Paolo Gasparini

List of Publications by Citations

313 papers	23,047 citations	73 h-index	146 g-index
335 ext. papers	27,974 ext. citations	9.7 avg, IF	5.5 L-index

#	Paper	IF	Citations
313	A reference panel of 64,976 haplotypes for genotype imputation. *Nature Genetics*, **2016**, 48, 1279-83	36.3	1447
312	Genome-wide association study identifies 74 loci associated with educational attainment. *Nature*, **2016**, 533, 539-42	50.4	850
311	The putative forkhead transcription factor FOXL2 is mutated in blepharophimosis/ptosis/epicanthus inversus syndrome. *Nature Genetics*, **2001**, 27, 159-66	36.3	790
310	The gene TFR2 is mutated in a new type of haemochromatosis mapping to 7q22. *Nature Genetics*, **2000**, 25, 14-5	36.3	649
309	Connexin-26 mutations in sporadic and inherited sensorineural deafness. *Lancet, The*, **1998**, 351, 394-8	40	542
308	Genome-wide association analyses identify 18 new loci associated with serum urate concentrations. *Nature Genetics*, **2013**, 45, 145-54	36.3	505
307	Mutations of SURF-1 in Leigh disease associated with cytochrome c oxidase deficiency. *American Journal of Human Genetics*, **1998**, 63, 1609-21	11	454
306	Dating the origin of the CCR5-Delta32 AIDS-resistance allele by the coalescence of haplotypes. *American Journal of Human Genetics*, **1998**, 62, 1507-15	11	428
305	Rare and low-frequency coding variants alter human adult height. *Nature*, **2017**, 542, 186-190	50.4	412
304	Parent-of-origin-specific allelic associations among 106 genomic loci for age at menarche. *Nature*, **2014**, 514, 92-97	50.4	401
303	Genetic analysis of over 1 million people identifies 535 new loci associated with blood pressure traits. *Nature Genetics*, **2018**, 50, 1412-1425	36.3	386
302	A general approach for haplotype phasing across the full spectrum of relatedness. *PLoS Genetics*, **2014**, 10, e1004234	6	377
301	Thirty new loci for age at menarche identified by a meta-analysis of genome-wide association studies. *Nature Genetics*, **2010**, 42, 1077-85	36.3	372
300	Autosomal-dominant hemochrom-atosis is associated with a mutation in the ferroportin (SLC11A3) gene. *Journal of Clinical Investigation*, **2001**, 108, 619-623	15.9	364
299	MPV17 encodes an inner mitochondrial membrane protein and is mutated in infantile hepatic mitochondrial DNA depletion. *Nature Genetics*, **2006**, 38, 570-5	36.3	341
298	Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. *Nature Genetics*, **2011**, 43, 1005-11	36.3	338
297	New gene functions in megakaryopoiesis and platelet formation. *Nature*, **2011**, 480, 201-8	50.4	330

296	Cystinuria caused by mutations in rBAT, a gene involved in the transport of cystine. *Nature Genetics*, **1994**, 6, 420-5	36.3	322
295	High carrier frequency of the 35delG deafness mutation in European populations. Genetic Analysis Consortium of GJB2 35delG. *European Journal of Human Genetics*, **2000**, 8, 19-23	5.3	318
294	Genome-wide association analysis identifies novel blood pressure loci and offers biological insights into cardiovascular risk. *Nature Genetics*, **2017**, 49, 403-415	36.3	313
293	Genetic associations at 53 loci highlight cell types and biological pathways relevant for kidney function. *Nature Communications*, **2016**, 7, 10023	17.4	295
292	Mutations in GJB6 cause nonsyndromic autosomal dominant deafness at DFNA3 locus. *Nature Genetics*, **1999**, 23, 16-8	36.3	293
291	Reelin gene alleles and haplotypes as a factor predisposing to autistic disorder. *Molecular Psychiatry*, **2001**, 6, 150-9	15.1	282
290	Genome-wide association analyses of risk tolerance and risky behaviors in over 1 million individuals identify hundreds of loci and shared genetic influences. *Nature Genetics*, **2019**, 51, 245-257	36.3	259
289	Seventy-five genetic loci influencing the human red blood cell. *Nature*, **2012**, 492, 369-75	50.4	257
288	Meta-analyses identify 13 loci associated with age at menopause and highlight DNA repair and immune pathways. *Nature Genetics*, **2012**, 44, 260-8	36.3	243
