Antonio Pizzuti

List of Publications by Year in Descending Order

172 papers	10,570 citations	39 h-index	101 g-index
179 ext. papers	11,527 ext. citations	5.7 avg, IF	5.02 L-index

#	Paper	IF	Citations
172	Molecular Approaches in Fetal Malformations, Dynamic Anomalies and Soft Markers: Diagnostic Rates and Challenges-Systematic Review of the Literature and Meta-Analysis.. *Diagnostics*, **2022**, 12,	3.8	1
171	Critical prenatal diagnosis and management of incidental exon 43-44 deletion in the dystrophin gene.. *European Journal of Obstetrics, Gynecology and Reproductive Biology*, **2022**,	2.4	
170	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	3.8	0
169	Myoclonic Epilepsy: Case Report of a Mild Phenotype in a Pediatric Patient Expanding Clinical Spectrum of Pathogenic Variants.. *Frontiers in Neurology*, **2021**, 12, 806516	4.1	
168	Pregnant women knowledge and behaviour to prevent cytomegalovirus infection: an observational study. *Journal of Perinatal Medicine*, **2021**, 49, 327-332	2.7	3
167	OTX015 Epi-Drug Exerts Antitumor Effects in Ovarian Cancer Cells by Blocking GNL3-Mediated Radioresistance Mechanisms: Cellular, Molecular and Computational Evidence. *Cancers*, **2021**, 13,	6.6	2
166	miR-125b/NRF2/HO-1 axis is involved in protection against oxidative stress of cystic fibrosis: A pilot study. *Experimental and Therapeutic Medicine*, **2021**, 21, 585	2.1	3
165	Fetal dacryocystocele: A pitfall in the third-trimester prenatal diagnosis of cleft lip. *Journal of Clinical Ultrasound*, **2021**, 49, 777-778	1	
164	Altered Expression of Candidate Genes in Mayer-Rokitansky-Küster-Hauser Syndrome May Influence Vaginal Keratinocytes Biology: A Focus on Protein Kinase X. *Biology*, **2021**, 10,	4.9	1
163	External hydrocephalus as a prenatal feature of noonan syndrome. *Annals of Human Genetics*, **2021**, 85, 249-252	2.2	1
162	Neonatal Marfan Syndrome by Inherited Mutation. *Indian Journal of Pediatrics*, **2021**, 88, 176-177	3	1
161	Recurrent prenatal PIEZO1-related lymphatic dysplasia: Expanding molecular and ultrasound findings. *European Journal of Medical Genetics*, **2021**, 64, 104106	2.6	3
160	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	2.4	1
159	GDF5 mutation case report and a systematic review of molecular and clinical spectrum: Expanding current knowledge on genotype-phenotype correlations. *Bone*, **2021**, 144, 115803	4.7	2
158	Prenatal Exome Sequencing: Background, Current Practice and Future Perspectives-A Systematic Review. *Diagnostics*, **2021**, 11,	3.8	5
157	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	8.1	5
156	Fetal early motor neuron disruption and prenatal molecular diagnosis in a severe BICD2-opathy. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1509-1514	2.5	1

155	Risk of neural tube defects according to maternal body mass index: a systematic review and meta-analysis. *Journal of Maternal-Fetal and Neonatal Medicine*, **2021**, 1-10	2	3
154	Genotype-Phenotype Correlations in Monogenic Parkinson Disease: A Review on Clinical and Molecular Findings. *Frontiers in Neurology*, **2021**, 12, 648588	4.1	3
153	X-linked dominant RPGR gene mutation in a familial Coats angiomatosis. *BMC Ophthalmology*, **2021**, 21, 37	2.3	1
152	Protein-protein interaction network analysis applied to DNA copy number profiling suggests new perspectives on the aetiology of Mayer-Rokitansky-Küster-Hauser syndrome. *Scientific Reports*, **2021**, 11, 448	4.9	5
151	Evidence of involvement of a novel VUS variant in the CHKB gene to congenital muscular dystrophy affection. *Meta Gene*, **2020**, 24, 100698	0.7	
