

Marcella Devoto

List of Publications by Year in Descending Order

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

136
papers

9,510
citations

45
h-index

96
g-index

143
ext. papers

10,643
ext. citations

9.6
avg, IF

4.85
L-index

#	Paper	IF	Citations
136	Variants in STXBP3 are Associated with Very Early Onset Inflammatory Bowel Disease, Bilateral Sensorineural Hearing Loss and Immune Dysregulation. <i>Journal of Crohn's and Colitis</i> , 2021 , 15, 1908-1919	19.5	0
135	Immune Dysregulation in Human ITCH Deficiency Successfully Treated with Hematopoietic Cell Transplantation. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2021 , 9, 2885-2893.e3	5.4	0
134	Colonoids From Patients With Pediatric Inflammatory Bowel Disease Exhibit Decreased Growth Associated With Inflammation Severity and Durable Upregulation of Antigen Presentation Genes. <i>Inflammatory Bowel Diseases</i> , 2021 , 27, 256-267	4.5	1
133	The Unique Disease Course of Children with Very Early onset-Inflammatory Bowel Disease. <i>Inflammatory Bowel Diseases</i> , 2020 , 26, 909-918	4.5	14
132	Impaired Redox and Protein Homeostasis as Risk Factors and Therapeutic Targets in Toxin-Induced Biliary Atresia. <i>Gastroenterology</i> , 2020 , 159, 1068-1084.e2	13.3	0
131	Exome Sequencing in Individuals with Isolated Biliary Atresia. <i>Scientific Reports</i> , 2020 , 10, 2709	4.9	7
130	Rare copy number variants in over 100,000 European ancestry subjects reveal multiple disease associations. <i>Nature Communications</i> , 2020 , 11, 255	17.4	17
129	Complex genetic signatures in immune cells underlie autoimmunity and inform therapy. <i>Nature Genetics</i> , 2020 , 52, 1036-1045	36.3	16
128	Neural crest-derived tumor neuroblastoma and melanoma share 1p13.2 as susceptibility locus that shows a long-range interaction with the SLC16A1 gene. <i>Carcinogenesis</i> , 2020 , 41, 284-295	4.6	11
127	Exploring Shared Susceptibility between Two Neural Crest Cells Originating Conditions: Neuroblastoma and Congenital Heart Disease. <i>Genes</i> , 2019 , 10,	4.2	8
126	Expanding the spectrum of genes responsible for hereditary motor neuropathies. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2019 , 90, 1171-1179	5.5	19
125	An Exploratory Association Analysis of the Insulin Gene Region With Diabetes Mellitus in Two Dog Breeds. <i>Journal of Heredity</i> , 2019 , 110, 793-800	2.4	2
124	Subphenotype meta-analysis of testicular cancer genome-wide association study data suggests a role for RBFOX family genes in cryptorchidism susceptibility. <i>Human Reproduction</i> , 2018 , 33, 967-977	5.7	7
123	Fine mapping of 2q35 high-risk neuroblastoma locus reveals independent functional risk variants and suggests full-length BARD1 as tumor-suppressor. <i>International Journal of Cancer</i> , 2018 , 143, 2828-2837	7.5	34
122	A genome-wide association study identifies a susceptibility locus for biliary atresia on 2p16.1 within the gene EFEMP1. <i>PLoS Genetics</i> , 2018 , 14, e1007532	6	23
121	Commentary on Mutations in Interleukin-10 Receptor and Clinical Phenotypes in Patients with Very Early-onset Inflammatory Bowel Disease: A Chinese VEO-IBD Collaboration Group Survey. <i>Inflammatory Bowel Diseases</i> , 2017 , 23, 591-592	4.5	2
120	Novel ZBTB24 Mutation Associated with Immunodeficiency, Centromere Instability, and Facial Anomalies Type-2 Syndrome Identified in a Patient with Very Early Onset Inflammatory Bowel Disease. <i>Inflammatory Bowel Diseases</i> , 2017 , 23, 2252-2255	4.5	15

