Massimo Carella

List of Publications by Year
in descending order

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219	7,745	53660	69108
papers	citations	45 h-index	77 g-index

222	222	222	15002
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	The gene TFR2 is mutated in a new type of haemochromatosis mapping to 7q22. Nature Genetics, 2000, 25, 14-15.	9.4	751
2	Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization. Nature Genetics, 2014, 46, 826-836.	9.4	281
3	Juvenile Hemochromatosis Locus Maps to Chromosome 1q. American Journal of Human Genetics, 1999, 64, 1388-1393.	2.6	229
4	MYO6, the Human Homologue of the Gene Responsible for Deafness in Snellâ€™s Waltzer Mice, Is Mutated in Autosomal Dominant Nonsyndromic Hearing Loss. American Journal of Human Genetics, 2001, 69, 635-640.	2.6	212
5	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.	1.4	208
6	Mitochondrial dysregulation and oxidative stress in patients with chronic kidney disease. BMC Genomics, 2009, 10, 388.	1.2	202
7	Gene amplification as double minutes or homogeneously staining regions in solid tumors: Origin and structure. Genome Research, 2010, 20, 1198-1206.	2.4	194
8	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, 2752-2768.	1.4	140
9	NF1 Gene Mutations Represent the Major Molecular Event Underlying Neurofibromatosis-Noonan Syndrome. American Journal of Human Genetics, 2005, 77, 1092-1101.	2.6	139
10	Stomatocytic haemolysis and macrothrombocytopenia (Mediterranean) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 387 Td (stomatocytosis/m British Journal of Haematology, 2005, 130, 297-309.	1.2	138
11	Mirna Expression Profiles Identify Drivers in Colorectal and Pancreatic Cancers. PLoS ONE, 2012, 7, e33663.	1.1	138
12	The KCNQ1OT1 imprinting control region and non-coding RNA: new properties derived from the study of Beckwithâ€“Wiedemann syndrome and Silverâ€“Russell syndrome cases. Human Molecular Genetics, 2012, 21, 10-25.	1.4	135
13	Intergenerational instability and marked anticipation in SCA-17. Neurology, 2003, 61, 1441-1443.	1.5	125
14	Clock Gene Expression Levels and Relationship With Clinical and Pathological Features in Colorectal Cancer Patients. Chronobiology International, 2011, 28, 841-851.	0.9	123
15	DPM2â€CDG: A muscular dystrophyâ€“dystroglycanopathy syndrome with severe epilepsy. Annals of Neurology, 2012, 72, 550-558.	2.8	121
16	Loss-of-Function Mutations in APPL1 in Familial Diabetes Mellitus. American Journal of Human Genetics, 2015, 97, 177-185.	2.6	114
17	Mitochondrial DNA mutations in patients with postlingual, nonsyndromic hearing impairment. European Journal of Human Genetics, 2005, 13, 26-33.	1.4	110
18	7q11.23 dosage-dependent dysregulation in human pluripotent stem cells affects transcriptional programs in disease-relevant lineages. Nature Genetics, 2015, 47, 132-141.	9.4	108

