# Marcella Devoto

## List of Publications by Citations



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| 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 | Identification of ALK as a major familial neuroblastoma predisposition gene. *Nature*, **2008**, 455, 930-5 | 50.4 | 960 |
| 135 | A common molecular basis for three inherited kidney stone diseases. *Nature*, **1996**, 379, 445-9 | 50.4 | 614 |
| 134 | Cornelia de Lange syndrome is caused by mutations in NIPBL, the human homolog of Drosophila melanogaster Nipped-B. *Nature Genetics*, **2004**, 36, 631-5 | 36.3 | 560 |
| 133 | Polymorphisms of alpha-adducin and salt sensitivity in patients with essential hypertension. *Lancet, The*, **1997**, 349, 1353-7 | 40 | 473 |
| 132 | A genome-wide association study identifies KIAA0350 as a type 1 diabetes gene. *Nature*, **2007**, 448, 591-4 | 50.4 | 424 |
| 131 | Charcot-Marie-Tooth type 4B is caused by mutations in the gene encoding myotubularin-related protein-2. *Nature Genetics*, **2000**, 25, 17-9 | 36.3 | 419 |
| 130 | Copy number variation at 1q21.1 associated with neuroblastoma. *Nature*, **2009**, 459, 987-91 | 50.4 | 285 |
| 129 | Genome-wide copy number variation study associates metabotropic glutamate receptor gene networks with attention deficit hyperactivity disorder. *Nature Genetics*, **2011**, 44, 78-84 | 36.3 | 279 |
| 128 | Variants of DENND1B associated with asthma in children. *New England Journal of Medicine*, **2010**, 362, 36-44 | 59.2 | 261 |
| 127 | NIPBL mutational analysis in 120 individuals with Cornelia de Lange syndrome and evaluation of genotype-phenotype correlations. *American Journal of Human Genetics*, **2004**, 75, 610-23 | 11 | 245 |
| 126 | Integrative genomics identifies LMO1 as a neuroblastoma oncogene. *Nature*, **2011**, 469, 216-20 | 50.4 | 231 |
| 125 | Common variations in BARD1 influence susceptibility to high-risk neuroblastoma. *Nature Genetics*, **2009**, 41, 718-23 | 36.3 | 226 |
| 124 | Chromosome 6p22 locus associated with clinically aggressive neuroblastoma. *New England Journal of Medicine*, **2008**, 358, 2585-93 | 59.2 | 224 |
| 123 | Common variation at 6q16 within HACE1 and LIN28B influences susceptibility to neuroblastoma. *Nature Genetics*, **2012**, 44, 1126-30 | 36.3 | 198 |
| 122 | A common mutation in Sardinian autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy patients. *Human Genetics*, **1998**, 103, 428-34 | 6.3 | 192 |
| 121 | New locus for autosomal dominant high myopia maps to the long arm of chromosome 17. *Investigative Ophthalmology and Visual Science*, **2003**, 44, 1830-6 | | 149 |
| 120 | Meta-analysis of genome-wide scans provides evidence for sex- and site-specific regulation of bone mass. *Journal of Bone and Mineral Research*, **2007**, 22, 173-183 | 6.3 | 128 |

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| 119 | IRAK-M is involved in the pathogenesis of early-onset persistent asthma. *American Journal of Human Genetics*, **2007**, 80, 1103-14 | 11 | 125 |
| 118 | Phenotype restricted genome-wide association study using a gene-centric approach identifies three low-risk neuroblastoma susceptibility Loci. *PLoS Genetics*, **2011**, 7, e1002026 | 6 | 123 |
| 117 | Mapping a gene for familial situs abnormalities to human chromosome Xq24-q27.1. *Nature Genetics* , **1993**, 5, 403-7 | 36.3 | 122 |
| 116 | A novel susceptibility locus for type 1 diabetes on Chr12q13 identified by a genome-wide association study. *Diabetes*, **2008**, 57, 1143-6 | 0.9 | 118 |
| 115 | Identification of a novel locus on 2q for autosomal dominant high-grade myopia. *Investigative Ophthalmology and Visual Science*, **2005**, 46, 2300-7 | | 100 |
| 114 | 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. *American Journal of Human Genetics*, **1999**, 64, 586-93 | 11 | 97 |
| 113 | Evidence from human and zebrafish that GPC1 is a biliary atresia susceptibility gene. *Gastroenterology*, **2013**, 144, 1107-1115.e3 | 13.3 | 87 |
