

Antonio Pizzuti

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/8734236/publications.pdf>

Version: 2024-02-01

177
papers

12,473
citations

71102

41
h-index

25787

108
g-index

180
all docs

180
docs citations

180
times ranked

13578
citing authors

#	ARTICLE	IF	CITATIONS
1	Identification of a gene (FMR-1) containing a CGG repeat coincident with a breakpoint cluster region exhibiting length variation in fragile X syndrome. <i>Cell</i> , 1991, 65, 905-914.	28.9	3,285
2	Variation of the CGG repeat at the fragile X site results in genetic instability: Resolution of the Sherman paradox. <i>Cell</i> , 1991, 67, 1047-1058.	28.9	2,007
3	Characterization of a murine gene expressed from the inactive X chromosome. <i>Nature</i> , 1991, 351, 325-329.	27.8	527
4	Founder and Recurrent CDH1 Mutations in Families With Hereditary Diffuse Gastric Cancer. <i>JAMA - Journal of the American Medical Association</i> , 2007, 297, 2360.	7.4	394
5	Grouping of Multiple-Lentiginos/LEOPARD and Noonan Syndromes on the PTPN11 Gene. <i>American Journal of Human Genetics</i> , 2002, 71, 389-394.	6.2	380
6	Unravelling the Complexity of T Cell Abnormalities in Common Variable Immunodeficiency. <i>Journal of Immunology</i> , 2007, 178, 3932-3943.	0.8	249
7	TDP-43 and FUS RNA-binding Proteins Bind Distinct Sets of Cytoplasmic Messenger RNAs and Differently Regulate Their Post-transcriptional Fate in Motoneuron-like Cells. <i>Journal of Biological Chemistry</i> , 2012, 287, 15635-15647.	3.4	233
8	A polymorphism (K121Q) of the human glycoprotein PC-1 gene coding region is strongly associated with insulin resistance. <i>Diabetes</i> , 1999, 48, 1881-1884.	0.6	228
9	Additive Effects of Genetic Variation in Dopamine Regulating Genes on Working Memory Cortical Activity in Human Brain. <i>Journal of Neuroscience</i> , 2006, 26, 3918-3922.	3.6	208
10	Germline Missense Mutations Affecting KRAS Isoform B Are Associated with a Severe Noonan Syndrome Phenotype. <i>American Journal of Human Genetics</i> , 2006, 79, 129-135.	6.2	205
11	Survival Motor-Neuron Gene Transcript Analysis in Muscles from Spinal Muscular-Atrophy Patients. <i>Biochemical and Biophysical Research Communications</i> , 1995, 213, 342-348.	2.1	182
12	A Variation in 3' UTR of hPTP1B Increases Specific Gene Expression and Associates with Insulin Resistance. <i>American Journal of Human Genetics</i> , 2002, 70, 806-812.	6.2	179
13	Mutations in KCNH1 and ATP6V1B2 cause Zimmermann-Laband syndrome. <i>Nature Genetics</i> , 2015, 47, 661-667.	21.4	177
14	HLA-DQA1 and HLA-DQB1 in Celiac disease predisposition: practical implications of the HLA molecular typing. <i>Journal of Biomedical Science</i> , 2012, 19, 88.	7.0	170
15	NF1 Gene Mutations Represent the Major Molecular Event Underlying Neurofibromatosis-Noonan Syndrome. <i>American Journal of Human Genetics</i> , 2005, 77, 1092-1101.	6.2	139
16	Mutations of ZFPM2/FOG2 gene in sporadic cases of tetralogy of Fallot. <i>Human Mutation</i> , 2003, 22, 372-377.	2.5	127
17	Assignment of a locus for autosomal dominant idiopathic scoliosis (IS) to human chromosome 17p11. <i>Human Genetics</i> , 2002, 111, 401-404.	3.8	125
18	Loss-of-Function Mutations in APPL1 in Familial Diabetes Mellitus. <i>American Journal of Human Genetics</i> , 2015, 97, 177-185.	6.2	114

