Antonio Pizzuti

List of Publications by Year
in descending order

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177	12,473	71102	25787
papers	citations	41 h-index	108 g-index

180	180	180	13578
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Risk of neural tube defects according to maternal body mass index: a systematic review and meta-analysis. Journal of Maternal-Fetal and Neonatal Medicine, 2022, 35, 7296-7305.	1.5	10
2	Molecular Approaches in Fetal Malformations, Dynamic Anomalies and Soft Markers: Diagnostic Rates and Challengesâ€"Systematic Review of the Literature and Meta-Analysis. Diagnostics, 2022, 12, 575.	2.6	11
3	Critical prenatal diagnosis and management of incidental exon 43â€“44 deletion in the dystrophin gene. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2022, , .	1.1	0
4	MiRLog and dbmiR: Prioritization and functional annotation tools to study human microRNA sequence variants. Human Mutation, 2022, , .	2.5	1
5	Potassium Channel KCNH1 Activating Variants Cause Altered Functional and Morphological Ciliogenesis. Molecular Neurobiology, 2022, 59, 4825-4838.	4.0	4
6	Chromosomal Microarray Analysis in Fetuses Detected with Isolated Cardiovascular Malformation: A Multicenter Study, Systematic Review of the Literature and Meta-Analysis. Diagnostics, 2022, 12, 1328.	2.6	2
7	Genomic Breakpointsâ€™ Characterization of a Large CHEK2 Duplication in an Italian Family with Hereditary Breast Cancer. Diagnostics, 2022, 12, 1520.	2.6	0
8	Susceptibility to ischaemic heart disease: Focusing on genetic variants for ATP-sensitive potassium channel beyond traditional risk factors. European Journal of Preventive Cardiology, 2021, 28, 1495-1500.	1.8	22
9	Neonatal Marfan Syndrome by Inherited Mutation. Indian Journal of Pediatrics, 2021, 88, 176-177.	0.8	1
10	Recurrent prenatal PIEZO1-related lymphatic dysplasia: Expanding molecular and ultrasound findings. European Journal of Medical Genetics, 2021, 64, 104106.	1.3	7
11	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
12	GDF5 mutation case report and a systematic review of molecular and clinical spectrum: Expanding current knowledge on genotype-phenotype correlations. Bone, 2021, 144, 115803.	2.9	7
13	Prenatal Exome Sequencing: Background, Current Practice and Future Perspectivesâ€"A Systematic Review. Diagnostics, 2021, 11, 224.	2.6	16
14	When to test fetuses for RASopathies? Proposition from a systematic analysis of 352 multicenter cases and a postnatal cohort. Genetics in Medicine, 2021, 23, 1116-1124.	2.4	17
15	Fetal early motor neuron disruption and prenatal molecular diagnosis in a severe BICD2 â€opathy. American Journal of Medical Genetics, Part A, 2021, 185, 1509-1514.	1.2	1
16	OTX015 Epi-Drug Exerts Antitumor Effects in Ovarian Cancer Cells by Blocking GNL3-Mediated Radioresistance Mechanisms: Cellular, Molecular and Computational Evidence. Cancers, 2021, 13, 1519.	3.7	7
17	miRâ€'125b/NRF2/HOâ€'1 axis is involved in protection against oxidative stress of cystic fibrosis: A pilot study. Experimental and Therapeutic Medicine, 2021, 21, 585.	1.8	6
18	Fetal dacryocystocele: A pitfall in the thirdâ€trimester prenatal diagnosis of cleft lip. Journal of Clinical Ultrasound, 2021, 49, 777-778.	0.8	0

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19	Altered Expression of Candidate Genes in Mayerâ€“Rokitanskyâ€“KÃ¼sterâ€“Hauser Syndrome May Influence Vaginal Keratinocytes Biology: A Focus on Protein Kinase X. Biology, 2021, 10, 450.	2.8	4
20	External hydrocephalus as a prenatal feature of noonan syndrome. Annals of Human Genetics, 2021, 85, 249-252.	0.8	3
21	Genotype-Phenotype Correlations in Monogenic Parkinson Disease: A Review on Clinical and Molecular Findings. Frontiers in Neurology, 2021, 12, 648588.	2.4	23