150	Susceptibility to ischaemic heart disease: Focusing on genetic variants for ATP-sensitive potassium channel beyond traditional risk factors. *European Journal of Preventive Cardiology*, **2020**, 2047487320926780	3.9	13
149	Myoclonic epilepsy, parkinsonism, schizophrenia and left-handedness as common neuropsychiatric features in 22q11.2 deletion syndrome. *Journal of Medical Genetics*, **2020**, 57, 151-159	5.8	5
148	T399I Polymorphism and Endometriosis in a Cohort of Italian Women. *Diagnostics*, **2020**, 10,	3.8	3
147	BET inhibition therapy counteracts cancer cell survival, clonogenic potential and radioresistance mechanisms in rhabdomyosarcoma cells. *Cancer Letters*, **2020**, 479, 71-88	9.9	8
146	An observational study to assess Italian obstetrics providers' knowledge about preventive practices and diagnosis of congenital cytomegalovirus. *Journal of Perinatal Medicine*, **2020**, 49, 67-72	2.7	1
145	Prenatal whole exome sequencing detects a new homozygous fukutin (FKTN) mutation in a fetus with an ultrasound suspicion of familial Dandy-Walker malformation. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1054	2.3	3
144	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	2.4	1
143	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	0.7	0
142	An enormous Italian pedigree of Marfan syndrome with a novel mutation in the FBN1 gene. *Clinical Case Reports (discontinued)*, **2020**, 8, 1445-1451	0.7	0
141	Heterozygous nonsense ARX mutation in a family highlights the complexity of clinical and molecular diagnosis in case of chromosomal and single gene disorder co-inheritance. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1336	2.3	1
140	Clinical and Molecular Spectrum of Myotonia and Periodic Paralyses Associated With Mutations in in a Large Cohort of Italian Patients. *Frontiers in Neurology*, **2020**, 11, 646	4.1	3
139	Role of ductus venosus agenesis in right ventricle development. *Journal of Maternal-Fetal and Neonatal Medicine*, **2020**, 1-4	2	1
138	Fetal tongue posture associated with micrognathia: An ultrasound marker of cleft secondary palate?. *Journal of Clinical Ultrasound*, **2020**, 48, 48-51	1	1

137	Small 7p22.3 microdeletion: Case report of Snx8 haploinsufficiency and neurological findings. *European Journal of Medical Genetics*, **2020**, 63, 103772	2.6	4
136	Unusual Segregation of APP Mutations in Monogenic Alzheimer Disease. *Neurodegenerative Diseases*, **2019**, 19, 96-100	2.3	1
135	Clinical and functional characterization of a novel RASopathy-causing SHOC2 mutation associated with prenatal-onset hypertrophic cardiomyopathy. *Human Mutation*, **2019**, 40, 1046-1056	4.7	6
134	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	5.5	9
133	Update in non-invasive prenatal testing. *Minerva Ginecologica*, **2019**, 71, 44-53	1.2	1
132	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	4.9	16
131	Midtrimester isolated short femur and perinatal outcomes: A systematic review and meta-analysis. *Acta Obstetricia Et Gynecologica Scandinavica*, **2019**, 98, 11-17	3.8	15
130	Whole exome sequencing in an Italian family with isolated maxillary canine agenesis and canine eruption anomalies. *Archives of Oral Biology*, **2018**, 91, 96-102	2.8	3
129	Molecular Analysis of PKU-Associated PAH Mutations: A Fast and Simple Genotyping Test. *Methods and Protocols*, **2018**, 1,	2.5	1
128	Rapid detection of copy number variations and point mutations in genes using a single workflow by ion semiconductor sequencing pipeline. *Oncotarget*, **2018**, 9, 33648-33655	3.3	9