119	Common variants upstream of MLF1 at 3q25 and within CPZ at 4p16 associated with neuroblastoma. <i>PLoS Genetics</i> , 2017 , 13, e1006787	6	40
118	IQ and hemizyosity for the Val Met functional polymorphism of COMT in 22q11DS. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2016 , 171, 1112-1115	3.5	5
117	Variants of the ACTG2 gene correlate with degree of severity and presence of megacystis in chronic intestinal pseudo-obstruction. <i>European Journal of Human Genetics</i> , 2016 , 24, 1211-5	5.3	27
116	P-197 Identification of a Homozygous Mutation in the ZBTB24 Gene in a Patient with Very Early Onset Inflammatory Bowel Disease. <i>Inflammatory Bowel Diseases</i> , 2016 , 22, S68	4.5	
115	O-003 Understanding the Relevance of Whole Exome Sequencing Identified Variants in Patients with Very Early-Onset-IBD. <i>Inflammatory Bowel Diseases</i> , 2016 , 22, S1-S2	4.5	
114	Rare copy number variants and congenital heart defects in the 22q11.2 deletion syndrome. <i>Human Genetics</i> , 2016 , 135, 273-85	6.3	31
113	Polygenic inheritance of cryptorchidism susceptibility in the LE/orl rat. <i>Molecular Human Reproduction</i> , 2016 , 22, 18-34	4.4	7
112	Compound heterozygous mutations in NEK8 in siblings with end-stage renal disease with hepatic and cardiac anomalies. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 750-3	2.5	17
111	Genomic copy number variation association study in Caucasian patients with nonsyndromic cryptorchidism. <i>BMC Urology</i> , 2016 , 16, 62	2.2	4
110	Is a Candidate Modifier of Liver Disease Severity in Alagille Syndrome. <i>Cellular and Molecular Gastroenterology and Hepatology</i> , 2016 , 2, 663-675.e2	7.9	21
109	Phenotype specific association of the TGFBR3 locus with nonsyndromic cryptorchidism. <i>Journal of Urology</i> , 2015 , 193, 1637-45	2.5	16
108	Integration of Omics Data in Genetic Epidemiology. <i>Human Heredity</i> , 2015 , 79, 109-10	1.1	4
107	Pathway analysis supports association of nonsyndromic cryptorchidism with genetic loci linked to cytoskeleton-dependent functions. <i>Human Reproduction</i> , 2015 , 30, 2439-51	5.7	19
106	Exome sequencing analysis reveals variants in primary immunodeficiency genes in patients with very early onset inflammatory bowel disease. <i>Gastroenterology</i> , 2015 , 149, 1415-24	13.3	68
105	Copy-Number Variation of the Glucose Transporter Gene SLC2A3 and Congenital Heart Defects in the 22q11.2 Deletion Syndrome. <i>American Journal of Human Genetics</i> , 2015 , 96, 753-64	11	54
104	Association of a Novel ACTA1 Mutation With a Dominant Progressive Scapulooperoneal Myopathy in an Extended Family. <i>JAMA Neurology</i> , 2015 , 72, 689-98	17.2	27
103	Heterozygous deletion of FOXA2 segregates with disease in a family with heterotaxy, panhypopituitarism, and biliary atresia. <i>Human Mutation</i> , 2015 , 36, 631-7	4.7	31
102	Exome sequencing reveals compound heterozygous mutations in ATP8B1 in a JAG1/NOTCH2 mutation-negative patient with clinically diagnosed Alagille syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 891-3	2.5	9