287	Genetic structure of Europeans: a view from the North-East. *PLoS ONE*, **2009**, 4, e5472	3.7	237
286	Prevalence and evolutionary origins of the del(GJB6-D13S1830) mutation in the DFNB1 locus in hearing-impaired subjects: a multicenter study. *American Journal of Human Genetics*, **2003**, 73, 1452-8	11	236
285	Non-type I cystinuria caused by mutations in SLC7A9, encoding a subunit (bo,+AT) of rBAT. *Nature Genetics*, **1999**, 23, 52-7	36.3	232
284	Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair. *Nature Genetics*, **2015**, 47, 1294-1303	36.3	226
283	Hereditary hemochromatosis in adults without pathogenic mutations in the hemochromatosis gene. *New England Journal of Medicine*, **1999**, 341, 725-32	59.2	224
282	A catalog of genetic loci associated with kidney function from analyses of a million individuals. *Nature Genetics*, **2019**, 51, 957-972	36.3	217
281	SDHAF1, encoding a LYR complex-II specific assembly factor, is mutated in SDH-defective infantile leukoencephalopathy. *Nature Genetics*, **2009**, 41, 654-6	36.3	209
280	Juvenile hemochromatosis locus maps to chromosome 1q. *American Journal of Human Genetics*, **1999**, 64, 1388-93	11	209
279	New mutations inactivating transferrin receptor 2 in hemochromatosis type 3. *Blood*, **2001**, 97, 2555-60	2.2	202

278	Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization. *Nature Genetics*, **2014**, 46, 826-36	36.3	199
277	Genome-wide analysis identifies 12 loci influencing human reproductive behavior. *Nature Genetics*, **2016**, 48, 1462-1472	36.3	198
276	Comparison between SLC3A1 and SLC7A9 cystinuria patients and carriers: a need for a new classification. *Journal of the American Society of Nephrology: JASN*, **2002**, 13, 2547-53	12.7	187
275	Population genetic differentiation of height and body mass index across Europe. *Nature Genetics*, **2015**, 47, 1357-62	36.3	186
274	MYO6, the human homologue of the gene responsible for deafness in Snell's waltzer mice, is mutated in autosomal dominant nonsyndromic hearing loss. *American Journal of Human Genetics*, **2001**, 69, 635-40	11	186
273	Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity. *Nature Genetics*, **2018**, 50, 26-41	36.3	186
272	Molecular genetics of hearing impairment due to mutations in gap junction genes encoding beta connexins. *Human Mutation*, **2000**, 16, 190-202	4.7	180
271	Ethylmalonic encephalopathy is caused by mutations in ETHE1, a gene encoding a mitochondrial matrix protein. *American Journal of Human Genetics*, **2004**, 74, 239-52	11	176
270	Mutations in a novel gene with transmembrane domains underlie Usher syndrome type 3. *American Journal of Human Genetics*, **2001**, 69, 673-84	11	171
269	Severe infantile encephalomyopathy caused by a mutation in COX6B1, a nucleus-encoded subunit of cytochrome c oxidase. *American Journal of Human Genetics*, **2008**, 82, 1281-9	11	151
268	Spectrum of hemojuvelin gene mutations in 1q-linked juvenile hemochromatosis. *Blood*, **2004**, 103, 4317-21	22	150
267	Age- and sex-related variations in platelet count in Italy: a proposal of reference ranges based on 40987 subjects' data. *PLoS ONE*, **2013**, 8, e54289	3.7	146
266	Genome-wide association and functional follow-up reveals new loci for kidney function. *PLoS Genetics*, **2012**, 8, e1002584	6	143
265	Variation in the bitter-taste receptor gene TAS2R38, and adiposity in a genetically isolated population in Southern Italy. *Obesity*, **2008**, 16, 2289-95	8	137
264	Genome sequencing elucidates Sardinian genetic architecture and augments association analyses for lipid and blood inflammatory markers. *Nature Genetics*, **2015**, 47, 1272-1281	36.3	129
263	Target genes, variants, tissues and transcriptional pathways influencing human serum urate levels. *Nature Genetics*, **2019**, 51, 1459-1474	36.3	122
262	Natural history of juvenile haemochromatosis. *British Journal of Haematology*, **2002**, 117, 973-9	4.5	121