127	Unusual association of SCN2A epileptic encephalopathy with severe cortical dysplasia detected by prenatal MRI. *European Journal of Paediatric Neurology*, **2017**, 21, 587-590	3.8	9
126	Lack of pathogenic mutations in SOS1 gene in phenytoin-induced gingival overgrowth patients. *Archives of Oral Biology*, **2017**, 80, 160-163	2.8	2
125	A sketch of known and novel MYCN-associated miRNA networks in neuroblastoma. *Oncology Reports*, **2017**, 38, 3-20	3.5	20
124	Role of fetal MRI in the evaluation of isolated and non-isolated corpus callosum dysgenesis: results of a cross-sectional study. *Prenatal Diagnosis*, **2017**, 37, 244-252	3.2	12
123	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	22.4	23
122	Pfeiffer syndrome: literature review of prenatal sonographic findings and genetic diagnosis. *Journal of Maternal-Fetal and Neonatal Medicine*, **2017**, 30, 2225-2231	2	10
121	An update on the metabolic syndrome epigenomic risk. *Minerva Endocrinology*, **2017**, 42, 376-384	2.5	
120	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, 654-7	1.6	6

119	Molecular analysis of sarcomeric and non-sarcomeric genes in patients with hypertrophic cardiomyopathy. *Gene*, **2016**, 577, 227-35	3.8	24
118	DNMT3B in vitro knocking-down is able to reverse embryonal rhabdomyosarcoma cell phenotype through inhibition of proliferation and induction of myogenic differentiation. *Oncotarget*, **2016**, 7, 79342-79356	5.3	27
117	Prenatal diagnosis of proximal focal femoral deficiency: Literature review of prenatal sonographic findings. *Journal of Clinical Ultrasound*, **2016**, 44, 252-9	1	10
116	Recessive Inactivating Mutations in TBCK, Encoding a Rab GTPase-Activating Protein, Cause Severe Infantile Syndromic Encephalopathy. *American Journal of Human Genetics*, **2016**, 98, 772-81	11	29
115	Mutations in KCNH1 and ATP6V1B2 cause Zimmermann-Laband syndrome. *Nature Genetics*, **2015**, 47, 661-7	36.3	128
114	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	12.8	38
113	The use of piezosurgery in cranial surgery in children. *Journal of Craniofacial Surgery*, **2015**, 26, 840-2	1.2	13
112	Loss-of-Function Mutations in APPL1 in Familial Diabetes Mellitus. *American Journal of Human Genetics*, **2015**, 97, 177-85	11	91
111	Single nucleotide polymorphisms in the promoter regions of Foxp3 and ICOSLG genes are associated with Alopecia areata. *Clinical and Experimental Medicine*, **2014**, 14, 91-7	4.9	25
110	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		2
109	Novel SMAD4 mutation causing Myhre syndrome. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 1835-40	2.5	23
108	Deep Sequencing the microRNA profile in rhabdomyosarcoma reveals down-regulation of miR-378 family members. *BMC Cancer*, **2014**, 14, 880	4.8	43
107	Immunogenetic investigation in vernal keratoconjunctivitis. *Pediatric Allergy and Immunology*, **2014**, 25, 508-10	4.2	12
106	Cytotoxic T-lymphocyte antigen 4 (CTLA4) +49AG and CT60 gene polymorphisms in Alopecia Areata: a case-control association study in the Italian population. *Archives of Dermatological Research*, **2013**, 305, 665-70	3.3	19
105	Clinical and genetic study of two patients with Zimmermann-Laband syndrome and literature review. *European Journal of Medical Genetics*, **2013**, 56, 570-6	2.6	23
104	Neurocognitive effects of methylphenidate on ADHD children with different DAT genotypes: a longitudinal open label trial. *European Journal of Paediatric Neurology*, **2013**, 17, 407-14	3.8	23