101	A de novo whole gene deletion of XIAP detected by exome sequencing analysis in very early onset inflammatory bowel disease: a case report. <i>BMC Gastroenterology</i> , 2015 , 15, 160	3	31
100	Rare variants in TP53 and susceptibility to neuroblastoma. <i>Journal of the National Cancer Institute</i> , 2014 , 106, dju047	9.7	83
99	Common genetic variants in NEFL influence gene expression and neuroblastoma risk. <i>Cancer Research</i> , 2014 , 74, 6913-24	10.1	69
98	Recessive and dominant mutations in COL12A1 cause a novel EDS/myopathy overlap syndrome in humans and mice. <i>Human Molecular Genetics</i> , 2014 , 23, 2339-52	5.6	78
97	Replication of a GWAS signal in a Caucasian population implicates ADD3 in susceptibility to biliary atresia. <i>Human Genetics</i> , 2014 , 133, 235-43	6.3	45
96	A candidate gene approach to identify modifiers of the palatal phenotype in 22q11.2 deletion syndrome patients. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2013 , 77, 123-7	1.7	11
95	Novel COCH mutation in a family with autosomal dominant late onset sensorineural hearing impairment and tinnitus. <i>American Journal of Otolaryngology - Head and Neck Medicine and Surgery</i> , 2013 , 34, 230-5	2.8	20
94	Evidence from human and zebrafish that GPC1 is a biliary atresia susceptibility gene. <i>Gastroenterology</i> , 2013 , 144, 1107-1115.e3	13.3	87
93	Developmental dysplasia of the hip: linkage mapping and whole exome sequencing identify a shared variant in CX3CR1 in all affected members of a large multigeneration family. <i>Journal of Bone and Mineral Research</i> , 2013 , 28, 2540-9	6.3	36
92	Replication of GWAS-identified neuroblastoma risk loci strengthens the role of BARD1 and affirms the cumulative effect of genetic variations on disease susceptibility. <i>Carcinogenesis</i> , 2013 , 34, 605-11	4.6	82
91	Computerized neurocognitive profile in young people with 22q11.2 deletion syndrome compared to youths with schizophrenia and at-risk for psychosis. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2012 , 159B, 87-93	3.5	31
90	Overt cleft palate phenotype and TBX1 genotype correlations in velo-cardio-facial/DiGeorge/22q11.2 deletion syndrome patients. <i>American Journal of Medical Genetics, Part A</i> , 2012 , 158A, 2781-7	2.5	19
89	Common variation at 6q16 within HACE1 and LIN28B influences susceptibility to neuroblastoma. <i>Nature Genetics</i> , 2012 , 44, 1126-30	36.3	198
88	Genetic mapping of quantitative trait loci for disease-related phenotypes. <i>Methods in Molecular Biology</i> , 2012 , 871, 281-311	1.4	2
87	Replication of neuroblastoma SNP association at the BARD1 locus in African-Americans. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2012 , 21, 658-63	4	48
86	Common variation at BARD1 results in the expression of an oncogenic isoform that influences neuroblastoma susceptibility and oncogenicity. <i>Cancer Research</i> , 2012 , 72, 2068-78	10.1	75
85	Genetic modifiers of β -thalassemia and clinical severity as assessed by age at first transfusion. <i>Haematologica</i> , 2012 , 97, 989-93	6.6	51
84	Genome-wide copy number variation study associates metabotropic glutamate receptor gene networks with attention deficit hyperactivity disorder. <i>Nature Genetics</i> , 2011 , 44, 78-84	36.3	279

83	Phenotype restricted genome-wide association study using a gene-centric approach identifies three low-risk neuroblastoma susceptibility Loci. <i>PLoS Genetics</i> , 2011 , 7, e1002026	6	123
82	Integrative genomics identifies LMO1 as a neuroblastoma oncogene. <i>Nature</i> , 2011 , 469, 216-20	50.4	231
81	Genotype and cardiovascular phenotype correlations with TBX1 in 1,022 velo-cardio-facial/DiGeorge/22q11.2 deletion syndrome patients. <i>Human Mutation</i> , 2011 , 32, 1278-89	4.7	48
80	Genome-wide linkage analysis to identify genetic modifiers of ALK mutation penetrance in familial neuroblastoma. <i>Human Heredity</i> , 2011 , 71, 135-9	1.1	23
79	Next-generation linkage analysis. <i>Human Heredity</i> , 2011 , 72, 227	1.1	4
78	Variants of DENND1B associated with asthma in children. <i>New England Journal of Medicine</i> , 2010 , 362, 36-44	59.2	261
77	The Otto Aufranc Award: Identification of a 4 Mb region on chromosome 17q21 linked to developmental dysplasia of the hip in one 18-member, multigeneration family. <i>Clinical Orthopaedics and Related Research</i> , 2010 , 468, 337-44	2.2	39
76	Analysis of GWAS top hits in ADHD suggests association to two polymorphisms located in genes expressed in the cerebellum. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2010 , 153B, 1127-33	3.5	17
75	Comparative analysis of different approaches for dealing with candidate regions in the context of a genome-wide association study. <i>BMC Proceedings</i> , 2009 , 3 Suppl 7, S93	2.3	4
74	Nocturnal enuresis: a suggestive endophenotype marker for a subgroup of inattentive attention-deficit/hyperactivity disorder. <i>Journal of Pediatrics</i> , 2009 , 155, 239-44.e5	3.6	41
73	Copy number variation at 1q21.1 associated with neuroblastoma. <i>Nature</i> , 2009 , 459, 987-91	50.4	285
72	Common variations in BARD1 influence susceptibility to high-risk neuroblastoma. <i>Nature Genetics</i> , 2009 , 41, 718-23	36.3	226
71	Candidate gene analysis in an on-going genome-wide association study of attention-deficit hyperactivity disorder: suggestive association signals in ADRA1A. <i>Psychiatric Genetics</i> , 2009 , 19, 134-41	2.9	27
70	Identification of ALK as a major familial neuroblastoma predisposition gene. <i>Nature</i> , 2008 , 455, 930-5	50.4	960
69	Association of a polymorphic variant of the adiponectin gene with insulin resistance in african americans. <i>Clinical and Translational Science</i> , 2008 , 1, 194-9	4.9	6
68	Estimates of genetic and environmental contribution to 43 quantitative traits support sharing of a homogeneous environment in an isolated population from South Tyrol, Italy. <i>Human Heredity</i> , 2008 , 65, 175-82	1.1	28
67	Advances in family-based association analysis. Introduction. <i>Human Heredity</i> , 2008 , 66, 65-6	1.1	3
66	A novel susceptibility locus for type 1 diabetes on Chr12q13 identified by a genome-wide association study. <i>Diabetes</i> , 2008 , 57, 1143-6	0.9	118