22	X-linked dominant RPGR gene mutation in a familial Coats angiomatosis. BMC Ophthalmology, 2021, 21, 37.	1.4	2
23	Proteinâ€“protein interaction network analysis applied to DNA copy number profiling suggests new perspectives on the aetiology of Mayerâ€“Rokitanskyâ€“KÃ¼sterâ€“Hauser syndrome. Scientific Reports, 2021, 11, 448.	3.3	13
24	An observational study to assess Italian obstetrics providersâ€™ knowledge about preventive practices and diagnosis of congenital cytomegalovirus. Journal of Perinatal Medicine, 2021, 49, 67-72.	1.4	2
25	Pregnant womenâ€™s knowledge and behaviour to prevent cytomegalovirus infection: an observational study. Journal of Perinatal Medicine, 2021, 49, 327-332.	1.4	7
26	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
27	Fetal tongue posture associated with micrognathia: An ultrasound marker of cleft secondary palate?. Journal of Clinical Ultrasound, 2020, 48, 48-51.	0.8	5
28	Small 7p22.3 microdeletion: Case report of Snx8 haploinsufficiency and neurological findings. European Journal of Medical Genetics, 2020, 63, 103772.	1.3	8
29	Prenatal whole exome sequencing detects a new homozygous fukutin (FKTN) mutation in a fetus with an ultrasound suspicion of familial Dandyâ€“Walker malformation. Molecular Genetics & Genomic Medicine, 2020, 8, e1054.	1.2	6
30	Obstetrical and perinatal outcomes in fetuses with early versus late sonographic diagnosis of short femur length: A single-center, prospective, cohort study. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2020, 254, 170-174.	1.1	3
31	Identification of a novel RUNX2 gene mutation and early diagnosis of CCD in a cleidocranial dysplasia suspected Iranian family. Clinical Case Reports (discontinued), 2020, 8, 2333-2340.	0.5	2
32	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
33	Heterozygous nonsense ARX mutation in a family highlights the complexity of clinical and molecular diagnosis in case of chromosomal and single gene disorder coâ€inheritance. Molecular Genetics & Genomic Medicine, 2020, 8, e1336.	1.2	4
34	Clinical and Molecular Spectrum of Myotonia and Periodic Paralyses Associated With Mutations in SCN4A in a Large Cohort of Italian Patients. Frontiers in Neurology, 2020, 11, 646.	2.4	7
35	Beyond BRCA1 and BRCA2: Deleterious Variants in DNA Repair Pathway Genes in Italian Families with Breast/Ovarian and Pancreatic Cancers. Journal of Clinical Medicine, 2020, 9, 3003.	2.4	5
36	Role of ductus venosus agenesis in right ventricle development. Journal of Maternal-Fetal and Neonatal Medicine, 2020, , 1-4.	1.5	1

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37	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
38	Myoclonic epilepsy, parkinsonism, schizophrenia and left-handedness as common neuropsychiatric features in 22q11.2 deletion syndrome. Journal of Medical Genetics, 2020, 57, 151-159.	3.2	9
39	TLR4 T399I Polymorphism and Endometriosis in a Cohort of Italian Women. Diagnostics, 2020, 10, 255.	2.6	6
40	BET inhibition therapy counteracts cancer cell survival, clonogenic potential and radioresistance mechanisms in rhabdomyosarcoma cells. Cancer Letters, 2020, 479, 71-88.	7.2	15
41	Defining the clinical-genetic and neuroradiological features in SPG54: description of eight additional cases and nine novel DDHD2 variants. Journal of Neurology, 2019, 266, 2657-2664.	3.6	19
42	Unusual Segregation of APP Mutations in Monogenic Alzheimer Disease. Neurodegenerative Diseases, 2019, 19, 96-100.	1.4	3
43	Clinical and functional characterization of a novel RASopathyâ€causingSHOC2mutation associated with prenatalâ€onset hypertrophic cardiomyopathy. Human Mutation, 2019, 40, 1046-1056.	2.5	18
44	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