103	Elevated levels of miR-145 correlate with SMAD3 down-regulation in cystic fibrosis patients. *Journal of Cystic Fibrosis*, **2013**, 12, 797-802	4.1	47
102	Loss of function of the E3 ubiquitin-protein ligase UBE3B causes Kaufman oculocerebrofacial syndrome. *Journal of Medical Genetics*, **2013**, 50, 493-9	5.8	33

101	From Nuremberg to bioethics: an educational project for students of dentistry and dental prosthesis. *Annali Di Stomatologia*, **2013**, 4, 138-41		
100	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, 15635-47	5.4	190
99	HLA-DQA1 and HLA-DQB1 in Celiac disease predisposition: practical implications of the HLA molecular typing. *Journal of Biomedical Science*, **2012**, 19, 88	13.3	120
98	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	3.7	70
97	Brain derived neurotrophic factor (BDNF) expression is regulated by microRNAs miR-26a and miR-26b allele-specific binding. *PLoS ONE*, **2011**, 6, e28656	3.7	86
96	Genetic association of HLA-DQB1 and HLA-DRB1 polymorphisms with alopecia areata in the Italian population. *British Journal of Dermatology*, **2011**, 165, 823-7	4	26
95	Familial spinal neurofibromatosis due to a multiexonic NF1 gene deletion. *Neurogenetics*, **2011**, 12, 233-40	3	6
94	Early ultrasound suspect of thanatophoric dysplasia followed by first trimester molecular diagnosis. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1756-8	2.5	3
93	Mitochondrial disfunction as a cause of ALS. *Archives Italiennes De Biologie*, **2011**, 149, 113-9	1.1	13
92	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, 150-2	3.2	12
91	Genetic variants in adipose triglyceride lipase influence lipid levels in familial combined hyperlipidemia. *Atherosclerosis*, **2010**, 213, 206-11	3.1	7
90	Two novel mutations affecting splicing in the IRF6 gene associated with van der Woude syndrome. *Journal of Craniofacial Surgery*, **2010**, 21, 1654-6	1.2	8
89	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, 107-9	2.5	29
88	Quantification of small non-coding RNAs allows an accurate comparison of miRNA expression profiles. *Journal of Biomedicine and Biotechnology*, **2009**, 2009, 659028		18
87	Clinical, neuropsychological, neurophysiologic, and genetic features of a new Italian pedigree with familial cortical myoclonic tremor with epilepsy. *Epilepsia*, **2009**, 50, 1284-8	6.4	29
86	High prevalence of epilepsy in a village in the Littoral Province of Cameroon. *Epilepsy Research*, **2008**, 82, 200-10	3	58
85	Clinical lumping and molecular splitting of LEOPARD and NF1/NF1-Noonan syndromes. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 1009-11	2.5	12
84	Functional analysis of splicing mutations in exon 7 of NF1 gene. *BMC Medical Genetics*, **2007**, 8, 4	2.1	25

83	In vitro effect of PPAR-gamma2 Pro12Ala polymorphism on the deposition of Alzheimer's amyloid-beta peptides. *Brain Research*, **2007**, 1173, 1-5	3.7	2
82	Case report of adult-onset Allgrove syndrome. *Neurological Sciences*, **2007**, 28, 331-5	3.5	12
81	Unravelling the complexity of T cell abnormalities in common variable immunodeficiency. *Journal of Immunology*, **2007**, 178, 3932-43	5.3	202
80	Founder and recurrent CDH1 mutations in families with hereditary diffuse gastric cancer. *JAMA - Journal of the American Medical Association*, **2007**, 297, 2360-72	27.4	324
79	Additional evidence that PTPN11 mutations play only a minor role in the pathogenesis of non-syndromic atrioventricular canal defect. *American Journal of Medical Genetics, Part A*, **2006**, 140, 1970-2	2.5	3