#	ARTICLE	IF	CITATIONS
19	SMT3A, a Human Homologue of the <i>S. cerevisiae</i> SMT3 Gene, Maps to Chromosome 21qter and Defines a Novel Gene Family. <i>Genomics</i> , 1997, 40, 362-366.	2.9	112
20	Brain Derived Neurotrophic Factor (BDNF) Expression Is Regulated by MicroRNAs miR-26a and miR-26b Allele-Specific Binding. <i>PLoS ONE</i> , 2011, 6, e28656.	2.5	110
21	Effect of nerve growth factor in adrenal autografts in parkinsonism. <i>Annals of Neurology</i> , 1990, 27, 341-342.	5.3	109
22	A homozygous GJA1 gene mutation causes a Hallermann-Streiff/ODDD spectrum phenotype. <i>Human Mutation</i> , 2004, 23, 286-286.	2.5	97
23	UFD1L, a Developmentally Expressed Ubiquitination Gene, is Deleted in CATCH 22 Syndrome. <i>Human Molecular Genetics</i> , 1997, 6, 259-265.	2.9	85
24	Pentanucleotide repeat length polymorphism at the human CD4 locus. <i>Nucleic Acids Research</i> , 1991, 19, 4791-4791.	14.5	82
25	An ATG Repeat in the 3' Untranslated Region of the Human Resistin Gene Is Associated with a Decreased Risk of Insulin Resistance. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2002, 87, 4403-4406.	3.6	82
26	Synergistic Post-Transcriptional Regulation of the Cystic Fibrosis Transmembrane conductance Regulator (CFTR) by miR-101 and miR-494 Specific Binding. <i>PLoS ONE</i> , 2011, 6, e26601.	2.5	80
27	Role of peroxisome proliferator-activated receptor β in amyloid precursor protein processing and amyloid β -mediated cell death. <i>Biochemical Journal</i> , 2005, 391, 693-698.	3.7	78
28	Mutations of the Nogo-66 receptor (RTN4R) gene in schizophrenia. <i>Human Mutation</i> , 2004, 24, 534-535.	2.5	77
29	High prevalence of epilepsy in a village in the Littoral Province of Cameroon. <i>Epilepsy Research</i> , 2008, 82, 200-210.	1.6	71
30	cDNA characterization and chromosomal mapping of two human homologues of the <i>Drosophila</i> dishevelled polarity gene. <i>Human Molecular Genetics</i> , 1996, 5, 953-958.	2.9	57
31	Epilepsy with auditory features: ALG11 gene mutation suggests a loss-of-function mechanism. <i>Annals of Neurology</i> , 2003, 53, 396-399.	5.3	57
32	Elevated levels of miR-145 correlate with SMAD3 down-regulation in Cystic Fibrosis patients. <i>Journal of Cystic Fibrosis</i> , 2013, 12, 797-802.	0.7	57
33	Deep Sequencing the microRNA profile in rhabdomyosarcoma reveals down-regulation of miR-378 family members. <i>BMC Cancer</i> , 2014, 14, 880.	2.6	56
34	The Q121 PC-1 Variant and Obesity Have Additive and Independent Effects in Causing Insulin Resistance. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2001, 86, 5888-5891.	3.6	53
35	Point mutations and polymorphisms in the human dystrophin gene identified in genomic DNA sequences amplified by multiplex PCR. <i>Human Genetics</i> , 1992, 89, 253-8.	3.8	52
36	Human neuronal cell viability demonstrated in culture after cryopreservation. <i>Brain Research</i> , 1988, 473, 169-174.	2.2	51