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19	Recurrent microdeletion at 17q12 as a cause of Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome: two case reports. Orphanet Journal of Rare Diseases, 2009, 4, 25.	1.2	105
20	Regulation ofKEAP1expression by promoter methylation in malignant gliomas and association with patientâ€™s outcome. Epigenetics, 2011, 6, 317-325.	1.3	94
21	Genomic organization and evolution of double minutes/homogeneously staining regions withMYCamplification in human cancer. Nucleic Acids Research, 2014, 42, 9131-9145.	6.5	91
22	Assisted Reproductive Techniques and Risk of Beckwith-Wiedemann Syndrome. Pediatrics, 2017, 140, .	1.0	87
23	Clinical and genetic studies in hereditary spastic paraplegia with thin corpus callosum. Neurology, 2004, 62, 262-268.	1.5	82
24	Wnt5a Drives an Invasive Phenotype in Human Glioblastoma Stem-like Cells. Cancer Research, 2017, 77, 996-1007.	0.4	75
25	Genomewide Search for Dehydrated Hereditary Stomatocytosis (Hereditary Xerocytosis): Mapping of Locus to Chromosome 16 (16q23-qter). American Journal of Human Genetics, 1998, 63, 810-816.	2.6	73
26	Changes in CpG Islands Promoter Methylation Patterns during Ductal Breast Carcinoma Progression. Cancer Epidemiology Biomarkers and Prevention, 2009, 18, 2694-2700.	1.1	73
27	The Common â^'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. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 1176-1180.	1.8	72
28	Pathogenetic role of the deafness-related M34T mutation of Cx26. Human Molecular Genetics, 2006, 15, 2569-2587.	1.4	71
29	MYC-containing amplicons in acute myeloid leukemia: genomic structures, evolution, and transcriptional consequences. Leukemia, 2018, 32, 2152-2166.	3.3	70
30	CDC73 mutations and parafibromin immunohistochemistry in parathyroid tumors: clinical correlations in a single-centre patient cohort. Cellular Oncology (Dordrecht), 2012, 35, 411-422.	2.1	67
31	Molecular and functional analysis ofSUMF1 mutations in multiple sulfatase deficiency. Human Mutation, 2004, 23, 576-581.	1.1	63
32	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.	1.4	63
33	A MiRNA Signature for Defining Aggressive Phenotype and Prognosis in Gliomas. PLoS ONE, 2014, 9, e108950.	1.1	60
34	Espin gene (ESPN) mutations associated with autosomal dominant hearing loss cause defects in microvillar elongation or organisation. Journal of Medical Genetics, 2005, 43, 157-161.	1.5	59
35	Altered expression of the clock gene machinery in kidney cancer patients. Biomedicine and Pharmacotherapy, 2012, 66, 175-179.	2.5	59
36	A novel CISD2 intragenic deletion, optic neuropathy and platelet aggregation defect in Wolfram syndrome type 2. BMC Medical Genetics, 2014, 15, 88.	2.1	59

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37	Testis development in the absence of SRY: chromosomal rearrangements at SOX9 and SOX3. European Journal of Human Genetics, 2015, 23, 1025-1032.	1.4	59
38	On the statistical assessment of classifiers using DNA microarray data. BMC Bioinformatics, 2006, 7, 387.	1.2	56
39	High-confidence assessment of functional impact of human mitochondrial non-synonymous genome variations by APOGEE. PLoS Computational Biology, 2017, 13, e1005628.	1.5	54
40	TBR1 is the candidate gene for intellectual disability in patients with a 2q24.2 interstitial deletion. American Journal of Medical Genetics, Part A, 2014, 164, 828-833.	0.7	52
41	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.	1.1	51
42	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.	2.0	51
43	Genomic instability and increased expression of BUB1B and MAD2L1 genes in ductal breast carcinoma. Cancer Letters, 2007, 254, 298-307.	3.2	50
44	Systematic analysis of circadian genes using genome-wide cDNA microarrays in the inflammatory bowel disease transcriptome. Chronobiology International, 2015, 32, 903-916.	0.9	50
45	Dementia, ataxia, extrapyramidal features, and epilepsy: phenotype spectrum in two Italian families with spinocerebellar ataxia type 17. Neurological Sciences, 2003, 24, 166-167.	0.9	49
46	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, 104-109.	1.5	46
47	GOAL: automated Gene Ontology analysis of expression profiles. Nucleic Acids Research, 2004, 32, W492-W499.	6.5	40
48	Hearing loss features in GJB2 biallelic mutations and GJB2/GJB6 digenic inheritance in a large Italian cohort. International Journal of Audiology, 2009, 48, 12-17.	0.9	40
49	High RAD51 mRNA expression characterize estrogen receptorâ€positive/progesteron receptorâ€negative breast cancer and is associated with patient's outcome. International Journal of Cancer, 2011, 129, 536-545.	2.3	40
50	Genetic Heterogeneity of Congenital Dyserythropoietic Anemia Type II. Blood, 1998, 92, 2593-2594.	0.6	39
51	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, 107-113.	1.0	38
52	Establishment of stable iPS-derived human neural stem cell lines suitable for cell therapies. Cell Death and Disease, 2018, 9, 937.	2.7	36
53	GABA (Î³-Amino-Butyric Acid) Neurotransmission: Identification and Fine Mapping of the Human GABABReceptor Gene. Biochemical and Biophysical Research Communications, 1998, 250, 240-245.	1.0	35
54	Incomplete penetrance and phenotypic variability of 6q16 deletions including SIM1. European Journal of Human Genetics, 2015, 23, 1010-1018.	1.4	35