261	Directional dominance on stature and cognition in diverse human populations. *Nature*, **2015**, 523, 459-462	50.4	119

260	Nonmuscle myosin heavy-chain gene MYH14 is expressed in cochlea and mutated in patients affected by autosomal dominant hearing impairment (DFNA4). *American Journal of Human Genetics*, **2004**, 74, 770-6	11	117
259	Functional analysis of mutations in SLC7A9, and genotype-phenotype correlation in non-Type I cystinuria. *Human Molecular Genetics*, **2001**, 10, 305-16	5.6	115
258	Stomatocytic haemolysis and macrothrombocytopenia (Mediterranean stomatocytosis/macrothrombocytopenia) is the haematological presentation of phytosterolaemia. *British Journal of Haematology*, **2005**, 130, 297-309	4.5	113
257	A functional study of plasma-membrane calcium-pump isoform 2 mutants causing digenic deafness. *Proceedings of the National Academy of Sciences of the United States of America*, **2007**, 104, 1516-21	11.5	104
256	Meta-analysis of genome-wide association studies identifies six new Loci for serum calcium concentrations. *PLoS Genetics*, **2013**, 9, e1003796	6	100
255	Mitochondrial DNA mutations in patients with postlingual, nonsyndromic hearing impairment. *European Journal of Human Genetics*, **2005**, 13, 26-33	5.3	96
254	Genetic variation in taste sensitivity to 6-n-propylthiouracil and its relationship to taste perception and food selection. *Annals of the New York Academy of Sciences*, **2009**, 1170, 126-39	6.5	95
253	Exploring influences on food choice in a large population sample: The Italian Taste project. *Food Quality and Preference*, **2017**, 59, 123-140	5.8	94
252	FASTKD2 nonsense mutation in an infantile mitochondrial encephalomyopathy associated with cytochrome c oxidase deficiency. *American Journal of Human Genetics*, **2008**, 83, 415-23	11	93
251	Multiple loci are associated with white blood cell phenotypes. *PLoS Genetics*, **2011**, 7, e1002113	6	92
250	Genetic variants linked to education predict longevity. *Proceedings of the National Academy of Sciences of the United States of America*, **2016**, 113, 13366-13371	11.5	90
249	Insights into the binding of Phenyltiocarbamide (PTC) agonist to its target human TAS2R38 bitter receptor. *PLoS ONE*, **2010**, 5, e12394	3.7	87
248	Novel Blood Pressure Locus and Gene Discovery Using Genome-Wide Association Study and Expression Data Sets From Blood and the Kidney. *Hypertension*, **2017**,	8.5	85
247	Multiple mutations of MYO1A, a cochlear-expressed gene, in sensorineural hearing loss. *American Journal of Human Genetics*, **2003**, 72, 1571-7	11	85
246	Hearing loss: frequency and functional studies of the most common connexin26 alleles. *Biochemical and Biophysical Research Communications*, **2002**, 296, 685-91	3.4	79
245	52 Genetic Loci Influencing Myocardial Mass. *Journal of the American College of Cardiology*, **2016**, 68, 1435-1448	15.1	76
244	Whole-Genome Sequencing Coupled to Imputation Discovers Genetic Signals for Anthropometric Traits. *American Journal of Human Genetics*, **2017**, 100, 865-884	11	74
243	Heterogeneity in Circulating Tumor Cells: The Relevance of the Stem-Cell Subset. *Cancers*, **2019**, 11,	6.6	73

242	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
241	Common variants in UMOD associate with urinary uromodulin levels: a meta-analysis. *Journal of the American Society of Nephrology: JASN*, **2014**, 25, 1869-82	12.7	71
240	1000 Genomes-based meta-analysis identifies 10 novel loci for kidney function. *Scientific Reports*, **2017**, 7, 45040	4.9	70
239	Screening hepcidin for mutations in juvenile hemochromatosis: identification of a new mutation (C70R). *Blood*, **2004**, 103, 2407-9	2.2	68
238	Existence of a genetic risk factor on chromosome 5q in Italian coeliac disease families. *Annals of Human Genetics*, **2001**, 65, 35-41	2.2	68
237	The 2q23.1 microdeletion syndrome: clinical and behavioural phenotype. *European Journal of Human Genetics*, **2010**, 18, 163-70	5.3	65
236	Evidence of inbreeding depression on human height. *PLoS Genetics*, **2012**, 8, e1002655	6	62