#	ARTICLE	IF	CITATIONS
37	A novel PTPN11 gene mutation bridges Noonan syndrome, multiple lentiginos/LEOPARD syndrome and Noonan-like/multiple giant cell lesion syndrome. <i>European Journal of Human Genetics</i> , 2004, 12, 1069-1072.	2.8	51
38	DiGeorge subtypes of nonsyndromic conotruncal defects: evidence against a major role of TBX1 Gene. <i>European Journal of Human Genetics</i> , 2003, 11, 349-351.	2.8	48
39	(CTG) _n Triplet Mutation and Phenotype Manifestations in Myotonic Dystrophy Patients. <i>Biochemical Medicine and Metabolic Biology</i> , 1993, 50, 85-92.	0.7	47
40	Hepatitis G virus infection in hemodialysis patients. <i>Kidney International</i> , 1997, 51, 348-352.	5.2	44
41	mRNA distribution in adult human brain of GRIN2B, a N-methyl-d-aspartate (NMDA) receptor subunit. <i>Neuroscience Letters</i> , 1997, 239, 49-53.	2.1	43
42	Familial blepharospasm is inherited as an autosomal dominant trait and relates to a novel unassigned gene. <i>Movement Disorders</i> , 2003, 18, 207-212.	3.9	43
43	Recessive Inactivating Mutations in TBCK, Encoding a Rab GTPase-Activating Protein, Cause Severe Infantile Syndromic Encephalopathy. <i>American Journal of Human Genetics</i> , 2016, 98, 772-781.	6.2	43
44	Crizotinib-induced antitumour activity in human alveolar rhabdomyosarcoma cells is not solely dependent on ALK and MET inhibition. <i>Journal of Experimental and Clinical Cancer Research</i> , 2015, 34, 112.	8.6	41
45	Clinical, neuropsychological, neurophysiologic, and genetic features of a new Italian pedigree with familial cortical myoclonic tremor with epilepsy. <i>Epilepsia</i> , 2009, 50, 1284-1288.	5.1	40
46	Loss of function of the E3 ubiquitin-protein ligase UBE3B causes Kaufman oculocerebrofacial syndrome. <i>Journal of Medical Genetics</i> , 2013, 50, 493-499.	3.2	40
47	A transposon-like element in the deletion-prone region of the dystrophin gene. <i>Genomics</i> , 1992, 13, 594-600.	2.9	39
48	The Myotonic Dystrophy Gene. <i>Archives of Neurology</i> , 1993, 50, 1173-1179.	4.5	37
49	Genomic structure, promoter characterisation and mutational analysis of the S100A7 gene: exclusion of a candidate for familial psoriasis susceptibility. <i>Human Genetics</i> , 1999, 104, 130-134.	3.8	37
50	DNMT3B <i>in vitro</i> knocking-down is able to reverse embryonal rhabdomyosarcoma cell phenotype through inhibition of proliferation and induction of myogenic differentiation. <i>Oncotarget</i> , 2016, 7, 79342-79356.	1.8	37
51	Mutations of UFD1L Are Not Responsible for the Majority of Cases of DiGeorge Syndrome/Velocardiofacial Syndrome without Deletions within Chromosome 22q11. <i>American Journal of Human Genetics</i> , 1999, 65, 247-249.	6.2	36
52	Clinical features and outcome of familial chronic lymphocytic leukemia. <i>Haematologica</i> , 2006, 91, 1117-20.	3.5	35
53	Single nucleotide polymorphisms in the promoter regions of Foxp3 and ICOSLG genes are associated with Alopecia Areata. <i>Clinical and Experimental Medicine</i> , 2014, 14, 91-97.	3.6	33
54	Narrowing the Duane syndrome critical region at chromosome 8q13 down to 40 kb. <i>European Journal of Human Genetics</i> , 2000, 8, 319-324.	2.8	32

#	ARTICLE	IF	CITATIONS
55	Functional analysis of splicing mutations in exon 7 of NF1 gene. BMC Medical Genetics, 2007, 8, 4.	2.1	32
56	Severe Neuropathy After Diphtheria-Tetanus-Pertussis Vaccination in a Child Carrying a Novel Frame-Shift Mutation in the Small Heat-Shock Protein 27 Gene. Journal of Child Neurology, 2010, 25, 107-109.	1.4	32
57	Clinical and genetic study of two patients with Zimmermann-Rabinovitch syndrome and literature review. European Journal of Medical Genetics, 2013, 56, 570-576.	1.3	32
58	Expression Study of Survival Motor Neuron Gene in Human Fetal Tissues. Biochemical and Molecular Medicine, 1997, 61, 102-106.	1.4	31
59	A peptidase gene in chromosome 8q is disrupted by a balanced translocation in a duane syndrome patient. Investigative Ophthalmology and Visual Science, 2002, 43, 3609-12.	3.3	31
60	Genetic association of HLA-DQB1 and HLA-DRB1 polymorphisms with alopecia areata in the Italian population. British Journal of Dermatology, 2011, 165, 823-827.	1.5	30
61	Novel SMAD4 mutation causing Myhre syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 1835-1840.	1.2	29
62	Pharmacological targeting of the ephrin receptor kinase signalling by GLPG1790 in vitro and in vivo reverts oncophenotype, induces myogenic differentiation and radiosensitizes embryonal rhabdomyosarcoma cells. Journal of Hematology and Oncology, 2017, 10, 161.	17.0	29
63	CRELD1 and GATA4 gene analysis in patients with nonsyndromic atrioventricular canal defects. American Journal of Medical Genetics, Part A, 2005, 139A, 236-238.	1.2	26
64	Neurocognitive effects of methylphenidate on ADHD children with different DAT genotypes: A longitudinal open label trial. European Journal of Paediatric Neurology, 2013, 17, 407-414.	1.6	26
65	Molecular analysis of sarcomeric and non-sarcomeric genes in patients with hypertrophic cardiomyopathy. Gene, 2016, 577, 227-235.	2.2	26
66	Induction of adhesion molecules on human Schwann cells by proinflammatory cytokines, an immunofluorescence study. Journal of the Neurological Sciences, 1999, 170, 124-130.	0.6	25
67	PARP inhibitors affect growth, survival and radiation susceptibility of human alveolar and embryonal rhabdomyosarcoma cell lines. Journal of Cancer Research and Clinical Oncology, 2019, 145, 137-152.	2.5	25
68	A sketch of known and novel MYCN-associated miRNA networks in neuroblastoma. Oncology Reports, 2017, 38, 3-20.	2.6	24
69	Human Elongation Factor EF-1 β : Cloning and Characterization of the EF1 β 5a Gene and Assignment of EF-1 β Isoforms to Chromosomes 2, 5, 15, and X. Biochemical and Biophysical Research Communications, 1993, 197, 154-162.	2.1	23
70	Novel Italian family supports clinical and genetic heterogeneity of primary adult-onset torsion dystonia. Movement Disorders, 2002, 17, 392-397.	3.9	23
71	Cytogenetic mapping of a novel locus for type II Waardenburg syndrome. Human Genetics, 2002, 110, 64-67.	3.8	23
72	Genotype-Phenotype Correlations in Monogenic Parkinson Disease: A Review on Clinical and Molecular Findings. Frontiers in Neurology, 2021, 12, 648588.	2.4	23