45	Midtrimester isolated short femur and perinatal outcomes: A systematic review and metaâ€analysis. Acta Obstetricia Et Gynecologica Scandinavica, 2019, 98, 11-17.	2.8	22
46	Update in non-invasive prenatal testing. Minerva Ginecologica, 2019, 71, 44-53.	0.8	5
47	Whole exome sequencing in an Italian family with isolated maxillary canine agenesis and canine eruption anomalies. Archives of Oral Biology, 2018, 91, 96-102.	1.8	8
48	Molecular Analysis of PKU-Associated PAH Mutations: A Fast and Simple Genotyping Test. Methods and Protocols, 2018, 1, 30.	2.0	3
49	Rapid detection of copy number variations and point mutations in BRCA1/2 genes using a single workflow by ion semiconductor sequencing pipeline. Oncotarget, 2018, 9, 33648-33655.	1.8	11
50	Unusual association of SCN2A epileptic encephalopathy with severe cortical dysplasia detected by prenatal MRI. European Journal of Paediatric Neurology, 2017, 21, 587-590.	1.6	16
51	Lack of pathogenic mutations in SOS1 gene in phenytoin-induced gingival overgrowth patients. Archives of Oral Biology, 2017, 80, 160-163.	1.8	3
52	A sketch of known and novel MYCN-associated miRNA networks in neuroblastoma. Oncology Reports, 2017, 38, 3-20.	2.6	24
53	Role of fetal MRI in the evaluation of isolated and nonâ€isolated corpus callosum dysgenesis: results of a crossâ€sectional study. Prenatal Diagnosis, 2017, 37, 244-252.	2.3	18
54	Pfeiffer syndrome: literature review of prenatal sonographic findings and genetic diagnosis. Journal of Maternal-Fetal and Neonatal Medicine, 2017, 30, 2225-2231.	1.5	16

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55	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
56	An update on the metabolic syndrome's epigenomic risk. Minerva Endocrinology, 2017, 42, 376-384.	1.1	0
57	Prenatal diagnosis of proximal focal femoral deficiency: Literature review of prenatal sonographic findings. Journal of Clinical Ultrasound, 2016, 44, 252-259.	0.8	13
58	Recessive Inactivating Mutations in TBCK, Encoding a Rab GTPase-Activating Protein, Cause Severe Infantile Syndromic Encephalopathy. American Journal of Human Genetics, 2016, 98, 772-781.	6.2	43
59	Comparative Analysis of Real-Time Polymerase Chain Reaction Methods to Typing HLA-B*57:01 in HIV-1-Positive Patients. AIDS Research and Human Retroviruses, 2016, 32, 654-657.	1.1	7
60	Molecular analysis of sarcomeric and non-sarcomeric genes in patients with hypertrophic cardiomyopathy. Gene, 2016, 577, 227-235.	2.2	26
61	DNMT3Bin vitroknocking-down is able to reverse embryonal rhabdomyosarcoma cell phenotype through inhibition of proliferation and induction of myogenic differentiation. Oncotarget, 2016, 7, 79342-79356.	1.8	37
62	Crizotinib-induced antitumour activity in human alveolar rhabdomyosarcoma cells is not solely dependent on ALK and MET inhibition. Journal of Experimental and Clinical Cancer Research, 2015, 34, 112.	8.6	41
63	The Use of Piezosurgery in Cranial Surgery in Children. Journal of Craniofacial Surgery, 2015, 26, 840-842.	0.7	16
64	Loss-of-Function Mutations in APPL1 in Familial Diabetes Mellitus. American Journal of Human Genetics, 2015, 97, 177-185.	6.2	114
65	Mutations in KCNH1 and ATP6V1B2 cause Zimmermann-Laband syndrome. Nature Genetics, 2015, 47, 661-667.	21.4	177
66	Deep Sequencing the microRNA profile in rhabdomyosarcoma reveals down-regulation of miR-378 family members. BMC Cancer, 2014, 14, 880.	2.6	56
67	Immunogenetic investigation in vernal keratoconjunctivitis. Pediatric Allergy and Immunology, 2014, 25, 508-510.	2.6	16
68	Single nucleotide polymorphisms in the promoter regions of Foxp3 and ICOSLG genes are associated with Alopecia Areata. Clinical and Experimental Medicine, 2014, 14, 91-97.	3.6	33