235	Incidence and expression of the N1303K mutation of the cystic fibrosis (CFTR) gene. *Human Genetics*, **1992**, 89, 653-8	6.3	62
234	Coarse-grained/molecular mechanics of the TAS2R38 bitter taste receptor: experimentally-validated detailed structural prediction of agonist binding. *PLoS ONE*, **2013**, 8, e64675	3.7	61
233	Multi-ancestry genome-wide gene-smoking interaction study of 387,272 individuals identifies new loci associated with serum lipids. *Nature Genetics*, **2019**, 51, 636-648	36.3	59
232	Genome-wide association meta-analysis of individuals of European ancestry identifies new loci explaining a substantial fraction of hair color variation and heritability. *Nature Genetics*, **2018**, 50, 652-656	36.3	59
231	A Large-Scale Multi-ancestry Genome-wide Study Accounting for Smoking Behavior Identifies Multiple Significant Loci for Blood Pressure. *American Journal of Human Genetics*, **2018**, 102, 375-400	11	59
230	Hearing function and thresholds: a genome-wide association study in European isolated populations identifies new loci and pathways. *Journal of Medical Genetics*, **2011**, 48, 369-74	5.8	59
229	Integration of genome-wide association studies with biological knowledge identifies six novel genes related to kidney function. *Human Molecular Genetics*, **2012**, 21, 5329-43	5.6	54
228	Mitochondrial 12S rRNA gene mutations affect RNA secondary structure and lead to variable penetrance in hearing impairment. *Biochemical and Biophysical Research Communications*, **2006**, 341, 950-7	3.4	54
227	Osteoporosis in beta-thalassaemia major patients: analysis of the genetic background. *British Journal of Haematology*, **2000**, 111, 461-6	4.5	54
226	A meta-analysis of genome-wide association studies of the electrocardiographic early repolarization pattern. *Heart Rhythm*, **2012**, 9, 1627-34	6.7	53
225	Discovery and refinement of genetic loci associated with cardiometabolic risk using dense imputation maps. *Nature Genetics*, **2016**, 48, 1303-1312	36.3	51

224	Genetic characterization of northeastern Italian population isolates in the context of broader European genetic diversity. *European Journal of Human Genetics*, **2013**, 21, 659-65	5.3	50
223	Mutations in the TMPRSS3 gene are a rare cause of childhood nonsyndromic deafness in Caucasian patients. *Journal of Molecular Medicine*, **2002**, 80, 124-31	5.5	50
222	Espin gene (ESPN) mutations associated with autosomal dominant hearing loss cause defects in microvillar elongation or organisation. *Journal of Medical Genetics*, **2006**, 43, 157-61	5.8	50
221	Autosomal dominant reticuloendothelial iron overload (HFE type 4) due to a new missense mutation in the FERROPORTIN 1 gene (SLC11A3) in a large French-Canadian family. *Haematologica*, **2003**, 88, 824-6	6.6	46
220	Hereditary hearing loss: a 96 gene targeted sequencing protocol reveals novel alleles in a series of Italian and Qatari patients. *Gene*, **2014**, 542, 209-16	3.8	45
219	Analysis of the complete coding region of the CFTR gene in a cohort of CF patients from north-eastern Italy: identification of 90% of the mutations. *Human Genetics*, **1995**, 95, 397-402	6.3	44
218	Protein-coding variants implicate novel genes related to lipid homeostasis contributing to body-fat distribution. *Nature Genetics*, **2019**, 51, 452-469	36.3	44
217	Global diversity in the TAS2R38 bitter taste receptor: revisiting a classic evolutionary PROPosal. *Scientific Reports*, **2016**, 6, 25506	4.9	43
216	Regulatory evaluation of Glybera in Europe - two committees, one mission. *Nature Reviews Drug Discovery*, **2013**, 12, 719	64.1	43
215	Fixed drug eruptions with feprazone are linked to HLA-B22. *Journal of the American Academy of Dermatology*, **1997**, 36, 782-4	4.5	43
214	Linkage study and exome sequencing identify a BDP1 mutation associated with hereditary hearing loss. *PLoS ONE*, **2013**, 8, e80323	3.7	43
213	Factors Influencing the Phenotypic Characterization of the Oral Marker, PROP. *Nutrients*, **2017**, 9,	6.7	42