#	ARTICLE	IF	CITATIONS
73	Structure and expression of the human ubiquitin fusionâ€“degradation gene (UFD1L). <i>Biochimica Et Biophysica Acta Gene Regulatory Mechanisms</i> , 1998, 1396, 158-162.	2.4	22
74	Isolation and Characterization of a Novel Transcript Embedded within HIRA, a Gene Deleted in DiGeorge Syndrome. <i>Molecular Genetics and Metabolism</i> , 1999, 67, 227-235.	1.1	22
75	Cytotoxic T-lymphocyte antigen 4 (CTLA4) +49AG and CT60 gene polymorphisms in Alopecia Areata: a caseâ€“control association study in the Italian population. <i>Archives of Dermatological Research</i> , 2013, 305, 665-670.	1.9	22
76	Midtrimester isolated short femur and perinatal outcomes: A systematic review and metaâ€“analysis. <i>Acta Obstetricia Et Gynecologica Scandinavica</i> , 2019, 98, 11-17.	2.8	22
77	Susceptibility to ischaemic heart disease: Focusing on genetic variants for ATP-sensitive potassium channel beyond traditional risk factors. <i>European Journal of Preventive Cardiology</i> , 2021, 28, 1495-1500.	1.8	22
78	Quantification of Small Non-Coding RNAs Allows an Accurate Comparison of miRNA Expression Profiles. <i>Journal of Biomedicine and Biotechnology</i> , 2009, 2009, 1-9.	3.0	21
79	Mutational analysis of parkin gene by denaturing high-performance liquid chromatography (DHPLC) in essential tremor. <i>Parkinsonism and Related Disorders</i> , 2004, 10, 357-362.	2.2	20
80	ZFPM2/FOG2 andHEY2 genes analysis in nonsyndromic tricuspid atresia. <i>American Journal of Medical Genetics, Part A</i> , 2005, 133A, 68-70.	1.2	19
81	Defining the clinical-genetic and neuroradiological features in SPG54: description of eight additional cases and nine novel DDHD2 variants. <i>Journal of Neurology</i> , 2019, 266, 2657-2664.	3.6	19
82	The Q121 PC-1 Variant and Obesity Have Additive and Independent Effects in Causing Insulin Resistance. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2001, 86, 5888-5891.	3.6	19
83	Role of fetal MRI in the evaluation of isolated and nonâ€“isolated corpus callosum dysgenesis: results of a crossâ€“sectional study. <i>Prenatal Diagnosis</i> , 2017, 37, 244-252.	2.3	18
84	Clinical and functional characterization of a novel RASopathyâ€“causing <i>SHOC2</i> mutation associated with prenatalâ€“onset hypertrophic cardiomyopathy. <i>Human Mutation</i> , 2019, 40, 1046-1056.	2.5	18
85	When to test fetuses for RASopathies? Proposition from a systematic analysis of 352 multicenter cases and a postnatal cohort. <i>Genetics in Medicine</i> , 2021, 23, 1116-1124.	2.4	17
86	The role of PC-1 and ACE genes in diabetic nephropathy in type 1 diabetic patients: evidence for a polygenic control of kidney disease progression. <i>Nephrology Dialysis Transplantation</i> , 2002, 17, 1402-1407.	0.7	16
87	LEOPARD syndrome: a new polyaneurysm association and an update on the molecular genetics of the disease. <i>Journal of Vascular Surgery</i> , 2004, 39, 897-900.	1.1	16
88	Immunogenetic investigation in vernal keratoconjunctivitis. <i>Pediatric Allergy and Immunology</i> , 2014, 25, 508-510.	2.6	16
89	The Use of Piezosurgery in Cranial Surgery in Children. <i>Journal of Craniofacial Surgery</i> , 2015, 26, 840-842.	0.7	16
90	Unusual association of SCN2A epileptic encephalopathy with severe cortical dysplasia detected by prenatal MRI. <i>European Journal of Paediatric Neurology</i> , 2017, 21, 587-590.	1.6	16