| 112 | Why is the cystic fibrosis gene so frequent?. *Human Genetics*, **1989**, 84, 1-5 | 6.3 | 84 |
| 111 | Rare variants in TP53 and susceptibility to neuroblastoma. *Journal of the National Cancer Institute*, **2014**, 106, dju047 | 9.7 | 83 |
| 110 | Replication of GWAS-identified neuroblastoma risk loci strengthens the role of BARD1 and affirms the cumulative effect of genetic variations on disease susceptibility. *Carcinogenesis*, **2013**, 34, 605-11 | 4.6 | 82 |
| 109 | Recessive and dominant mutations in COL12A1 cause a novel EDS/myopathy overlap syndrome in humans and mice. *Human Molecular Genetics*, **2014**, 23, 2339-52 | 5.6 | 78 |
| 108 | Close linkage with the RET protooncogene and boundaries of deletion mutations in autosomal dominant Hirschsprung disease. *Human Molecular Genetics*, **1993**, 2, 1803-8 | 5.6 | 77 |
| 107 | Common variation at BARD1 results in the expression of an oncogenic isoform that influences neuroblastoma susceptibility and oncogenicity. *Cancer Research*, **2012**, 72, 2068-78 | 10.1 | 75 |
| 106 | Genetic analysis in Italian families with inflammatory bowel disease supports linkage to the IBD1 locus--a GISC study. *European Journal of Human Genetics*, **1999**, 7, 567-73 | 5.3 | 73 |
| 105 | Precocious sister chromatid separation (PSCS) in Cornelia de Lange syndrome. *American Journal of Medical Genetics, Part A*, **2005**, 138, 27-31 | 2.5 | 72 |
| 104 | Common genetic variants in NEFL influence gene expression and neuroblastoma risk. *Cancer Research*, **2014**, 74, 6913-24 | 10.1 | 69 |
| 103 | Exome sequencing analysis reveals variants in primary immunodeficiency genes in patients with very early onset inflammatory bowel disease. *Gastroenterology*, **2015**, 149, 1415-24 | 13.3 | 68 |
| 102 | Search for linkage to schizophrenia on the X and Y chromosomes. *American Journal of Medical Genetics Part A*, **1994**, 54, 113-21 | | 60 |

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|---|---|---|---|
| 101 | Genetic linkage study of high-grade myopia in a Hutterite population from South Dakota. *Molecular Vision*, **2007**, 13, 229-36 | 2.3 | 57 |
| 100 | A linkage study of schizophrenia to markers within Xp11 near the MAOB gene. *Psychiatry Research*, **1997**, 70, 131-43 | 9.9 | 55 |
| 99 | Copy-Number Variation of the Glucose Transporter Gene SLC2A3 and Congenital Heart Defects in the 22q11.2 Deletion Syndrome. *American Journal of Human Genetics*, **2015**, 96, 753-64 | 11 | 54 |
| 98 | Association of variants of the interleukin-23 receptor gene with susceptibility to pediatric Crohn's disease. *Clinical Gastroenterology and Hepatology*, **2007**, 5, 972-6 | 6.9 | 53 |
| 97 | Association of the T300A non-synonymous variant of the ATG16L1 gene with susceptibility to paediatric Crohn's disease. *Gut*, **2007**, 56, 1171-3 | 19.2 | 53 |
| 96 | Genetic modifiers of β-thalassemia and clinical severity as assessed by age at first transfusion. *Haematologica*, **2012**, 97, 989-93 | 6.6 | 51 |
| 95 | Genotype and cardiovascular phenotype correlations with TBX1 in 1,022 velo-cardio-facial/DiGeorge/22q11.2 deletion syndrome patients. *Human Mutation*, **2011**, 32, 1278-89 | 4.7 | 48 |
| 94 | Replication of neuroblastoma SNP association at the BARD1 locus in African-Americans. *Cancer Epidemiology Biomarkers and Prevention*, **2012**, 21, 658-63 | 4 | 48 |
| 93 | A genotype-phenotype correlation with gender-effect for hearing impairment caused by TECTA mutations. *Cellular Physiology and Biochemistry*, **2004**, 14, 369-76 | 3.9 | 46 |
| 92 | Replication of a GWAS signal in a Caucasian population implicates ADD3 in susceptibility to biliary atresia. *Human Genetics*, **2014**, 133, 235-43 | 6.3 | 45 |
| 91 | A common haplotype at the 5' end of the RET proto-oncogene, overrepresented in Hirschsprung patients, is associated with reduced gene expression. *Human Mutation*, **2005**, 25, 189-95 | 4.7 | 44 |