65	Chromosome 6p22 locus associated with clinically aggressive neuroblastoma. <i>New England Journal of Medicine</i> , 2008 , 358, 2585-93	59.2	224
64	Variants of ST8SIA1 are associated with risk of developing multiple sclerosis. <i>PLoS ONE</i> , 2008 , 3, e2653	3.7	10
63	Genetic loci linked to type 1 diabetes and multiple sclerosis families in Sardinia. <i>BMC Medical Genetics</i> , 2008 , 9, 3	2.1	18
62	Association of variants of the interleukin-23 receptor gene with susceptibility to pediatric Crohn's disease. <i>Clinical Gastroenterology and Hepatology</i> , 2007 , 5, 972-6	6.9	53
61	A novel locus for syndromic chronic idiopathic intestinal pseudo-obstruction maps to chromosome 8q23-q24. <i>European Journal of Human Genetics</i> , 2007 , 15, 889-97	5.3	24
60	A genome-wide association study identifies KIAA0350 as a type 1 diabetes gene. <i>Nature</i> , 2007 , 448, 591-4	40.4	424
59	ADHD genetics: 2007 update. <i>Current Psychiatry Reports</i> , 2007 , 9, 434-9	9.1	31
58	Association of the T300A non-synonymous variant of the ATG16L1 gene with susceptibility to paediatric Crohn's disease. <i>Gut</i> , 2007 , 56, 1171-3	19.2	53
57	Genetic predisposition to familial neuroblastoma: identification of two novel genomic regions at 2p and 12p. <i>Human Heredity</i> , 2007 , 63, 205-11	1.1	31
56	IRAK-M is involved in the pathogenesis of early-onset persistent asthma. <i>American Journal of Human Genetics</i> , 2007 , 80, 1103-14	11	125
55	Meta-analysis of genome-wide scans provides evidence for sex- and site-specific regulation of bone mass. <i>Journal of Bone and Mineral Research</i> , 2007 , 22, 173-183	6.3	128
54	Genetic linkage study of high-grade myopia in a Hutterite population from South Dakota. <i>Molecular Vision</i> , 2007 , 13, 229-36	2.3	57
53	Contribution of IBD5 locus to clinical features of IBD patients. <i>American Journal of Gastroenterology</i> , 2006 , 101, 318-25	0.7	24
52	Ophthalmologic findings in Cornelia de Lange syndrome: a genotype-phenotype correlation study. <i>JAMA Ophthalmology</i> , 2006 , 124, 552-7		21
51	Statistical tools for linkage analysis and genetic association studies. <i>Expert Review of Molecular Diagnostics</i> , 2005 , 5, 781-96	3.8	11
50	Univariate and bivariate variance component linkage analysis of a whole-genome scan for loci contributing to bone mineral density. <i>European Journal of Human Genetics</i> , 2005 , 13, 781-8	5.3	37
49	A common haplotype at the 5' end of the RET proto-oncogene, overrepresented in Hirschsprung patients, is associated with reduced gene expression. <i>Human Mutation</i> , 2005 , 25, 189-95	4.7	44
48	Precocious sister chromatid separation (PSCS) in Cornelia de Lange syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2005 , 138, 27-31	2.5	72

