phenotype after genetic assessment in a master runner athlete. <i>Journal of Electrocardiology</i> , 2019 , 53, 95-99	1.4	7
67	Clinical and molecular characterization of a second family with the 12q14 microdeletion syndrome and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2017 , 173, 1922-1930	2.5	6
66	Epigenetically induced ectopic expression of UNCX impairs the proliferation and differentiation of myeloid cells. <i>Haematologica</i> , 2017 , 102, 1204-1214	6.6	6
65	Target sequencing approach intended to discover new mutations in non-syndromic intellectual disability. <i>Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis</i> , 2015 , 781, 32-6	3.3	6
64	Comparison of the Genomic Profile of Cancer Stem Cells and Their Non-Stem Counterpart: The Case of Ovarian Cancer. <i>Journal of Clinical Medicine</i> , 2020 , 9,	5.1	6
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