69	Lack of association between serotonin transporter 5-HTT gene polymorphism and endometriosis in an Italian patient population. Journal of Negative Results in BioMedicine, 2014, 13, 12.	1.4	2
70	Novel SMAD4 mutation causing Myhre syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 1835-1840.	1.2	29
71	Cytotoxic T-lymphocyte antigen 4 (CTLA4) +49AG and CT60 gene polymorphisms in Alopecia Areata: a caseâ€“control association study in the Italian population. Archives of Dermatological Research, 2013, 305, 665-670.	1.9	22
72	Clinical and genetic study of two patients with Zimmermannâ€“Laband syndrome and literature review. European Journal of Medical Genetics, 2013, 56, 570-576.	1.3	32

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73	Neurocognitive effects of methylphenidate on ADHD children with different DAT genotypes: AÂlongitudinal open label trial. European Journal of Paediatric Neurology, 2013, 17, 407-414.	1.6	26
74	Elevated levels of miR-145 correlate with SMAD3 down-regulation in Cystic Fibrosis patients. Journal of Cystic Fibrosis, 2013, 12, 797-802.	0.7	57
75	Loss of function of the E3 ubiquitin-protein ligase UBE3B causes Kaufman oculocerebrofacial syndrome. Journal of Medical Genetics, 2013, 50, 493-499.	3.2	40
76	From Nuremberg to bioethics: an educational project for students of dentistry and dental prosthesis. Annali Di Stomatologia, 2013, 4, 138-41.	0.6	0
77	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. Journal of Biological Chemistry, 2012, 287, 15635-15647.	3.4	233
78	HLA-DQA1 and HLA-DQB1 in Celiac disease predisposition: practical implications of the HLA molecular typing. Journal of Biomedical Science, 2012, 19, 88.	7.0	170
79	Synergistic Post-Transcriptional Regulation of the Cystic Fibrosis Transmembrane conductance Regulator (CFTR) by miR-101 and miR-494 Specific Binding. PLoS ONE, 2011, 6, e26601.	2.5	80
80	Brain Derived Neurotrophic Factor (BDNF) Expression Is Regulated by MicroRNAs miR-26a and miR-26b Allele-Specific Binding. PLoS ONE, 2011, 6, e28656.	2.5	110
81	Genetic association of HLAâ€ÐQB1 and HLAâ€ÐRB1 polymorphisms with alopecia areata in the Italian population. British Journal of Dermatology, 2011, 165, 823-827.	1.5	30
82	Familial spinal neurofibromatosis due to a multiexonic NF1 gene deletion. Neurogenetics, 2011, 12, 233-240.	1.4	8
83	Early ultrasound suspect of thanatophoric dysplasia followed by first trimester molecular diagnosis. American Journal of Medical Genetics, Part A, 2011, 155, 1756-1758.	1.2	4
84	Mitochondrial disfunction as a cause of ALS. Archives Italiennes De Biologie, 2011, 149, 113-9.	0.4	14
85	Two Novel Mutations Affecting Splicing in the IRF6 Gene Associated With van der Woude Syndrome. Journal of Craniofacial Surgery, 2010, 21, 1654-1656.	0.7	9
86	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
87	Triple A syndrome: A novel compound heterozygous mutation in the AAAS gene in an Italian patient without adrenal insufficiency. Journal of the Neurological Sciences, 2010, 290, 150-152.	0.6	15
88	Genetic variants in adipose triglyceride lipase influence lipid levels in familial combined hyperlipidemia. Atherosclerosis, 2010, 213, 206-211.	0.8	8
89	Quantification of Small Non-Coding RNAs Allows an Accurate Comparison of miRNA Expression Profiles. Journal of Biomedicine and Biotechnology, 2009, 2009, 1-9.	3.0	21
90	Clinical, neuropsychological, neurophysiologic, and genetic features of a new Italian pedigree with familial cortical myoclonic tremor with epilepsy. Epilepsia, 2009, 50, 1284-1288.	5.1	40

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91	High prevalence of epilepsy in a village in the Littoral Province of Cameroon. Epilepsy Research, 2008, 82, 200-210.	1.6	71