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55	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.	0.5	34
56	Overexpression of the cohesin-core subunit SMC1A contributes to colorectal cancer development. Journal of Experimental and Clinical Cancer Research, 2019, 38, 108.	3.5	34
57	A splicing mutation of the HMGA2 gene is associated with Silverâ€“Russell syndrome phenotype. Journal of Human Genetics, 2015, 60, 287-293.	1.1	33
58	Promoter methylation correlates with reduced NDRG2expression in advanced colon tumour. BMC Medical Genomics, 2009, 2, 11.	0.7	32
59	Microdeletion of 12q24.31: Report of a girl with intellectual disability, stereotypies, seizures and facial dysmorphisms. American Journal of Medical Genetics, Part A, 2015, 167, 438-444.	0.7	32
60	Charcotâ€“Marieâ€“Tooth disease type 2C: a distinct genetic entity. Clinical and molecular characterization of the first European family. Neuromuscular Disorders, 2002, 12, 399-404.	0.3	31
61	The Emerging Role of Altered d-Aspartate Metabolism in Schizophrenia: New Insights From Preclinical Models and Human Studies. Frontiers in Psychiatry, 2018, 9, 559.	1.3	31
62	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 Parkinsonâ€™s disease patients. Scientific Reports, 2019, 9, 8898.	1.6	31
63	Genetic heterogeneity of FG syndrome: a fourth locus (FGS4) maps to Xp11.4-p11.3 in an Italian family. Human Genetics, 2003, 112, 124-130.	1.8	30
64	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.	1.5	29
65	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.	1.6	29
66	Juvenile hemochromatosis locus maps to chromosome 1q in a French Canadian population. European Journal of Human Genetics, 2003, 11, 585-589.	1.4	28
67	Are MYO1C and MYO1F associated with hearing loss?. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2009, 1792, 27-32.	1.8	28
68	Variable phenotype in 17q12 microdeletions: Clinical and molecular characterization of a new case. Gene, 2014, 538, 373-378.	1.0	28
69	Rhodobacter sphaeroidesadaptation to high concentrations of cobalt ions requires energetic metabolism changes. FEMS Microbiology Ecology, 2014, 88, 345-357.	1.3	28
70	Functional characterization of a novel Cx26 (T55N) mutation associated to non-syndromic hearing loss. Biochemical and Biophysical Research Communications, 2005, 337, 799-805.	1.0	27
71	Molecular analysis of the HuD gene in neuroendocrine lung cancers. Lung Cancer, 2010, 67, 69-75.	0.9	27
72	22q11.2 Distal Deletion Syndrome: Description of a New Case with Truncus Arteriosus Type 2 and Review. Molecular Syndromology, 2011, 2, 35-44.	0.3	26