| 90 | A single-nucleotide polymorphic variant of the RET proto-oncogene is underrepresented in sporadic Hirschsprung disease. *European Journal of Human Genetics*, **2000**, 8, 721-4 | 5.3 | 42 |
| 89 | Nocturnal enuresis: a suggestive endophenotype marker for a subgroup of inattentive attention-deficit/hyperactivity disorder. *Journal of Pediatrics*, **2009**, 155, 239-44.e5 | 3.6 | 41 |
| 88 | Common variants upstream of MLF1 at 3q25 and within CPZ at 4p16 associated with neuroblastoma. *PLoS Genetics*, **2017**, 13, e1006787 | 6 | 40 |
| 87 | 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. *Clinical Orthopaedics and Related Research*, **2010**, 468, 337-44 | 2.2 | 39 |
| 86 | A rare haplotype of the RET proto-oncogene is a risk-modifying allele in hirschsprung disease. *American Journal of Human Genetics*, **2002**, 71, 969-74 | 11 | 39 |
| 85 | Four new mutations of the CFTR gene (541delC, R347H, R352Q, E585X) detected by DGGE analysis in Italian CF patients, associated with different clinical phenotypes. *Human Mutation*, **1992**, 1, 314-9 | 4.7 | 39 |
| 84 | Univariate and bivariate variance component linkage analysis of a whole-genome scan for loci contributing to bone mineral density. *European Journal of Human Genetics*, **2005**, 13, 781-8 | 5.3 | 37 |

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| 83 | 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. *Journal of Bone and Mineral Research*, **2013**, 28, 2540-9 | 6.3 | 36 |
| 82 | Fine mapping of 2q35 high-risk neuroblastoma locus reveals independent functional risk variants and suggests full-length BARD1 as tumor-suppressor. *International Journal of Cancer*, **2018**, 143, 2828-2837 | 7.5 | 34 |
| 81 | Weak linkage at 4p16 to predisposition for human neuroblastoma. *Oncogene*, **2002**, 21, 8356-60 | 9.2 | 34 |
| 80 | Gender-dependent disease severity in autosomal polycystic kidney disease of rats. *Kidney International*, **1995**, 48, 496-500 | 9.9 | 34 |
| 79 | Heterozygous deletion of FOXA2 segregates with disease in a family with heterotaxy, panhypopituitarism, and biliary atresia. *Human Mutation*, **2015**, 36, 631-7 | 4.7 | 31 |
| 78 | Rare copy number variants and congenital heart defects in the 22q11.2 deletion syndrome. *Human Genetics*, **2016**, 135, 273-85 | 6.3 | 31 |
| 77 | Computerized neurocognitive profile in young people with 22q11.2 deletion syndrome compared to youths with schizophrenia and at-risk for psychosis. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2012**, 159B, 87-93 | 3.5 | 31 |
| 76 | A de novo whole gene deletion of XIAP detected by exome sequencing analysis in very early onset inflammatory bowel disease: a case report. *BMC Gastroenterology*, **2015**, 15, 160 | 3 | 31 |
| 75 | ADHD genetics: 2007 update. *Current Psychiatry Reports*, **2007**, 9, 434-9 | 9.1 | 31 |
| 74 | Genetic predisposition to familial neuroblastoma: identification of two novel genomic regions at 2p and 12p. *Human Heredity*, **2007**, 63, 205-11 | 1.1 | 31 |
| 73 | CARD15 genotyping in inflammatory bowel disease patients by multiplex pyrosequencing. *Clinical Chemistry*, **2003**, 49, 1675-9 | 5.5 | 29 |
| 72 | Estimates of genetic and environmental contribution to 43 quantitative traits support sharing of a homogeneous environment in an isolated population from South Tyrol, Italy. *Human Heredity*, **2008**, 65, 175-82 | 1.1 | 28 |
| 71 | Association of a Novel ACTA1 Mutation With a Dominant Progressive Scapuloperoneal Myopathy in an Extended Family. *JAMA Neurology*, **2015**, 72, 689-98 | 17.2 | 27 |
| 70 | Variants of the ACTG2 gene correlate with degree of severity and presence of megacystis in chronic intestinal pseudo-obstruction. *European Journal of Human Genetics*, **2016**, 24, 1211-5 | 5.3 | 27 |