#	ARTICLE	IF	CITATIONS
91	Pfeiffer syndrome: literature review of prenatal sonographic findings and genetic diagnosis. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2017, 30, 2225-2231.	1.5	16
92	Prenatal Exome Sequencing: Background, Current Practice and Future Perspectivesâ”A Systematic Review. <i>Diagnostics</i> , 2021, 11, 224.	2.6	16
93	Genomic Organization, Physical Mapping, and Involvement in Yq Microdeletions of the VCY2 (BPY 2) Gene. <i>Genomics</i> , 2001, 72, 153-157.	2.9	15
94	Nonsyndromic Pulmonary Valve Stenosis and thePTPN11 Gene. <i>American Journal of Medical Genetics Part A</i> , 2003, 116A, 389-390.	2.4	15
95	Case report of adult-onset Allgrove syndrome. <i>Neurological Sciences</i> , 2007, 28, 331-335.	1.9	15
96	Triple A syndrome: A novel compound heterozygous mutation in the AAAS gene in an Italian patient without adrenal insufficiency. <i>Journal of the Neurological Sciences</i> , 2010, 290, 150-152.	0.6	15
97	BET inhibition therapy counteracts cancer cell survival, clonogenic potential and radioresistance mechanisms in rhabdomyosarcoma cells. <i>Cancer Letters</i> , 2020, 479, 71-88.	7.2	15
98	Motor neurone metabolism. <i>Journal of the Neurological Sciences</i> , 1999, 169, 161-169.	0.6	14
99	Familial aggregation of genetically heterogeneous hypertrophic cardiomyopathy: A boy with LEOPARD syndrome due to PTPN11 mutation and his nonsyndromic father lacking PTPN11 mutations. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2004, 70, 95-98.	1.6	14
100	Association of the Matrix Metalloproteinase-3 (MMP-3) Promoter Polymorphism With Celiac Disease in Male Subjects. <i>Human Immunology</i> , 2005, 66, 715-719.	2.4	14
101	Mitochondrial dysfunction as a cause of ALS. <i>Archives Italiennes De Biologie</i> , 2011, 149, 113-9.	0.4	14
102	Postzygotic instability of the myotonic dystrophy p[AGC] _n repeat supported by larger expansions in muscle and reduced amplifications in sperm. <i>Journal of Neurology</i> , 1995, 242, 379-383.	3.6	13
103	LG11 gene mutation screening in sporadic partial epilepsy with auditory features. <i>Journal of Neurology</i> , 2005, 252, 62-66.	3.6	13
104	Clinical lumping and molecular splitting of LEOPARD and NF1/NF1-Noonan syndromes. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 1009-1011.	1.2	13
105	Prenatal diagnosis of proximal focal femoral deficiency: Literature review of prenatal sonographic findings. <i>Journal of Clinical Ultrasound</i> , 2016, 44, 252-259.	0.8	13
106	Proteinâ”protein interaction network analysis applied to DNA copy number profiling suggests new perspectives on the aetiology of Mayerâ”Rokitanskyâ”KÅ¼sterâ”Hauser syndrome. <i>Scientific Reports</i> , 2021, 11, 448.	3.3	13
107	cDNA sequence of human Î²-NGF. <i>Nucleic Acids Research</i> , 1990, 18, 4020-4020.	14.5	12
108	Immunomagnetic isolation of human developing motor neurons. <i>NeuroReport</i> , 1998, 9, 1143-1147.	1.2	12