212	Estrogen-related receptor gamma and hearing function: evidence of a role in humans and mice. *Neurobiology of Aging*, **2013**, 34, 2077.e1-9	5.6	42
211	Multi-ancestry study of blood lipid levels identifies four loci interacting with physical activity. *Nature Communications*, **2019**, 10, 376	17.4	41
210	Associations of autozygosity with a broad range of human phenotypes. *Nature Communications*, **2019**, 10, 4957	17.4	40
209	Multiancestry Genome-Wide Association Study of Lipid Levels Incorporating Gene-Alcohol Interactions. *American Journal of Epidemiology*, **2019**, 188, 1033-1054	3.8	39
208	PR interval genome-wide association meta-analysis identifies 50 loci associated with atrial and atrioventricular electrical activity. *Nature Communications*, **2018**, 9, 2904	17.4	39
207	A novel mutation in the mitochondrial tRNA(Val) gene associated with a complex neurological presentation. *Annals of Neurology*, **1998**, 43, 98-101	9.4	39

206	First all-in-one diagnostic tool for DNA intelligence: genome-wide inference of biogeographic ancestry, appearance, relatedness, and sex with the Identitas v1 Forensic Chip. *International Journal of Legal Medicine*, **2013**, 127, 559-72	3.1	38
205	Enrichment of low-frequency functional variants revealed by whole-genome sequencing of multiple isolated European populations. *Nature Communications*, **2017**, 8, 15927	17.4	37
204	Genetics of food preferences: a first view from silk road populations. *Journal of Food Science*, **2012**, 77, S413-8	3.4	37
203	Linkage analysis of 6p21 polymorphic markers and the hereditary hemochromatosis: localization of the gene centromeric to HLA-F. *Human Molecular Genetics*, **1993**, 2, 571-6	5.6	37
202	Autosomal recessive Stickler syndrome due to a loss of function mutation in the COL9A3 gene. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 42-7	2.5	36
201	Vezatin, an integral membrane protein of adherens junctions, is required for the sound resilience of cochlear hair cells. *EMBO Molecular Medicine*, **2009**, 1, 125-38	12	36
200	Transforming growth factor-beta1 gene polymorphism, bone turnover, and bone mass in Italian postmenopausal women. *Journal of Bone and Mineral Research*, **2000**, 15, 634-9	6.3	36
199	GOAL: automated Gene Ontology analysis of expression profiles. *Nucleic Acids Research*, **2004**, 32, W492-9	20.1	36
198	DNA mismatch repair gene MSH6 implicated in determining age at natural menopause. *Human Molecular Genetics*, **2014**, 23, 2490-7	5.6	35
197	Polymorphism in intron 4 of HFE does not compromise haemochromatosis mutation results. The European Haemochromatosis Consortium. *Nature Genetics*, **1999**, 23, 271	36.3	34
196	Association analysis of bitter receptor genes in five isolated populations identifies a significant correlation between TAS2R43 variants and coffee liking. *PLoS ONE*, **2014**, 9, e92065	3.7	32
195	Heritability and demographic analyses in the large isolated population of Val Borbera suggest advantages in mapping complex traits genes. *PLoS ONE*, **2009**, 4, e7554	3.7	32
194	Evidence for three genetic loci involved in both anorexia nervosa risk and variation of body mass index. *Molecular Psychiatry*, **2017**, 22, 192-201	15.1	31
193	Characterization of 14 novel deletions underlying Rubinstein-Taybi syndrome: an update of the CREBBP deletion repertoire. *Human Genetics*, **2015**, 134, 613-26	6.3	31
192	Novel genetic associations for blood pressure identified via gene-alcohol interaction in up to 570K individuals across multiple ancestries. *PLoS ONE*, **2018**, 13, e0198166	3.7	31
191	A novel deletion mutation involving TMEM38B in a patient with autosomal recessive osteogenesis imperfecta. *Gene*, **2014**, 545, 290-2	3.8	31
190	Genetic homogeneity of lysinuric protein intolerance. *European Journal of Human Genetics*, **1998**, 6, 612-5	5.3	31
189	GABA (gamma-amino-butyric acid) neurotransmission: identification and fine mapping of the human GABAB receptor gene. *Biochemical and Biophysical Research Communications*, **1998**, 250, 240-5	3.4	31