92	Unravelling the Complexity of T Cell Abnormalities in Common Variable Immunodeficiency. Journal of Immunology, 2007, 178, 3932-3943.	0.8	249
93	Founder and Recurrent CDH1 Mutations in Families With Hereditary Diffuse Gastric Cancer. JAMA - Journal of the American Medical Association, 2007, 297, 2360.	7.4	394
94	Clinical lumping and molecular splitting of LEOPARD and NF1/NF1-Noonan syndromes. American Journal of Medical Genetics, Part A, 2007, 143A, 1009-1011.	1.2	13
95	Functional analysis of splicing mutations in exon 7 of NF1gene. BMC Medical Genetics, 2007, 8, 4.	2.1	32
96	In vitro effect of PPAR-Î³2 Pro12Ala polymorphism on the deposition of Alzheimer's amyloid-Î² peptides. Brain Research, 2007, 1173, 1-5.	2.2	2
97	Case report of adult-onset Allgrove syndrome. Neurological Sciences, 2007, 28, 331-335.	1.9	15
98	Germline Missense Mutations Affecting KRAS Isoform B Are Associated with a Severe Noonan Syndrome Phenotype. American Journal of Human Genetics, 2006, 79, 129-135.	6.2	205
99	Additional evidence thatPTPN11 mutations play only a minor role in the pathogenesis of non-syndromic atrioventricular canal defect. American Journal of Medical Genetics, Part A, 2006, 140A, 1970-1972.	1.2	4
100	Additive Effects of Genetic Variation in Dopamine Regulating Genes on Working Memory Cortical Activity in Human Brain. Journal of Neuroscience, 2006, 26, 3918-3922.	3.6	208
101	Clinical features and outcome of familial chronic lymphocytic leukemia. Haematologica, 2006, 91, 1117-20.	3.5	35
102	Role of peroxisome proliferator-activated receptor Î³ in amyloid precursor protein processing and amyloid Î²-mediated cell death. Biochemical Journal, 2005, 391, 693-698.	3.7	78
103	ZFPM2/FOG2 andHEY2 genes analysis in nonsyndromic tricuspid atresia. American Journal of Medical Genetics, Part A, 2005, 133A, 68-70.	1.2	19
104	Hyperthrophic cardiomyopathy and thePTPN11 gene. American Journal of Medical Genetics, Part A, 2005, 136A, 93-94.	1.2	8
105	CRELD1 andGATA4 gene analysis in patients with nonsyndromic atrioventricular canal defects. American Journal of Medical Genetics, Part A, 2005, 139A, 236-238.	1.2	26
106	LGI1 gene mutation screening in sporadic partial epilepsy with auditory features. Journal of Neurology, 2005, 252, 62-66.	3.6	13
107	NF1 Gene Mutations Represent the Major Molecular Event Underlying Neurofibromatosis-Noonan Syndrome. American Journal of Human Genetics, 2005, 77, 1092-1101.	6.2	139
108	Association of the Matrix Metalloproteinase-3 (MMP-3) Promoter Polymorphism With Celiac Disease in Male Subjects. Human Immunology, 2005, 66, 715-719.	2.4	14

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109	A novel PTPN11 gene mutation bridges Noonan syndrome, multiple lentigines/LEOPARD syndrome and Noonan-like/multiple giant cell lesion syndrome. European Journal of Human Genetics, 2004, 12, 1069-1072.	2.8	51
110	Familial aggregation of genetically heterogeneous hypertrophic cardiomyopathy: A boy with LEOPARD syndrome due to PTPN11 mutation and his nonsyndromic father lacking PTPN11 mutations. Birth Defects Research Part A: Clinical and Molecular Teratology, 2004, 70, 95-98.	1.6	14
111	A homozygousGJA1 gene mutation causes a Hallermann-Streiff/ODDD spectrum phenotype. Human Mutation, 2004, 23, 286-286.	2.5	97
112	Mutations of the Nogo-66 receptor (RTN4R) gene in schizophrenia. Human Mutation, 2004, 24, 534-535.	2.5	77
113	LEOPARD syndrome: a new polyaneurysm association and an update on the molecular genetics of the disease. Journal of Vascular Surgery, 2004, 39, 897-900.	1.1	16
114	Mutational analysis of parkin gene by denaturing high-performance liquid chromatography (DHPLC) in essential tremor. Parkinsonism and Related Disorders, 2004, 10, 357-362.	2.2	20