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73	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.	1.5	26
74	DHPLC analysis of the MECP2 gene in Italian Rett patients. Human Mutation, 2001, 18, 132-140.	1.1	25
75	A second locus mapping to 2q35â€“36 for familial pseudohyperkalaemia. European Journal of Human Genetics, 2004, 12, 1073-1076.	1.4	25
76	Clinical Significance of Circulating miR-1273g-3p and miR-122-5p in Pancreatic Cancer. Frontiers in Oncology, 2020, 10, 44.	1.3	25
77	Analysis of clock gene-miRNA correlation networks reveals candidate drivers in colorectal cancer. Oncotarget, 2016, 7, 45444-45461.	0.8	25
78	Multiple spinal ganglioneuromas in a patient harboring a pathogenic NF1 mutation. Clinical Genetics, 2010, 77, 293-297.	1.0	24
79	A novel deletion in 2q24.1q24.2 in a girl with mental retardation and generalized hypotonia: a case report. Molecular Cytogenetics, 2012, 5, 1.	0.4	24
80	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.	2.0	24
81	Spontaneous remission in a Diamondâ€Blackfan anaemia patient due to a revertant uniparental disomy ablating a de novo RPS19 mutation. British Journal of Haematology, 2019, 185, 994-998.	1.2	24
82	High Specificity of Quantitative Methylation-Specific PCR Analysis forMGMTPromoter Hypermethylation Detection in Gliomas. Journal of Biomedicine and Biotechnology, 2009, 2009, 1-8.	3.0	23
83	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-e260.	4.3	23
84	Gene expression of muscular and neuronal pathways is cooperatively dysregulated in patients with idiopathic achalasia. Scientific Reports, 2016, 6, 31549.	1.6	23
85	Insights From Molecular Characterization of Adult Patients of Families With Multigenerational Diabetes. Diabetes, 2018, 67, 137-145.	0.3	23
86	Hereditary Hemochromatosis: Generation of a Transcription Map within a Refined and Extended Map of the HLA Class I Region. Genomics, 1996, 31, 319-326.	1.3	22
87	Statistical assessment of functional categories of genes deregulated in pathological conditions by using microarray data. Bioinformatics, 2007, 23, 2063-2072.	1.8	22
88	Genome-wide Pathway Analysis Using Gene Expression Data of Colonic Mucosa in Patients with Inflammatory Bowel Disease. Inflammatory Bowel Diseases, 2015, 21, 1.	0.9	22
89	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.	1.8	22
90	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.	1.8	22

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91	Congenital dyserythropoietic anemia type II: exclusion of seven candidate genes. Blood Cells, Molecules, and Diseases, 2003, 30, 22-29.	0.6	21
92	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, 332-335.	1.0	21
93	MitImpact 3: modeling the residue interaction network of the Respiratory Chain subunits. Nucleic Acids Research, 2021, 49, D1282-D1288.	6.5	21
94	TGFbeta and miRNA regulation in familial and sporadic breast cancer. Oncotarget, 2017, 8, 50715-50723.	0.8	20
95	Construction of a YAC Contig Covering Human Chromosome 6p22. Genomics, 1996, 36, 399-407.	1.3	19
96	Hereditary hemochromatosis: aHpal polymorphism within the HLA-H gene. Molecular and Cellular Probes, 1997, 11, 229-230.	0.9	19
97	Genome-Wide Analysis of Differentially Expressed Genes and Splicing Isoforms in Clear Cell Renal Cell Carcinoma. PLoS ONE, 2013, 8, e78452.	1.1	19
98	De novo microduplication of CHL1 in a patient with non-syndromic developmental phenotypes. Molecular Cytogenetics, 2015, 8, 66.	0.4	19
99	Excess of runs of homozygosity is associated with severe cognitive impairment in intellectual disability. Genetics in Medicine, 2015, 17, 396-399.	1.1	19
100	The expression of leucine-rich repeat gene family members in colorectal cancer. Experimental Biology and Medicine, 2012, 237, 1123-1128.	1.1	18
101	Identification and Functional Characterization of Three NoLS (Nucleolar Localisation Signals) Mutations of the CDC73 Gene. PLoS ONE, 2013, 8, e82292.	1.1	18
102	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, 155, 1697-1705.	0.7	17
103	Interstitial 16p13.3 microduplication: Case report and critical review of genotypeâ€“phenotype correlation. European Journal of Medical Genetics, 2012, 55, 747-752.	0.7	17
104	A rare S33C mutation of CTNNB1 encoding Î²-catenin in a parathyroid adenoma found in an Italian primary hyperparathyroid cohort. Endocrine, 2012, 41, 152-155.	1.1	17
105	Multiple tumor types including leiomyoma and Wilms tumor in a patient with Gorlin syndrome due to 9q22.3 microdeletion encompassing the PTCH1 and FANCâ€€ loci. American Journal of Medical Genetics, Part A, 2013, 161, 2894-2901.	0.7	17
106	Functional Implications of MicroRNAs in Crohnâ€™s Disease Revealed by Integrating MicroRNA and Messenger RNA Expression Profiling. International Journal of Molecular Sciences, 2017, 18, 1580.	1.8	17
107	Genomic inversions and GOLGA core duplicons underlie disease instability at the 15q25 locus. PLoS Genetics, 2019, 15, e1008075.	1.5	17
108	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, 466-471.	0.7	16