78	Additive effects of genetic variation in dopamine regulating genes on working memory cortical activity in human brain. *Journal of Neuroscience*, **2006**, 26, 3918-22	6.6	198
77	Germline missense mutations affecting KRAS Isoform B are associated with a severe Noonan syndrome phenotype. *American Journal of Human Genetics*, **2006**, 79, 129-35	11	183
76	Clinical features and outcome of familial chronic lymphocytic leukemia. *Haematologica*, **2006**, 91, 1117-20	6.6	31
75	NF1 gene mutations represent the major molecular event underlying neurofibromatosis-Noonan syndrome. *American Journal of Human Genetics*, **2005**, 77, 1092-101	11	115
74	Association of the matrix metalloproteinase-3 (MMP-3) promoter polymorphism with celiac disease in male subjects. *Human Immunology*, **2005**, 66, 716-20	2.3	11
73	Role of peroxisome proliferator-activated receptor gamma in amyloid precursor protein processing and amyloid beta-mediated cell death. *Biochemical Journal*, **2005**, 391, 693-8	3.8	75
72	ZFPM2/FOG2 and HEY2 genes analysis in nonsyndromic tricuspid atresia. *American Journal of Medical Genetics, Part A*, **2005**, 133A, 68-70	2.5	12
71	Hyperthrophic cardiomyopathy and the PTPN11 gene. *American Journal of Medical Genetics, Part A*, **2005**, 136, 93-4	2.5	8
70	CRELD1 and GATA4 gene analysis in patients with nonsyndromic atrioventricular canal defects. *American Journal of Medical Genetics, Part A*, **2005**, 139, 236-8	2.5	22
69	LGI1 gene mutation screening in sporadic partial epilepsy with auditory features. *Journal of Neurology*, **2005**, 252, 62-6	5.5	10
68	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, 1069-72	5.3	44
67	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, 95-8		11
66	A homozygous GJA1 gene mutation causes a Hallermann-Streiff/ODDD spectrum phenotype. *Human Mutation*, **2004**, 23, 286	4.7	77

65	Mutations of the Nogo-66 receptor (RTN4R) gene in schizophrenia. *Human Mutation*, **2004**, 24, 534-5	4.7	69
64	LEOPARD syndrome: a new polyaneurysm association and an update on the molecular genetics of the disease. *Journal of Vascular Surgery*, **2004**, 39, 897-900	3.5	10
63	Mutational analysis of parkin gene by denaturing high-performance liquid chromatography (DHPLC) in essential tremor. *Parkinsonism and Related Disorders*, **2004**, 10, 357-62	3.6	16
62	Mutations of ZFPM2/FOG2 gene in sporadic cases of tetralogy of Fallot. *Human Mutation*, **2003**, 22, 372-7	4.7	113
61	Familial blepharospasm is inherited as an autosomal dominant trait and relates to a novel unassigned gene. *Movement Disorders*, **2003**, 18, 207-12	7	38
60	Nonsyndromic pulmonary valve stenosis and the PTPN11 gene. *American Journal of Medical Genetics Part A*, **2003**, 116A, 389-90		14
59	Epilepsy with auditory features: a LGI1 gene mutation suggests a loss-of-function mechanism. *Annals of Neurology*, **2003**, 53, 396-9	9.4	51
58	DiGeorge subtypes of nonsyndromic conotruncal defects: evidence against a major role of TBX1 gene. *European Journal of Human Genetics*, **2003**, 11, 349-51	5.3	45
57	Genetic variants of modulators of insulin action. *International Congress Series*, **2003**, 1253, 45-53		
56	Novel Italian family supports clinical and genetic heterogeneity of primary adult-onset torsion dystonia. *Movement Disorders*, **2002**, 17, 392-7	7	21
55	Cytogenetic mapping of a novel locus for type II Waardenburg syndrome. *Human Genetics*, **2002**, 110, 64-7	6.3	17
54	Assignment of a locus for autosomal dominant idiopathic scoliosis (IS) to human chromosome 17p11. *Human Genetics*, **2002**, 111, 401-4	6.3	111
