Paolo Gasparini

List of Publications by Year in Descending Order

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	Eating disinhibition and food liking are influenced by variants in CAV1 (caveolin 1) gene. *Food Quality and Preference*, **2022**, 96, 104447	5.8	
312	Combined influence of TAS2R38 genotype and PROP phenotype on the intensity of basic tastes, astringency and pungency in the Italian taste project. *Food Quality and Preference*, **2022**, 95, 104361	5.8	2
311	Polygenic prediction of educational attainment within and between families from genome-wide association analyses in 3 million individuals.. *Nature Genetics*, **2022**,	36.3	7
310	Genetic variations associated with the soapy flavor perception in Gorgonzola PDO cheese. *Food Quality and Preference*, **2022**, 99, 104569	5.8	
309	Large-scale GWAS of food liking reveals genetic determinants and genetic correlations with distinct neurophysiological traits.. *Nature Communications*, **2022**, 13, 2743	17.4	0
308	Differential and shared genetic effects on kidney function between diabetic and non-diabetic individuals. *Communications Biology*, **2022**, 5,	6.7	1
307	Gene-educational attainment interactions in a multi-ancestry genome-wide meta-analysis identify novel blood pressure loci. *Molecular Psychiatry*, **2021**, 26, 2111-2125	15.1	3
306	Genome-wide association study in almost 195,000 individuals identifies 50 previously unidentified genetic loci for eye color. *Science Advances*, **2021**, 7,	14.3	11
305	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. *American Journal of Human Genetics*, **2021**, 108, 502-516	11	12
304	Runs of homozygosity are associated with staging of periodontitis in isolated populations. *Human Molecular Genetics*, **2021**, 30, 1154-1159	5.6	2
303	Natural human knockouts and Mendelian disorders: deep phenotyping in Italian isolates. *European Journal of Human Genetics*, **2021**, 29, 1272-1281	5.3	1
302	Systematic analysis of factors that improve homologous direct repair (HDR) efficiency in CRISPR/Cas9 technique. *PLoS ONE*, **2021**, 16, e0247603	3.7	4
301	Dietary Macronutrient Composition in Relation to Circulating HDL and Non-HDL Cholesterol: A Federated Individual-Level Analysis of Cross-Sectional Data from Adolescents and Adults in 8 European Studies. *Journal of Nutrition*, **2021**, 151, 2317-2329	4.1	1
300	Variants in USP48 encoding ubiquitin hydrolase are associated with autosomal dominant non-syndromic hereditary hearing loss. *Human Molecular Genetics*, **2021**, 30, 1785-1796	5.6	1
299	Relationship between clone metrics and clinical outcome in clonal cytopenia. *Blood*, **2021**, 138, 965-976	2.2	5
298	Endocrine and Growth Abnormalities in 4H Leukodystrophy Caused by Variants in POLR3A, POLR3B, and POLR1C. *Journal of Clinical Endocrinology and Metabolism*, **2021**, 106, e660-e674	5.6	9
297	Differences in taste and smell perception between type 2 diabetes mellitus patients and healthy controls. *Nutrition, Metabolism and Cardiovascular Diseases*, **2021**, 31, 193-200	4.5	8

296	Hearing loss **2021**, 305-322		0
295	Genetics, odor perception and food liking: The intriguing role of cinnamon. *Food Quality and Preference*, **2021**, 93, 104277	5.8	1
294	Taste perception and expression in stomach of bitter taste receptor tas2r38 in obese and lean subjects. *Appetite*, **2021**, 166, 105595	4.5	1
293	Multi-ancestry GWAS of the electrocardiographic PR interval identifies 202 loci underlying cardiac conduction. *Nature Communications*, **2020**, 11, 2542	17.4	16
292	New age-related hearing loss candidate genes in humans: an ongoing challenge. *Gene*, **2020**, 742, 144561	3.8	4
291	Deleterious variants in genes associated with bone mineral density are linked to susceptibility to periodontitis development. *Meta Gene*, **2020**, 24, 100670	0.7	2
290	A population-based approach for gene prioritization in understanding complex traits. *Human Genetics*, **2020**, 139, 647-655	6.3	2
289	A bird's-eye view of Italian genomic variation through whole-genome sequencing. *European Journal of Human Genetics*, **2020**, 28, 435-444	5.3	16
288	Molecular testing for the study of non-syndromic hearing loss. *Hearing, Balance and Communication* , **2020**, 18, 270-277	0.7	3
287	Target genes, variants, tissues and transcriptional pathways influencing human serum urate levels. *Nature Genetics*, **2019**, 51, 1459-1474	36.3	122
286	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
285	Multiancestry Genome-Wide Association Study of Lipid Levels Incorporating Gene-Alcohol Interactions. *American Journal of Epidemiology*, **2019**, 188, 1033-1054	3.8	39
284	Multi-ancestry study of blood lipid levels identifies four loci interacting with physical activity. *Nature Communications*, **2019**, 10, 376	17.4	41
283	A catalog of genetic loci associated with kidney function from analyses of a million individuals. *Nature Genetics*, **2019**, 51, 957-972	36.3	217
282	TAS2R38 bitter taste genotype is associated with complementary feeding behavior in infants. *Genes and Nutrition*, **2019**, 14, 13	4.3	5
281	Next Generation Sequencing and Animal Models Reveal as a New Gene Involved in Human Age-Related Hearing Loss. *Frontiers in Genetics*, **2019**, 10, 142	4.5	6
280	Heterogeneity in Circulating Tumor Cells: The Relevance of the Stem-Cell Subset. *Cancers*, **2019**, 11,	6.6	73
279	A multi-ancestry genome-wide study incorporating gene-smoking interactions identifies multiple new loci for pulse pressure and mean arterial pressure. *Human Molecular Genetics*, **2019**, 28, 2615-2633	5.6	14