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109	Molecular pathways undergoing dramatic transcriptomic changes during tumor development in the human colon. BMC Cancer, 2012, 12, 608.	1.1	16
110	MicroRNA expression profiling in male and female familial breast cancer. British Journal of Cancer, 2014, 111, 2361-2368.	2.9	16
111	miRNA profiling in serum and tissue samples to assess noninvasive biomarkers for NSCLC clinical outcome. Tumor Biology, 2016, 37, 5503-5513.	0.8	16
112	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.	1.1	16
113	A novel autosomal dominant non-syndromic deafness locus (DFNA48) maps to 12q13-q14 in a large Italian family. Human Genetics, 2003, 112, 319-320.	1.8	15
114	Paternal uniparental disomy chromosome 14â€like syndrome due a maternal de novo 160â€‰kb deletion at the 14q32.2 region not encompassing the IGâ€•and the MEG3â€DMRs: Patient report and genotypeâ€“phenotype correlation. American Journal of Medical Genetics, Part A, 2015, 167, 3130-3138.	0.7	15
115	Clinical and molecular characterization of an emerging chromosome 22q13.31 microdeletion syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 391-398.	0.7	15
116	Audiometric evaluation of carriers of the connexin 26 mutation 35delG. European Archives of Oto-Rhino-Laryngology, 2005, 262, 921-924.	0.8	14
117	An emerging phenotype of interstitial 15q25.2 microdeletions: Clinical report and review. American Journal of Medical Genetics, Part A, 2012, 158A, 3182-3189.	0.7	14
118	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.	1.1	14
119	Identification and Clinical Characterization of Adult Patients with Multigenerational Diabetes Mellitus. PLoS ONE, 2015, 10, e0135855.	1.1	14
120	Multifaceted enrichment analysis of RNAâ€“RNA crosstalk reveals cooperating micro-societies in human colorectal cancer. Nucleic Acids Research, 2016, 44, 4025-4036.	6.5	14
121	The Hidden Genomic and Transcriptomic Plasticity of Giant Marker Chromosomes in Cancer. Genetics, 2018, 208, 951-961.	1.2	13
122	Are Gaming-Enabled Graphic Processing Unit Cards Convenient for Molecular Dynamics Simulation?. Evolutionary Bioinformatics, 2019, 15, 117693431985014.	0.6	13
123	Molecular basis of hypoxanthine-guanine phosphoribosyltransferase deficiency in Italian Lesch-Nyhan patients: Identification of nine novel mutations. Journal of Inherited Metabolic Disease, 2004, 27, 767-773.	1.7	12
124	MtDNA mutation associated with mitochondrial dysfunction in megakaryoblastic leukaemic cells. Leukemia, 2008, 22, 1938-1941.	3.3	12
125	On the reproducibility of results of pathway analysis in genome-wide expression studies of colorectal cancers. Journal of Biomedical Informatics, 2010, 43, 397-406.	2.5	12
126	EYA1-related disorders: Two clinical cases and a literature review. International Journal of Pediatric Otorhinolaryngology, 2014, 78, 1201-1210.	0.4	12