53	An ATG repeat in the 3Q untranslated region of the human resistin gene is associated with a decreased risk of insulin resistance. *Journal of Clinical Endocrinology and Metabolism*, **2002**, 87, 4403-6	5.6	73
52	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, 1402-7	4.3	13
51	A variation in 3QUTR of hPTP1B increases specific gene expression and associates with insulin resistance. *American Journal of Human Genetics*, **2002**, 70, 806-12	11	165
50	Grouping of multiple-lentigines/LEOPARD and Noonan syndromes on the PTPN11 gene. *American Journal of Human Genetics*, **2002**, 71, 389-94	11	321
49	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. *Neuromuscular Disorders*, **2002**, 12, 286-91	2.9	7
48	Leiomyosarcoma of the larynx: case report with pathologic and surgical considerations. *The Journal of Otolaryngology*, **2002**, 31, 393-6		3

47	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		27
46	Genomic organization, physical mapping, and involvement in Yq microdeletions of the VCY2 (BPY 2) gene. *Genomics*, **2001**, 72, 153-7	4.3	14
45	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 Suppl 1, S69-76		4
44	The Q121 PC-1 variant and obesity have additive and independent effects in causing insulin resistance. *Journal of Clinical Endocrinology and Metabolism*, **2001**, 86, 5888-91	5.6	50
43	Narrowing the Duane syndrome critical region at chromosome 8q13 down to 40 kb. *European Journal of Human Genetics*, **2000**, 8, 319-24	5.3	29
42	A polymorphism (K121Q) of the human glycoprotein PC-1 gene coding region is strongly associated with insulin resistance. *Diabetes*, **1999**, 48, 1881-4	0.9	208
41	Genomic structure, promoter characterisation and mutational analysis of the S100A7 gene: exclusion of a candidate for familial psoriasis susceptibility. *Human Genetics*, **1999**, 104, 130-4	6.3	29
40	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, 247-9	11	30
39	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, 273-7	3.8	8
38	Induction of adhesion molecules on human schwann cells by proinflammatory cytokines, an immunofluorescence study. *Journal of the Neurological Sciences*, **1999**, 170, 124-30	3.2	21
37	Motor neurone metabolism. *Journal of the Neurological Sciences*, **1999**, 169, 161-9	3.2	13
36	Isolation and characterization of a novel transcript embedded within HIRA, a gene deleted in DiGeorge syndrome. *Molecular Genetics and Metabolism*, **1999**, 67, 227-35	3.7	17
35	Structure and expression of the human ubiquitin fusion-degradation gene (UFD1L). *Biochimica Et Biophysica Acta Gene Regulatory Mechanisms*, **1998**, 1396, 158-62		20
34	Immunomagnetic isolation of human developing motor neurons. *NeuroReport*, **1998**, 9, 1143-7	1.7	10
33	UFD1L, a developmentally expressed ubiquitination gene, is deleted in CATCH 22 syndrome. *Human Molecular Genetics*, **1997**, 6, 259-65	5.6	75
32	SMT3A, a human homologue of the S. cerevisiae SMT3 gene, maps to chromosome 21qter and defines a novel gene family. *Genomics*, **1997**, 40, 362-6	4.3	105
31	mRNA distribution in adult human brain of GRIN2B, a N-methyl-D-aspartate (NMDA) receptor subunit. *Neuroscience Letters*, **1997**, 239, 49-53	3.3	41
30	Hepatitis G virus infection in hemodialysis patients. *Kidney International*, **1997**, 51, 348-52	9.9	29

29	Expression study of survival motor neuron gene in human fetal tissues. *Biochemical and Molecular Medicine*, **1997**, 61, 102-6		27
28	Deletion analysis of SMN and NAIP genes in spinal muscular atrophy Italian families. *Muscle and Nerve*, **1996**, 19, 378-80	3.4	6