188	Modulation of genetic associations with serum urate levels by body-mass-index in humans. *PLoS ONE*, **2015**, 10, e0119752	3.7	31
187	and Loci Identified through Large-Scale Exome Chip Analysis Regulate Kidney Development and Function. *Journal of the American Society of Nephrology: JASN*, **2017**, 28, 981-994	12.7	30
186	Expression and replication studies to identify new candidate genes involved in normal hearing function. *PLoS ONE*, **2014**, 9, e85352	3.7	30
185	Genetic evidence for an origin of the Armenians from Bronze Age mixing of multiple populations. *European Journal of Human Genetics*, **2016**, 24, 931-6	5.3	29
184	Cx26 partial loss causes accelerated presbycusis by redox imbalance and dysregulation of Nfr2 pathway. *Redox Biology*, **2018**, 19, 301-317	11.3	28
183	Influence of age, sex and ethnicity on platelet count in five Italian geographic isolates: mild thrombocytopenia may be physiological. *British Journal of Haematology*, **2012**, 157, 384-7	4.5	28
182	Molecular diagnosis of Usher syndrome: application of two different next generation sequencing-based procedures. *PLoS ONE*, **2012**, 7, e43799	3.7	28
181	Understanding the role of personality and alexithymia in food preferences and PROP taste perception. *Physiology and Behavior*, **2016**, 157, 72-8	3.5	27
180	Mutations in L-type amino acid transporter-2 support as a novel gene involved in age-related hearing loss. *ELife*, **2018**, 7,	8.9	27
179	Next generation sequencing in nonsyndromic intellectual disability: from a negative molecular karyotype to a possible causative mutation detection. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 170-6	2.5	27
178	Common variants in Mendelian kidney disease genes and their association with renal function. *Journal of the American Society of Nephrology: JASN*, **2013**, 24, 2105-17	12.7	27
177	Phospholipase C-β is a key modulator of IL-8 expression in cystic fibrosis bronchial epithelial cells. *Journal of Immunology*, **2011**, 186, 4946-58	5.3	27
176	Juvenile hemochromatosis locus maps to chromosome 1q in a French Canadian population. *European Journal of Human Genetics*, **2003**, 11, 585-9	5.3	27
175	An expression atlas of connexin genes in the mouse. *Genomics*, **2004**, 83, 812-20	4.3	27
174	D184E mutation in aquaporin-4 gene impairs water permeability and links to deafness. *Neuroscience*, **2011**, 197, 80-8	3.9	26
173	Does epidermal thickening explain GJB2 high carrier frequency and heterozygote advantage?. *European Journal of Human Genetics*, **2009**, 17, 284-6	5.3	26
172	Genetic heterogeneity of FG syndrome: a fourth locus (FGS4) maps to Xp11.4-p11.3 in an Italian family. *Human Genetics*, **2003**, 112, 124-30	6.3	26
171	Exome-chip meta-analysis identifies novel loci associated with cardiac conduction, including ADAMTS6. *Genome Biology*, **2018**, 19, 87	18.3	25

170	A novel P2RX2 mutation in an Italian family affected by autosomal dominant nonsyndromic hearing loss. *Gene*, **2014**, 534, 236-9	3.8	25
169	A population-based approach to study the impact of PROP perception on food liking in populations along the Silk Road. *PLoS ONE*, **2014**, 9, e91716	3.7	25
168	Rapid sizing of polymorphic microsatellite markers by capillary array electrophoresis. *Journal of Chromatography A*, **1997**, 781, 295-305	4.5	25
167	Molecular epidemiology of Usher syndrome in Italy. *Molecular Vision*, **2011**, 17, 1662-8	2.3	25
166	Genome-wide association analysis on normal hearing function identifies PCDH20 and SLC28A3 as candidates for hearing function and loss. *Human Molecular Genetics*, **2015**, 24, 5655-64	5.6	24
165	Are MYO1C and MYO1F associated with hearing loss?. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2009**, 1792, 27-32	6.9	24
164	A second locus mapping to 2q35-36 for familial pseudohyperkalaemia. *European Journal of Human Genetics*, **2004**, 12, 1073-6	5.3	24
163	DHPLC analysis of the MECP2 gene in Italian Rett patients. *Human Mutation*, **2001**, 18, 132-40	4.7	24