#	ARTICLE	IF	CITATIONS
109	Detection of β -nerve growth factor mRNA in the human fetal brain. <i>Brain Research</i> , 1990, 518, 337-341.	2.2	11
110	Rapid detection of copy number variations and point mutations in BRCA1/2 genes using a single workflow by ion semiconductor sequencing pipeline. <i>Oncotarget</i> , 2018, 9, 33648-33655.	1.8	11
111	Molecular Approaches in Fetal Malformations, Dynamic Anomalies and Soft Markers: Diagnostic Rates and Challenges—Systematic Review of the Literature and Meta-Analysis. <i>Diagnostics</i> , 2022, 12, 575.	2.6	11
112	Cryopreservation of human fetal adrenal medullary cells. <i>Brain Research</i> , 1988, 454, 383-386.	2.2	10
113	Risk of neural tube defects according to maternal body mass index: a systematic review and meta-analysis. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2022, 35, 7296-7305.	1.5	10
114	Different Expression of the Myotonin Protein Kinase Gene in Discrete Areas of Human Brain. <i>Biochemical and Biophysical Research Communications</i> , 1995, 216, 489-494.	2.1	9
115	A family with autosomal dominant mutilating neuropathy not linked to either Charcot-Marie-Tooth disease type 2B (CMT2B) or hereditary sensory neuropathy type I (HSN I) loci. <i>Neuromuscular Disorders</i> , 2002, 12, 286-291.	0.6	9
116	Two Novel Mutations Affecting Splicing in the IRF6 Gene Associated With van der Woude Syndrome. <i>Journal of Craniofacial Surgery</i> , 2010, 21, 1654-1656.	0.7	9
117	Myoclonic epilepsy, parkinsonism, schizophrenia and left-handedness as common neuropsychiatric features in 22q11.2 deletion syndrome. <i>Journal of Medical Genetics</i> , 2020, 57, 151-159.	3.2	9
118	Primary Cultures of Human Caudate Nucleus. <i>Stereotactic and Functional Neurosurgery</i> , 1988, 51, 10-20.	1.5	8
119	Mapping of the MYCL2 processed gene to Xq22-23 and identification of an additional L MYC-related sequence in Xq27.2. <i>FEBS Letters</i> , 1999, 446, 273-277.	2.8	8
120	Hypertrophic cardiomyopathy and the PTPN11 gene. <i>American Journal of Medical Genetics, Part A</i> , 2005, 136A, 93-94.	1.2	8
121	Genetic variants in adipose triglyceride lipase influence lipid levels in familial combined hyperlipidemia. <i>Atherosclerosis</i> , 2010, 213, 206-211.	0.8	8
122	Familial spinal neurofibromatosis due to a multiexonic NF1 gene deletion. <i>Neurogenetics</i> , 2011, 12, 233-240.	1.4	8
123	Whole exome sequencing in an Italian family with isolated maxillary canine agenesis and canine eruption anomalies. <i>Archives of Oral Biology</i> , 2018, 91, 96-102.	1.8	8
124	Small 7p22.3 microdeletion: Case report of Snx8 haploinsufficiency and neurological findings. <i>European Journal of Medical Genetics</i> , 2020, 63, 103772.	1.3	8
125	Isolation of a New Gene in the Friedreich Ataxia Candidate Region on Human Chromosome 9 by cDNA Direct Selection. <i>Biochemical Medicine and Metabolic Biology</i> , 1994, 52, 115-119.	0.7	7
126	Identification of Multiple Transcribed Sequences from the Spinal Muscular Atrophy Region on Human Chromosome 5. <i>Biochemical and Biophysical Research Communications</i> , 1995, 206, 294-301.	2.1	7