115	Mutations ofZFPM2/FOG2 gene in sporadic cases of tetralogy of Fallot. Human Mutation, 2003, 22, 372-377.	2.5	127
116	Familial blepharospasm is inherited as an autosomal dominant trait and relates to a novel unassigned gene. Movement Disorders, 2003, 18, 207-212.	3.9	43
117	Nonsyndromic Pulmonary Valve Stenosis and thePTPN11 Gene. American Journal of Medical Genetics Part A, 2003, 116A, 389-390.	2.4	15
118	Epilepsy with auditory features: ALGI1 gene mutation suggests a loss-of-function mechanism. Annals of Neurology, 2003, 53, 396-399.	5.3	57
119	DiGeorge subtypes of nonsyndromic conotruncal defects: evidence against a major role of TBX1 Gene. European Journal of Human Genetics, 2003, 11, 349-351.	2.8	48
120	Genetic variants of modulators of insulin action. International Congress Series, 2003, 1253, 45-53.	0.2	0
121	An ATG Repeat in the 3â€²-Untranslated Region of the Human Resistin Gene Is Associated with a Decreased Risk of Insulin Resistance. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 4403-4406.	3.6	82
122	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. Nephrology Dialysis Transplantation, 2002, 17, 1402-1407.	0.7	16
123	A Variation in 3â€² UTR of hPTP1B Increases Specific Gene Expression and Associates with Insulin Resistance. American Journal of Human Genetics, 2002, 70, 806-812.	6.2	179
124	Grouping of Multiple-Lentigines/LEOPARD and Noonan Syndromes on the PTPN11 Gene. American Journal of Human Genetics, 2002, 71, 389-394.	6.2	380
125	A family with autosomal dominant mutilating neuropathy not linked to either Charcotâ€“Marieâ€“Tooth disease type 2B (CMT2B) or hereditary sensory neuropathy type I (HSN I) loci. Neuromuscular Disorders, 2002, 12, 286-291.	0.6	9
126	Novel Italian family supports clinical and genetic heterogeneity of primary adult-onset torsion dystonia. Movement Disorders, 2002, 17, 392-397.	3.9	23

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127	Cytogenetic mapping of a novel locus for typeÂII Waardenburg syndrome. Human Genetics, 2002, 110, 64-67.	3.8	23
128	Assignment of a locus for autosomal dominant idiopathic scoliosis (IS) to human chromosome 17p11. Human Genetics, 2002, 111, 401-404.	3.8	125
129	Leiomyosarcoma of the Larynx: Case Report with Pathologic and Surgical Considerations. The Journal of Otolaryngology, 2002, 31, 393.	0.6	5
130	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
131	Genomic Organization, Physical Mapping, and Involvement in Yq Microdeletions of the VCY2 (BPY 2) Gene. Genomics, 2001, 72, 153-157.	2.9	15
132	Human developing motor neurons as a tool to study ALS. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders: Official Publication of the World Federation of Neurology, Research Group on Motor Neuron Diseases, 2001, 2, 69-76.	1.2	4
133	The Q121 PC-1 Variant and Obesity Have Additive and Independent Effects in Causing Insulin Resistance. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 5888-5891.	3.6	53
134	The Q121 PC-1 Variant and Obesity Have Additive and Independent Effects in Causing Insulin Resistance. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 5888-5891.	3.6	19
135	Narrowing the Duane syndrome critical region at chromosomeÂ8q13 down to 40Âkb. European Journal of Human Genetics, 2000, 8, 319-324.	2.8	32
136	A polymorphism (K121Q) of the human glycoprotein PC-1 gene coding region is strongly associated with insulin resistance.. Diabetes, 1999, 48, 1881-1884.	0.6	228
137	Genomic structure, promoter characterisation and mutational analysis of the S100A7 gene: exclusion of a candidate for familial psoriasis susceptibility. Human Genetics, 1999, 104, 130-134.	3.8	37