| 69 | Candidate gene analysis in an on-going genome-wide association study of attention-deficit hyperactivity disorder: suggestive association signals in ADRA1A. *Psychiatric Genetics*, **2009**, 19, 134-41 | 2.9 | 27 |
| 68 | Linkage analysis conditional on HLA status in a large North American pedigree supports the presence of a multiple sclerosis susceptibility locus on chromosome 12p12. *Human Molecular Genetics*, **2002**, 11, 295-300 | 5.6 | 27 |
| 67 | Exclusion of linkage to the CDL1 gene region on chromosome 3q26.3 in some familial cases of Cornelia de Lange syndrome. *American Journal of Medical Genetics Part A*, **2001**, 101, 120-129 | | 26 |
| 66 | A novel locus for syndromic chronic idiopathic intestinal pseudo-obstruction maps to chromosome 8q23-q24. *European Journal of Human Genetics*, **2007**, 15, 889-97 | 5.3 | 24 |

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| 65 | Contribution of IBD5 locus to clinical features of IBD patients. *American Journal of Gastroenterology* , **2006**, 101, 318-25 | 0.7 | 24 |
| 64 | Heterogeneity in the magnitude of the insulin gene effect on HLA risk in type 1 diabetes. *Diabetes*, **2004**, 53, 3286-91 | 0.9 | 24 |
| 63 | A genome-wide association study identifies a susceptibility locus for biliary atresia on 2p16.1 within the gene EFEMP1. *PLoS Genetics*, **2018**, 14, e1007532 | 6 | 23 |
| 62 | Genome-wide linkage analysis to identify genetic modifiers of ALK mutation penetrance in familial neuroblastoma. *Human Heredity*, **2011**, 71, 135-9 | 1.1 | 23 |
| 61 | Five families with arginine 519-cysteine mutation in COL2A1: evidence for three distinct founders. *Human Mutation*, **1998**, 12, 172-6 | 4.7 | 23 |
| 60 | Ophthalmologic findings in Cornelia de Lange syndrome: a genotype-phenotype correlation study. *JAMA Ophthalmology*, **2006**, 124, 552-7 | | 21 |
| 59 | A nonsense mutation (R1158X) and a splicing mutation (3849 + 4A----G) in exon 19 of the cystic fibrosis transmembrane conductance regulator gene. *Genomics*, **1992**, 12, 417-8 | 4.3 | 21 |
| 58 | Is a Candidate Modifier of Liver Disease Severity in Alagille Syndrome. *Cellular and Molecular Gastroenterology and Hepatology*, **2016**, 2, 663-675.e2 | 7.9 | 21 |
| 57 | Novel COCH mutation in a family with autosomal dominant late onset sensorineural hearing impairment and tinnitus. *American Journal of Otolaryngology - Head and Neck Medicine and Surgery*, **2013**, 34, 230-5 | 2.8 | 20 |
| 56 | Expanding the spectrum of genes responsible for hereditary motor neuropathies. *Journal of Neurology, Neurosurgery and Psychiatry*, **2019**, 90, 1171-1179 | 5.5 | 19 |
| 55 | Pathway analysis supports association of nonsyndromic cryptorchidism with genetic loci linked to cytoskeleton-dependent functions. *Human Reproduction*, **2015**, 30, 2439-51 | 5.7 | 19 |
| 54 | Overt cleft palate phenotype and TBX1 genotype correlations in velo-cardio-facial/DiGeorge/22q11.2 deletion syndrome patients. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2781-7 | 2.5 | 19 |
| 53 | Genetic loci linked to type 1 diabetes and multiple sclerosis families in Sardinia. *BMC Medical Genetics*, **2008**, 9, 3 | 2.1 | 18 |
| 52 | Rare copy number variants in over 100,000 European ancestry subjects reveal multiple disease associations. *Nature Communications*, **2020**, 11, 255 | 17.4 | 17 |
| 51 | Analysis of GWAS top hits in ADHD suggests association to two polymorphisms located in genes expressed in the cerebellum. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2010**, 153B, 1127-33 | 3.5 | 17 |
| 50 | Genetic refinement and physical mapping of the CMT4B gene on chromosome 11q22. *Genomics*, **2000**, 63, 271-8 | 4.3 | 17 |
| 49 | Compound heterozygous mutations in NEK8 in siblings with end-stage renal disease with hepatic and cardiac anomalies. *American Journal of Medical Genetics, Part A*, **2016**, 170, 750-3 | 2.5 | 17 |
| 48 | Phenotype specific association of the TGFBR3 locus with nonsyndromic cryptorchidism. *Journal of Urology*, **2015**, 193, 1637-45 | 2.5 | 16 |