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127	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.	7.9	12
128	Report of a patient and further clinical and molecular characterization of interstitial 4p16.3 microduplication. Molecular Cytogenetics, 2015, 8, 15.	0.4	12
129	A novel dominant-negative FGFR1 variant causes Hartsfield syndrome by deregulating RAS/ERK1/2 pathway. European Journal of Human Genetics, 2019, 27, 1113-1120.	1.4	12
130	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.	1.3	12
131	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.	0.8	12
132	Molecular detection of neuron-specific ELAV-like-positive cells in the peripheral blood of patients with small-cell lung cancer. Cellular Oncology, 2008, 30, 291-7.	1.9	12
133	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. Cellular Oncology, 2009, 31, 203-11.	1.9	12
134	Stemness underpinning all steps of human colorectal cancer defines the core of effective therapeutic strategies. EBioMedicine, 2019, 44, 346-360.	2.7	11
135	microRNAâ€mRNA network model in patients with achalasia. Neurogastroenterology and Motility, 2020, 32, e13764.	1.6	11
136	Pyntacle: a parallel computing-enabled framework for large-scale network biology analysis. GigaScience, 2020, 9, .	3.3	11
137	Detection of C282Y and H63D in theHFEGene. Genetic Testing and Molecular Biomarkers, 2000, 4, 115-120.	1.7	10
138	Identification of a novel mutation in the SLC26A4 gene in an Italian with fluctuating sensorineural hearing loss. International Journal of Pediatric Otorhinolaryngology, 2009, 73, 1458-1463.	0.4	10
139	Constitutional ring chromosome 11 mosaicism in a Wilms tumor patient: Cytogenetic, molecular and clinicoâ€pathological studies. American Journal of Medical Genetics, Part A, 2010, 152A, 1756-1763.	0.7	10
140	CASR gene activating mutations in two families with autosomal dominant hypocalcemia. Molecular Genetics and Metabolism, 2012, 107, 548-552.	0.5	10
141	Target sequencing approach intended to discover new mutations in non-syndromic intellectual disability. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2015, 781, 32-36.	0.4	10
142	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.	0.7	10
143	Sudden cardiac death in J wave syndrome with short QT associated to a novel mutation in Nav 1.8 coding gene SCN10A: First case report for a possible pharmacogenomic role. Journal of Electrocardiology, 2018, 51, 809-813.	0.4	10
144	Novel TNXB Variants in Two Italian Patients with Classical-Like Ehlers-Danlos Syndrome. Genes, 2019, 10, 967.	1.0	10

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145	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.	0.4	10
146	A Private 16q24.2q24.3 Microduplication in a Boy with Intellectual Disability, Speech Delay and Mild Dysmorphic Features. Genes, 2020, 11, 707.	1.0	10
147	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, 368.	1.0	10
148	Development of real-time quantitative reverse transcription-PCR for Her2 detection in peripheral blood from patients with breast cancer. Clinica Chimica Acta, 2007, 384, 52-56.	0.5	9
149	Combined microRNA and ER expression: a new classifier for familial and sporadic breast cancer patients. Journal of Translational Medicine, 2014, 12, 319.	1.8	9
150	Two maternal duplications involving the CDKN1C gene are associated with contrasting growth phenotypes. Clinical Epigenetics, 2016, 8, 69.	1.8	9
151	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.	1.9	9
152	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.	1.5	9
153	Dissecting molecular mechanisms of resistance to NOTCH1-targeted therapy in T-cell acute lymphoblastic leukemia xenografts. Haematologica, 2020, 105, 1317-1328.	1.7	9
154	MicroRNA co-expression networks exhibit increased complexity in pancreatic ductal compared to Vater's papilla adenocarcinoma. Oncotarget, 2017, 8, 105320-105339.	0.8	9
155	Cloning of a New Gene (FB19) within HLA Class I Region. Biochemical and Biophysical Research Communications, 1998, 250, 555-557.	1.0	8
156	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, 120-123.	0.7	8
157	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, 209-213.	1.0	8
158	Putative TMPRSS3/GJB2 digenic inheritance of hearing loss detected by targeted resequencing. Molecular and Cellular Probes, 2017, 33, 24-27.	0.9	8
159	Epigenetically induced ectopic expression of UNCX impairs the proliferation and differentiation of myeloid cells. Haematologica, 2017, 102, 1204-1214.	1.7	8
160	Whole Exome Sequencing Reveals a Novel AUTS2 In-Frame Deletion in a Boy with Global Developmental Delay, Absent Speech, Dysmorphic Features, and Cerebral Anomalies. Genes, 2021, 12, 229.	1.0	8
161	A de novo 11p13 Microduplication in a Patient with Some Features Invoking Silver-Russell Syndrome. Molecular Syndromology, 2014, 5, 11-18.	0.3	7
162	Establishment and genetic characterization of ANGM-CSS, a novel, immortal cell line derived from a human glioblastoma multiforme. International Journal of Oncology, 2014, 44, 717-724.	1.4	7

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