138	Mutations of UFD1L Are Not Responsible for the Majority of Cases of DiGeorge Syndrome/Velocardiofacial Syndrome without Deletions within Chromosome 22q11. American Journal of Human Genetics, 1999, 65, 247-249.	6.2	36
139	Mapping of the MYCL2 processed gene to Xq22-23 and identification of an additional L MYC-related sequence in Xq27.2. FEBS Letters, 1999, 446, 273-277.	2.8	8
140	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
141	Motor neurone metabolism. Journal of the Neurological Sciences, 1999, 169, 161-169.	0.6	14
142	Isolation and Characterization of a Novel Transcript Embedded within HIRA, a Gene Deleted in DiGeorge Syndrome. Molecular Genetics and Metabolism, 1999, 67, 227-235.	1.1	22
143	Structure and expression of the human ubiquitin fusionâ€“degradation gene (UFD1L). Biochimica Et Biophysica Acta Gene Regulatory Mechanisms, 1998, 1396, 158-162.	2.4	22
144	Immunomagnetic isolation of human developing motor neurons. NeuroReport, 1998, 9, 1143-1147.	1.2	12

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145	UFD1L, a Developmentally Expressed Ubiquitination Gene, is Deleted in CATCH 22 Syndrome. Human Molecular Genetics, 1997, 6, 259-265.	2.9	85
146	SMT3A,a Human Homologue of theS. cerevisiae SMT3Gene, Maps to Chromosome 21qter and Defines a Novel Gene Family. Genomics, 1997, 40, 362-366.	2.9	112
147	mRNA distribution in adult human brain of GRIN2B, a N-methyl-d-aspartate (NMDA) receptor subunit. Neuroscience Letters, 1997, 239, 49-53.	2.1	43
148	Hepatitis G virus infection in hemodialysis patients. Kidney International, 1997, 51, 348-352.	5.2	44
149	Expression Study of Survival Motor Neuron Gene in Human Fetal Tissues. Biochemical and Molecular Medicine, 1997, 61, 102-106.	1.4	31
150	Deletion analysis of SMN and NAIP genes in spinal muscular atrophy Italian families. , 1996, 19, 378-380.		6
151	Deletion analysis of the simple tandem repeat loci physically linked to the spinal muscular atrophy locus. Human Mutation, 1996, 7, 198-201.	2.5	7
152	cDNA characterization and chromosomal mapping of two human homologues of the Drosophila dishevelled polarity gene. Human Molecular Genetics, 1996, 5, 953-958.	2.9	57
153	Postzygotic instability of the myotonic dystrophy p[AGC]n repeat supported by larger expansions in muscle and reduced amplifications in sperm. Journal of Neurology, 1995, 242, 379-383.	3.6	13
154	Identification of Multiple Transcribed Sequences from the Spinal Muscular Atrophy Region on Human Chromosome 5. Biochemical and Biophysical Research Communications, 1995, 206, 294-301.	2.1	7
155	Survival Motor-Neuron Gene Transcript Analysis in Muscles from Spinal Muscular-Atrophy Patients. Biochemical and Biophysical Research Communications, 1995, 213, 342-348.	2.1	182
156	Different Expression of the Myotonin Protein Kinase Gene in Discrete Areas of Human Brain. Biochemical and Biophysical Research Communications, 1995, 216, 489-494.	2.1	9
157	Isolation of a New Gene in the Friedreich Ataxia Candidate Region on Human Chromosome 9 by cDNA Direct Selection. Biochemical Medicine and Metabolic Biology, 1994, 52, 115-119.	0.7	7
158	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
159	(CTG)n Triplet Mutation and Phenotype Manifestations in Myotonic Dystrophy Patients. Biochemical Medicine and Metabolic Biology, 1993, 50, 85-92.	0.7	47
160	Human Elongation Factor EF-1Î²: Cloning and Characterization of the EF1Î²5a Gene and Assignment of EF-1Î² Isoforms to Chromosomes 2, 5, 15, and X. Biochemical and Biophysical Research Communications, 1993, 197, 154-162.	2.1	23
161	The Myotonic Dystrophy Gene. Archives of Neurology, 1993, 50, 1173-1179.	4.5	37
162	A transposon-like element in the deletion-prone region of the dystrophin gene. Genomics, 1992, 13, 594-600.	2.9	39

#	Article	IF	Citations
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