47	Identification of a novel locus on 2q for autosomal dominant high-grade myopia. <i>Investigative Ophthalmology and Visual Science</i> , 2005 , 46, 2300-7		100
46	Heterogeneity in the magnitude of the insulin gene effect on HLA risk in type 1 diabetes. <i>Diabetes</i> , 2004 , 53, 3286-91	0.9	24
45	A genotype-phenotype correlation with gender-effect for hearing impairment caused by TECTA mutations. <i>Cellular Physiology and Biochemistry</i> , 2004 , 14, 369-76	3.9	46
44	Cornelia de Lange syndrome is caused by mutations in NIPBL, the human homolog of Drosophila melanogaster Nipped-B. <i>Nature Genetics</i> , 2004 , 36, 631-5	36.3	560
43	NIPBL mutational analysis in 120 individuals with Cornelia de Lange syndrome and evaluation of genotype-phenotype correlations. <i>American Journal of Human Genetics</i> , 2004 , 75, 610-23	11	245
42	CARD15 genotyping in inflammatory bowel disease patients by multiplex pyrosequencing. <i>Clinical Chemistry</i> , 2003 , 49, 1675-9	5.5	29
41	New locus for autosomal dominant high myopia maps to the long arm of chromosome 17. <i>Investigative Ophthalmology and Visual Science</i> , 2003 , 44, 1830-6		149
40	Linkage analysis in families with recurrent neuroblastoma. <i>Annals of the New York Academy of Sciences</i> , 2002 , 963, 74-84	6.5	16
39	Weak linkage at 4p16 to predisposition for human neuroblastoma. <i>Oncogene</i> , 2002 , 21, 8356-60	9.2	34
38	Second family with hearing impairment linked to 19q13 and refined DFNA4 localisation. <i>European Journal of Human Genetics</i> , 2002 , 10, 95-9	5.3	11
37	Linkage analysis conditional on HLA status in a large North American pedigree supports the presence of a multiple sclerosis susceptibility locus on chromosome 12p12. <i>Human Molecular Genetics</i> , 2002 , 11, 295-300	5.6	27
36	A rare haplotype of the RET proto-oncogene is a risk-modifying allele in hirschsprung disease. <i>American Journal of Human Genetics</i> , 2002 , 71, 969-74	11	39
35	Comparison of sib pair-based approaches for identifying quantitative trait loci underlying asthma in the Busselton families. <i>Genetic Epidemiology</i> , 2001 , 21 Suppl 1, S198-203	2.6	3
34	Exclusion of linkage to the CDL1 gene region on chromosome 3q26.3 in some familial cases of Cornelia de Lange syndrome. <i>American Journal of Medical Genetics Part A</i> , 2001 , 101, 120-129		26
33	Novel X-linked mental retardation syndrome with short stature maps to Xq24. <i>American Journal of Medical Genetics Part A</i> , 2001 , 103, 1-8		12
32	Charcot-Marie-Tooth type 4B is caused by mutations in the gene encoding myotubularin-related protein-2. <i>Nature Genetics</i> , 2000 , 25, 17-9	36.3	419
31	A single-nucleotide polymorphic variant of the RET proto-oncogene is underrepresented in sporadic Hirschsprung disease. <i>European Journal of Human Genetics</i> , 2000 , 8, 721-4	5.3	42
30	Combined segregation and linkage analysis of inflammatory bowel disease in the IBD1 region using severity to characterise Crohn's disease and ulcerative colitis. On behalf of the GISC. <i>European Journal of Human Genetics</i> , 2000 , 8, 846-52	5.3	16