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| 47 | Linkage analysis in families with recurrent neuroblastoma. *Annals of the New York Academy of Sciences*, **2002**, 963, 74-84 | 6.5 | 16 |
| 46 | 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. *European Journal of Human Genetics*, **2000**, 8, 846-52 | 5.3 | 16 |
| 45 | Complex genetic signatures in immune cells underlie autoimmunity and inform therapy. *Nature Genetics*, **2020**, 52, 1036-1045 | 36.3 | 16 |
| 44 | 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. *Inflammatory Bowel Diseases*, **2017**, 23, 2252-2255 | 4.5 | 15 |
| 43 | A refined physical and transcriptional map of the SPG9 locus on 10q23.3-q24.2. *European Journal of Human Genetics*, **2000**, 8, 777-82 | 5.3 | 15 |
| 42 | Exclusion of linkage between RET and neuronal intestinal dysplasia type B. *American Journal of Medical Genetics Part A*, **1996**, 62, 195-8 | | 15 |
| 41 | Haplotype distribution and molecular defects at the phenylalanine hydroxylase locus in Italy. *Human Genetics*, **1990**, 86, 69-72 | 6.3 | 15 |
| 40 | The Unique Disease Course of Children with Very Early onset-Inflammatory Bowel Disease. *Inflammatory Bowel Diseases*, **2020**, 26, 909-918 | 4.5 | 14 |
| 39 | Novel X-linked mental retardation syndrome with short stature maps to Xq24. *American Journal of Medical Genetics Part A*, **2001**, 103, 1-8 | | 12 |
| 38 | Frequency of consanguineous marriages among parents and grandparents of Down patients. *Human Genetics*, **1985**, 70, 256-8 | 6.3 | 12 |
| 37 | A candidate gene approach to identify modifiers of the palatal phenotype in 22q11.2 deletion syndrome patients. *International Journal of Pediatric Otorhinolaryngology*, **2013**, 77, 123-7 | 1.7 | 11 |
| 36 | Statistical tools for linkage analysis and genetic association studies. *Expert Review of Molecular Diagnostics*, **2005**, 5, 781-96 | 3.8 | 11 |
| 35 | Second family with hearing impairment linked to 19q13 and refined DFNA4 localisation. *European Journal of Human Genetics*, **2002**, 10, 95-9 | 5.3 | 11 |
| 34 | Neural crest-derived tumor neuroblastoma and melanoma share 1p13.2 as susceptibility locus that shows a long-range interaction with the SLC16A1 gene. *Carcinogenesis*, **2020**, 41, 284-295 | 4.6 | 11 |
| 33 | Variants of ST8SIA1 are associated with risk of developing multiple sclerosis. *PLoS ONE*, **2008**, 3, e2653 | 3.7 | 10 |
| 32 | Genetic differences in cystic fibrosis patients with and without pancreatic insufficiency. An Italian collaborative study. *Human Genetics*, **1990**, 84, 435-8 | 6.3 | 10 |
| 31 | Exome sequencing reveals compound heterozygous mutations in ATP8B1 in a JAG1/NOTCH2 mutation-negative patient with clinically diagnosed Alagille syndrome. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 891-3 | 2.5 | 9 |
| 30 | Analysis of linkage disequilibrium between different cystic fibrosis mutations and three intragenic microsatellites in the Italian population. *Human Mutation*, **1995**, 5, 23-7 | 4.7 | 9 |

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|---|---|---|---|
| 29 | Frequency of the delta F508 mutation in a sample of 175 Italian cystic fibrosis patients. *Human Genetics*, **1990**, 85, 400-2 | 6.3 | 9 |
| 28 | Exploring Shared Susceptibility between Two Neural Crest Cells Originating Conditions: Neuroblastoma and Congenital Heart Disease. *Genes*, **2019**, 10, | 4.2 | 8 |
| 27 | Exome Sequencing in Individuals with Isolated Biliary Atresia. *Scientific Reports*, **2020**, 10, 2709 | 4.9 | 7 |
| 26 | Subphenotype meta-analysis of testicular cancer genome-wide association study data suggests a role for RBFOX family genes in cryptorchidism susceptibility. *Human Reproduction*, **2018**, 33, 967-977 | 5.7 | 7 |
| 25 | Polygenic inheritance of cryptorchidism susceptibility in the LE/orl rat. *Molecular Human Reproduction*, **2016**, 22, 18-34 | 4.4 | 7 |