27	Deletion analysis of the simple tandem repeat loci physically linked to the spinal muscular atrophy locus. *Human Mutation*, **1996**, 7, 198-201	4.7	5
26	cDNA characterization and chromosomal mapping of two human homologues of the Drosophila dishevelled polarity gene. *Human Molecular Genetics*, **1996**, 5, 953-8	5.6	50
25	Postzygotic instability of the myotonic dystrophy p[AGC] in repeat supported by larger expansions in muscle and reduced amplifications in sperm. *Journal of Neurology*, **1995**, 242, 379-83	5.5	12
24	Identification of multiple transcribed sequences from the spinal muscular atrophy region of human chromosome 5. *Biochemical and Biophysical Research Communications*, **1995**, 206, 294-301	3.4	5
23	Survival motor neuron gene transcript analysis in muscles from spinal muscular atrophy patients. *Biochemical and Biophysical Research Communications*, **1995**, 213, 342-8	3.4	169
22	Different expression of the myotonin protein kinase gene in discrete areas of human brain. *Biochemical and Biophysical Research Communications*, **1995**, 216, 489-94	3.4	4
21	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, 115-9		5
20	High conservation of the trinucleotide [CTG]n repeat at the myotonic dystrophy locus in nonhuman primates. *Human Evolution*, **1994**, 9, 315-321		
19	Human elongation factor EF-1 beta: cloning and characterization of the EF1 beta 5a gene and assignment of EF-1 beta isoforms to chromosomes 2,5,15 and X. *Biochemical and Biophysical Research Communications*, **1993**, 197, 154-62	3.4	23
18	The myotonic dystrophy gene. *Archives of Neurology*, **1993**, 50, 1173-9		31
17	(CTG)n triplet mutation and phenotype manifestations in myotonic dystrophy patients. *Biochemical Medicine and Metabolic Biology*, **1993**, 50, 85-92		37
16	A transposon-like element in the deletion-prone region of the dystrophin gene. *Genomics*, **1992**, 13, 594-600	4.9	37
15	Point mutations and polymorphisms in the human dystrophin gene identified in genomic DNA sequences amplified by multiplex PCR. *Human Genetics*, **1992**, 89, 253-8	6.3	45
14	Characterization of a murine gene expressed from the inactive X chromosome. *Nature*, **1991**, 351, 325-9	50.4	471
13	Pentanucleotide repeat length polymorphism at the human CD4 locus. *Nucleic Acids Research*, **1991**, 19, 4791	20.1	68
12	Human fetal brain beta-nerve growth factor cDNA: molecular cloning of 5′ and 3′ untranslated regions. *Neuroscience Letters*, **1991**, 127, 117-20	3.3	2

11	Variation of the CGG repeat at the fragile X site results in genetic instability: resolution of the Sherman paradox. *Cell*, **1991**, 67, 1047-58	56.2	1786
10	Identification of a gene (FMR-1) containing a CGG repeat coincident with a breakpoint cluster region exhibiting length variation in fragile X syndrome. *Cell*, **1991**, 65, 905-14	56.2	2848
9	Adrenal medulla autograft in 3 parkinsonian patients: results using two different approaches. *Progress in Brain Research*, **1990**, 82, 677-82	2.9	4
8	Effect of nerve growth factor in adrenal autografts in parkinsonism. *Annals of Neurology*, **1990**, 27, 341-2	9.4	108
7	Detection of beta-nerve growth factor mRNA in the human fetal brain. *Brain Research*, **1990**, 518, 337-41	3.7	11
6	cDNA sequence of human beta-NGF. *Nucleic Acids Research*, **1990**, 18, 4020	20.1	11
5	Human neuronal cell viability demonstrated in culture after cryopreservation. *Brain Research*, **1988**, 473, 169-74	3.7	43
4	Cryopreservation of human fetal adrenal medullary cells. *Brain Research*, **1988**, 454, 383-6	3.7	9
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