29	A refined physical and transcriptional map of the SPG9 locus on 10q23.3-q24.2. <i>European Journal of Human Genetics</i> , 2000 , 8, 777-82	5.3	15
28	Genetic refinement and physical mapping of the CMT4B gene on chromosome 11q22. <i>Genomics</i> , 2000 , 63, 271-8	4.3	17
27	Linkage study of early-onset obesity to leptin receptor gene in Italian children. <i>Nutrition Research</i> , 2000 , 20, 1059-1063	4	2
26	Genetic analysis in Italian families with inflammatory bowel disease supports linkage to the IBD1 locus—a GISC study. <i>European Journal of Human Genetics</i> , 1999 , 7, 567-73	5.3	73
25	Genetic mapping to 10q23.3-q24.2, in a large Italian pedigree, of a new syndrome showing bilateral cataracts, gastroesophageal reflux, and spastic paraparesis with amyotrophy. <i>American Journal of Human Genetics</i> , 1999 , 64, 586-93	11	97
24	Five families with arginine 519-cysteine mutation in COL2A1: evidence for three distinct founders. <i>Human Mutation</i> , 1998 , 12, 172-6	4.7	23
23	A common mutation in Sardinian autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy patients. <i>Human Genetics</i> , 1998 , 103, 428-34	6.3	192
22	Polymorphisms of alpha-adducin and salt sensitivity in patients with essential hypertension. <i>Lancet, The</i> , 1997 , 349, 1353-7	4.0	473
21	A linkage study of schizophrenia to markers within Xp11 near the MAOB gene. <i>Psychiatry Research</i> , 1997 , 70, 131-43	9.9	55
20	Exclusion of linkage between RET and neuronal intestinal dysplasia type B. <i>American Journal of Medical Genetics Part A</i> , 1996 , 62, 195-8		15
19	A common molecular basis for three inherited kidney stone diseases. <i>Nature</i> , 1996 , 379, 445-9	50.4	614
18	Gender-dependent disease severity in autosomal polycystic kidney disease of rats. <i>Kidney International</i> , 1995 , 48, 496-500	9.9	34
17	Analysis of linkage disequilibrium between different cystic fibrosis mutations and three intragenic microsatellites in the Italian population. <i>Human Mutation</i> , 1995 , 5, 23-7	4.7	9
16	Search for linkage to schizophrenia on the X and Y chromosomes. <i>American Journal of Medical Genetics Part A</i> , 1994 , 54, 113-21		60
15	Close linkage with the RET protooncogene and boundaries of deletion mutations in autosomal dominant Hirschsprung disease. <i>Human Molecular Genetics</i> , 1993 , 2, 1803-8	5.6	77
14	Mapping a gene for familial situs abnormalities to human chromosome Xq24-q27.1. <i>Nature Genetics</i> , 1993 , 5, 403-7	36.3	122
13	A nonsense mutation (R1158X) and a splicing mutation (3849 + 4A----G) in exon 19 of the cystic fibrosis transmembrane conductance regulator gene. <i>Genomics</i> , 1992 , 12, 417-8	4.3	21
12	Four new mutations of the CFTR gene (541delC, R347H, R352Q, E585X) detected by DGGE analysis in Italian CF patients, associated with different clinical phenotypes. <i>Human Mutation</i> , 1992 , 1, 314-9	4.7	39

11	Frequency of the delta F508 mutation in a sample of 175 Italian cystic fibrosis patients. <i>Human Genetics</i> , 1990 , 85, 400-2	6.3	9
10	Preliminary results on the frequency of the delta F508 mutation in cystic fibrosis patients from the USSR. <i>Human Genetics</i> , 1990 , 85, 423-4	6.3	
9	Genetic differences in cystic fibrosis patients with and without pancreatic insufficiency. An Italian collaborative study. <i>Human Genetics</i> , 1990 , 84, 435-8	6.3	10
8	Haplotype distribution and molecular defects at the phenylalanine hydroxylase locus in Italy. <i>Human Genetics</i> , 1990 , 86, 69-72	6.3	15
7	Prenatal diagnosis and linkage disequilibrium with cystic fibrosis for markers surrounding D7S8. <i>Human Genetics</i> , 1990 , 85, 275-8	6.3	2
6	Why is the cystic fibrosis gene so frequent?. <i>Human Genetics</i> , 1989 , 84, 1-5	6.3	84
5	N-myc oncogene amplification and catecholamine metabolism in patients with neuroblastoma. <i>Lancet, The</i> , 1987 , 2, 795	4.0	1
4	Frequency of consanguineous marriages among parents and grandparents of Down patients. <i>Human Genetics</i> , 1985 , 70, 256-8	6.3	12
3	Attention-deficit hyperactivity disorder 168-182		
2	Impaired redox and protein homeostasis as risk factors and therapeutic targets in toxin-induced biliary atresia		1
1	Sequencing of over 100,000 individuals identifies multiple genes and rare variants associated with Crohns disease susceptibility		2