#	ARTICLE	IF	CITATIONS
127	Deletion analysis of the simple tandem repeat loci physically linked to the spinal muscular atrophy locus. <i>Human Mutation</i> , 1996, 7, 198-201.	2.5	7
128	Comparative Analysis of Real-Time Polymerase Chain Reaction Methods to Typing HLA-B*57:01 in HIV-1-Positive Patients. <i>AIDS Research and Human Retroviruses</i> , 2016, 32, 654-657.	1.1	7
129	Clinical and Molecular Spectrum of Myotonia and Periodic Paralysis Associated With Mutations in SCN4A in a Large Cohort of Italian Patients. <i>Frontiers in Neurology</i> , 2020, 11, 646.	2.4	7
130	Recurrent prenatal PIEZO1-related lymphatic dysplasia: Expanding molecular and ultrasound findings. <i>European Journal of Medical Genetics</i> , 2021, 64, 104106.	1.3	7
131	GDF5 mutation case report and a systematic review of molecular and clinical spectrum: Expanding current knowledge on genotype-phenotype correlations. <i>Bone</i> , 2021, 144, 115803.	2.9	7
132	OTX015 Epi-Drug Exerts Antitumor Effects in Ovarian Cancer Cells by Blocking GNL3-Mediated Radioresistance Mechanisms: Cellular, Molecular and Computational Evidence. <i>Cancers</i> , 2021, 13, 1519.	3.7	7
133	Pregnant women's knowledge and behaviour to prevent cytomegalovirus infection: an observational study. <i>Journal of Perinatal Medicine</i> , 2021, 49, 327-332.	1.4	7
134	Deletion analysis of SMN and NAIP genes in spinal muscular atrophy Italian families. , 1996, 19, 378-380.		6
135	Prenatal whole exome sequencing detects a new homozygous fukutin (FKTN) mutation in a fetus with an ultrasound suspicion of familial Dandy-Walker malformation. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1054.	1.2	6
136	TLR4 T399I Polymorphism and Endometriosis in a Cohort of Italian Women. <i>Diagnostics</i> , 2020, 10, 255.	2.6	6
137	miR-125b/NRF2/HO-1 axis is involved in protection against oxidative stress of cystic fibrosis: A pilot study. <i>Experimental and Therapeutic Medicine</i> , 2021, 21, 585.	1.8	6
138	Chapter 76 Adrenal medulla autograft in 3 parkinsonian patients: results using two different approaches. <i>Progress in Brain Research</i> , 1990, 82, 677-682.	1.4	5
139	Fetal tongue posture associated with micrognathia: An ultrasound marker of cleft secondary palate?. <i>Journal of Clinical Ultrasound</i> , 2020, 48, 48-51.	0.8	5
140	Beyond BRCA1 and BRCA2: Deleterious Variants in DNA Repair Pathway Genes in Italian Families with Breast/Ovarian and Pancreatic Cancers. <i>Journal of Clinical Medicine</i> , 2020, 9, 3003.	2.4	5
141	Leiomyosarcoma of the Larynx: Case Report with Pathologic and Surgical Considerations. <i>The Journal of Otolaryngology</i> , 2002, 31, 393.	0.6	5
142	Update in non-invasive prenatal testing. <i>Minerva Ginecologica</i> , 2019, 71, 44-53.	0.8	5
143	Human Fetal Adrenal Medulla for Transplantation in Parkinsonian Patients. <i>Annals of the New York Academy of Sciences</i> , 1987, 495, 771-773.	3.8	4
144	Human developing motor neurons as a tool to study ALS. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders: Official Publication of the World Federation of Neurology, Research Group on Motor Neuron Diseases</i> , 2001, 2, 69-76.	1.2	4