| 24 | Association of a polymorphic variant of the adiponectin gene with insulin resistance in african americans. *Clinical and Translational Science*, **2008**, 1, 194-9 | 4.9 | 6 |
| 23 | IQ and hemizygosity for the Val Met functional polymorphism of COMT in 22q11DS. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2016**, 171, 1112-1115 | 3.5 | 5 |
| 22 | Integration of Omics Data in Genetic Epidemiology. *Human Heredity*, **2015**, 79, 109-10 | 1.1 | 4 |
| 21 | Comparative analysis of different approaches for dealing with candidate regions in the context of a genome-wide association study. *BMC Proceedings*, **2009**, 3 Suppl 7, S93 | 2.3 | 4 |
| 20 | Next-generation linkage analysis. *Human Heredity*, **2011**, 72, 227 | 1.1 | 4 |
| 19 | Genomic copy number variation association study in Caucasian patients with nonsyndromic cryptorchidism. *BMC Urology*, **2016**, 16, 62 | 2.2 | 4 |
| 18 | Advances in family-based association analysis. Introduction. *Human Heredity*, **2008**, 66, 65-6 | 1.1 | 3 |
| 17 | Comparison of sib pair-based approaches for identifying quantitative trait loci underlying asthma in the Busselton families. *Genetic Epidemiology*, **2001**, 21 Suppl 1, S198-203 | 2.6 | 3 |
| 16 | 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. *Inflammatory Bowel Diseases*, **2017**, 23, 591-592 | 4.5 | 2 |
| 15 | An Exploratory Association Analysis of the Insulin Gene Region With Diabetes Mellitus in Two Dog Breeds. *Journal of Heredity*, **2019**, 110, 793-800 | 2.4 | 2 |
| 14 | Genetic mapping of quantitative trait loci for disease-related phenotypes. *Methods in Molecular Biology*, **2012**, 871, 281-311 | 1.4 | 2 |
| 13 | Linkage study of early-onset obesity to leptin receptor gene in Italian children. *Nutrition Research*, **2000**, 20, 1059-1063 | 4 | 2 |
| 12 | Prenatal diagnosis and linkage disequilibrium with cystic fibrosis for markers surrounding D7S8. *Human Genetics*, **1990**, 85, 275-8 | 6.3 | 2 |

# List of Publications

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|---|---|---|---|
| 11 | Sequencing of over 100,000 individuals identifies multiple genes and rare variants associated with Crohns disease susceptibility | | 2 |
| 10 | N-myc oncogene amplification and catecholamine metabolism in patients with neuroblastoma. *Lancet, The*, **1987**, 2, 795 | 40 | 1 |
| 9 | Impaired redox and protein homeostasis as risk factors and therapeutic targets in toxin-induced biliary atresia | | 1 |
| 8 | Colonoids From Patients With Pediatric Inflammatory Bowel Disease Exhibit Decreased Growth Associated With Inflammation Severity and Durable Upregulation of Antigen Presentation Genes. *Inflammatory Bowel Diseases*, **2021**, 27, 256-267 | 4.5 | 1 |
| 7 | Impaired Redox and Protein Homeostasis as Risk Factors and Therapeutic Targets in Toxin-Induced Biliary Atresia. *Gastroenterology*, **2020**, 159, 1068-1084.e2 | 13.3 | 0 |
| 6 | Variants in STXBP3 are Associated with Very Early Onset Inflammatory Bowel Disease, Bilateral Sensorineural Hearing Loss and Immune Dysregulation. *Journal of Crohns and Colitis*, **2021**, 15, 1908-1919 | 7.5 | 0 |
| 5 | Immune Dysregulation in Human ITCH Deficiency Successfully Treated with Hematopoietic Cell Transplantation. *Journal of Allergy and Clinical Immunology: in Practice*, **2021**, 9, 2885-2893.e3 | 5.4 | 0 |
| 4 | P-197 Identification of a Homozygous Mutation in the ZBTB24 Gene in a Patient with Very Early Onset Inflammatory Bowel Disease. *Inflammatory Bowel Diseases*, **2016**, 22, S68 | 4.5 | |
| 3 | O-003 Understanding the Relevance of Whole Exome Sequencing Identified Variants in Patients with Very Early-Onset-IBD. *Inflammatory Bowel Diseases*, **2016**, 22, S1-S2 | 4.5 | |
| 2 | Attention-deficit hyperactivity disorder168-182 | | |
| 1 | Preliminary results on the frequency of the delta F508 mutation in cystic fibrosis patients from the USSR. *Human Genetics*, **1990**, 85, 423-4 | 6.3 | |