162	Splice-site mutation in the PDS gene may result in intrafamilial variability for deafness in Pendred syndrome. *Human Mutation*, **1999**, 14, 520-6	4.7	24
161	Salt-inducible kinase 3, SIK3, is a new gene associated with hearing. *Human Molecular Genetics*, **2014**, 23, 6407-18	5.6	23
160	Variation of hemoglobin levels in normal Italian populations from genetic isolates. *Haematologica*, **2008**, 93, 1372-5	6.6	23
159	Cystinuria type I: identification of eight new mutations in SLC3A1. *Kidney International*, **2001**, 59, 1250-6	9.9	23
158	Detection of two novel large deletions in SLC3A1 by semi-quantitative fluorescent multiplex PCR. *Human Mutation*, **2000**, 15, 373-9	4.7	23
157	Increased rate of deleterious variants in long runs of homozygosity of an inbred population from Qatar. *Human Heredity*, **2015**, 79, 14-9	1.1	22
156	Cohen syndrome resulting from a novel large intragenic COH1 deletion segregating in an isolated Greek island population. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 2221-6	2.5	22
155	Common variants in CLDN14 are associated with differential excretion of magnesium over calcium in urine. *Pflugers Archiv European Journal of Physiology*, **2017**, 469, 91-103	4.6	21
154	Multicohort analysis of the maternal age effect on recombination. *Nature Communications*, **2015**, 6, 7846	17.4	21
153	Detection of epidermal thickening in GJB2 carriers with epidermal US. *Radiology*, **2009**, 251, 280-6	20.5	21

152	Identification of a novel mutation in the myosin VIIA motor domain in a family with autosomal dominant hearing loss (DFNA11). *Audiology and Neuro-Otology*, **2006**, 11, 157-64	2.2	21
151	Assignment of the gene responsible for cystinuria (rBAT) and of markers D2S119 and D2S177 to 2p16 by fluorescence in situ hybridization. *Human Genetics*, **1995**, 95, 633-6	6.3	21
150	Hereditary hemochromatosis: generation of a transcription map within a refined and extended map of the HLA class I region. *Genomics*, **1996**, 31, 319-26	4.3	21
149	Congenital hyperinsulinism: clinical and molecular analysis of a large Italian cohort. *Gene*, **2013**, 521, 160-58	3.8	20
148	Individual differences in prefrontal cortex activity during perception of bitter taste using fNIRS methodology. *Chemical Senses*, **2010**, 35, 801-12	4.8	20
147	Polymorphisms in sweet taste genes (TAS1R2 and GLUT2), sweet liking, and dental caries prevalence in an adult Italian population. *Genes and Nutrition*, **2015**, 10, 485	4.3	19
146	Genome-Wide Meta-Analysis Unravels Interactions between Magnesium Homeostasis and Metabolic Phenotypes. *Journal of the American Society of Nephrology: JASN*, **2018**, 29, 335-348	12.7	19
145	Non-additive genome-wide association scan reveals a new gene associated with habitual coffee consumption. *Scientific Reports*, **2016**, 6, 31590	4.9	19
144	Does the 1.5 Mb microduplication in chromosome band Xp22.31 have a pathogenetic role? New contribution and a review of the literature. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 461-4	2.5	19
143	Analysis of 31 CFTR mutations by polymerase chain reaction/oligonucleotide ligation assay in a pilot screening of 4476 newborns for cystic fibrosis. *Journal of Medical Screening*, **1999**, 6, 67-9	1.4	19
142	Distribution of Ha-RAS-1 proto-oncogene alleles in breast cancer patients and in a control population. *Breast Cancer Research and Treatment*, **1988**, 11, 147-53	4.4	19
141	Genome-wide association meta-analysis of 30,000 samples identifies seven novel loci for quantitative ECG traits. *European Journal of Human Genetics*, **2019**, 27, 952-962	5.3	18
140	A novel founder MYO15A frameshift duplication is the major cause of genetic hearing loss in Oman. *Journal of Human Genetics*, **2017**, 62, 259-264	4.3	18
139	Age-related hearing loss in four Italian genetic isolates: an epidemiological study. *International Journal of Audiology*, **2009**, 48, 465-72	2.6	17
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