#	ARTICLE	IF	CITATIONS
145	Additional evidence that PTPN11 mutations play only a minor role in the pathogenesis of non-syndromic atrioventricular canal defect. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 1970-1972.	1.2	4
146	Early ultrasound suspect of thanatophoric dysplasia followed by first trimester molecular diagnosis. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1756-1758.	1.2	4
147	Heterozygous nonsense <i>ARX</i> mutation in a family highlights the complexity of clinical and molecular diagnosis in case of chromosomal and single gene disorder coinheritance. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1336.	1.2	4
148	Altered Expression of Candidate Genes in Mayer-Rokitansky-Kuster-Hauser Syndrome May Influence Vaginal Keratinocytes Biology: A Focus on Protein Kinase X. <i>Biology</i> , 2021, 10, 450.	2.8	4
149	Potassium Channel KCNH1 Activating Variants Cause Altered Functional and Morphological Ciliogenesis. <i>Molecular Neurobiology</i> , 2022, 59, 4825-4838.	4.0	4
150	Chapter 71 Characterization of purified populations of human fetal chromaffin cells: considerations for grafting in parkinsonian patients. <i>Progress in Brain Research</i> , 1988, 78, 551-557.	1.4	3
151	Lack of pathogenic mutations in <i>SOS1</i> gene in phenytoin-induced gingival overgrowth patients. <i>Archives of Oral Biology</i> , 2017, 80, 160-163.	1.8	3
152	Molecular Analysis of PKU-Associated PAH Mutations: A Fast and Simple Genotyping Test. <i>Methods and Protocols</i> , 2018, 1, 30.	2.0	3
153	Unusual Segregation of APP Mutations in Monogenic Alzheimer Disease. <i>Neurodegenerative Diseases</i> , 2019, 19, 96-100.	1.4	3
154	Obstetrical and perinatal outcomes in fetuses with early versus late sonographic diagnosis of short femur length: A single-center, prospective, cohort study. <i>European Journal of Obstetrics, Gynecology and Reproductive Biology</i> , 2020, 254, 170-174.	1.1	3
155	External hydrocephalus as a prenatal feature of noonan syndrome. <i>Annals of Human Genetics</i> , 2021, 85, 249-252.	0.8	3
156	Human fetal brain β -nerve growth factor cDNA: molecular cloning of 5' and 3' untranslated regions. <i>Neuroscience Letters</i> , 1991, 127, 117-120.	2.1	2
157	In vitro effect of PPAR- β Pro12Ala polymorphism on the deposition of Alzheimer's amyloid- β peptides. <i>Brain Research</i> , 2007, 1173, 1-5.	2.2	2
158	Lack of association between serotonin transporter 5-HTT gene polymorphism and endometriosis in an Italian patient population. <i>Journal of Negative Results in BioMedicine</i> , 2014, 13, 12.	1.4	2
159	Identification of a novel <i>RUNX2</i> gene mutation and early diagnosis of CCD in a cleidocranial dysplasia suspected Iranian family. <i>Clinical Case Reports (discontinued)</i> , 2020, 8, 2333-2340.	0.5	2
160	X-linked dominant <i>RPGR</i> gene mutation in a familial Coats angiomatosis. <i>BMC Ophthalmology</i> , 2021, 21, 37.	1.4	2
161	An observational study to assess Italian obstetrics providers' knowledge about preventive practices and diagnosis of congenital cytomegalovirus. <i>Journal of Perinatal Medicine</i> , 2021, 49, 67-72.	1.4	2
162	Chromosomal Microarray Analysis in Fetuses Detected with Isolated Cardiovascular Malformation: A Multicenter Study, Systematic Review of the Literature and Meta-Analysis. <i>Diagnostics</i> , 2022, 12, 1328.	2.6	2

#	ARTICLE	IF	CITATIONS
163	An enormous Italian pedigree of Marfan syndrome with a novel mutation in the FBN1 gene. Clinical Case Reports (discontinued), 2020, 8, 1445-1451.	0.5	1
164	Role of ductus venosus agenesis in right ventricle development. Journal of Maternal-Fetal and Neonatal Medicine, 2020, , 1-4.	1.5	1
165	Neonatal Marfan Syndrome by Inherited Mutation. Indian Journal of Pediatrics, 2021, 88, 176-177.	0.8	1
166	Incidental SOS1 variant identified by non-invasive prenatal screening: Prenatal diagnosis and family clinical reassessment. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2021, 256, 518-520.	1.1	1
167	Fetal early motor neuron disruption and prenatal molecular diagnosis in a severe BICD2opathy. American Journal of Medical Genetics, Part A, 2021, 185, 1509-1514.	1.2	1
168	MiRLog and dbmiR: Prioritization and functional annotation tools to study human microRNA sequence variants. Human Mutation, 2022, , .	2.5	1
169	High conservation of the trinucleotide [CTG] _n repeat at the myotonic dystrophy locus in nonhuman primates. Human Evolution, 1994, 9, 315-321.	2.0	0
170	Genetic variants of modulators of insulin action. International Congress Series, 2003, 1253, 45-53.	0.2	0
171	Evidence of involvement of a novel VUS variant in the CHKB gene to congenital muscular dystrophy affection. Meta Gene, 2020, 24, 100698.	0.6	0
172	Fetal dacryocystocele: A pitfall in the third trimester prenatal diagnosis of cleft lip. Journal of Clinical Ultrasound, 2021, 49, 777-778.	0.8	0
173	An update on the metabolic syndrome's epigenomic risk. Minerva Endocrinology, 2017, 42, 376-384.	1.1	0
174	From Nuremberg to bioethics: an educational project for students of dentistry and dental prosthesis. Annali Di Stomatologia, 2013, 4, 138-41.	0.6	0
175	Myoclonic Epilepsy: Case Report of a Mild Phenotype in a Pediatric Patient Expanding Clinical Spectrum of KCNA2 Pathogenic Variants. Frontiers in Neurology, 2021, 12, 806516.	2.4	0
176	Critical prenatal diagnosis and management of incidental exon 43-44 deletion in the dystrophin gene. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2022, , .	1.1	0
177	Genomic Breakpoints™ Characterization of a Large CHEK2 Duplication™ in an Italian Family with Hereditary Breast Cancer. Diagnostics, 2022, 12, 1520.	2.6	0