278	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
277	Two further patients with Warsaw breakage syndrome. Is a mild phenotype possible?. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e639	2.3	8
276	Mutations in PLS1, encoding fimbrin, cause autosomal dominant nonsyndromic hearing loss. *Human Mutation*, **2019**, 40, 2286-2295	4.7	14
275	Next generation sequencing study in a cohort of Italian patients with syndromic hearing loss. *Hearing Research*, **2019**, 381, 107769	3.9	2
274	A Brief Review of Genetic Approaches to the Study of Food Preferences: Current Knowledge and Future Directions. *Nutrients*, **2019**, 11,	6.7	12
273	Genome-wide association meta-analysis identifies five novel loci for age-related hearing impairment. *Scientific Reports*, **2019**, 9, 15192	4.9	14
272	Associations of autozygosity with a broad range of human phenotypes. *Nature Communications*, **2019**, 10, 4957	17.4	40
271	Protein-coding variants implicate novel genes related to lipid homeostasis contributing to body-fat distribution. *Nature Genetics*, **2019**, 51, 452-469	36.3	44
270	Factors associated with food liking and their relationship with metabolic traits in Italian cohorts. *Food Quality and Preference*, **2019**, 75, 64-70	5.8	7
269	Next-generation sequencing identified SPATC1L as a possible candidate gene for both early-onset and age-related hearing loss. *European Journal of Human Genetics*, **2019**, 27, 70-79	5.3	14
268	Investigation of the link between PROP taste perception and vegetables consumption using FAOSTAT data. *International Journal of Food Sciences and Nutrition*, **2019**, 70, 484-490	3.7	2
267	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
266	TBL1Y: a new gene involved in syndromic hearing loss. *European Journal of Human Genetics*, **2019**, 27, 466-474	5.3	8
265	IBS clinical management in Italy: The AIGO survey. *Digestive and Liver Disease*, **2019**, 51, 782-789	3.3	13
264	Association of LTA gene haploblock with periodontal disease in Italian adults. *Journal of Periodontal Research*, **2019**, 54, 128-133	4.3	3
263	Joint Data Analysis in Nutritional Epidemiology: Identification of Observational Studies and Minimal Requirements. *Journal of Nutrition*, **2018**, 148, 285-297	4.1	7
262	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
261	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

260	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
259	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
258	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
257	Exome-chip meta-analysis identifies novel loci associated with cardiac conduction, including ADAMTS6. *Genome Biology*, **2018**, 19, 87	18.3	25
256	Whole-genome sequencing reveals new insights into age-related hearing loss: cumulative effects, pleiotropy and the role of selection. *European Journal of Human Genetics*, **2018**, 26, 1167-1179	5.3	14
255	Cx26 partial loss causes accelerated presbycusis by redox imbalance and dysregulation of Nfr2 pathway. *Redox Biology*, **2018**, 19, 301-317	11.3	28
254	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
253	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
252	19p13 microduplications encompassing NFIX are responsible for intellectual disability, short stature and small head circumference. *European Journal of Human Genetics*, **2018**, 26, 85-93	5.3	4
251	Genetic Landscape of Slovenians: Past Admixture and Natural Selection Pattern. *Frontiers in Genetics*, **2018**, 9, 551	4.5	3
250	Genomic Studies in a Large Cohort of Hearing Impaired Italian Patients Revealed Several New Alleles, a Rare Case of Uniparental Disomy (UPD) and the Importance to Search for Copy Number Variations. *Frontiers in Genetics*, **2018**, 9, 681	4.5	13
249	A genome-wide association study identifies an association between variants in EFCAB4B gene and periodontal disease in an Italian isolated population. *Journal of Periodontal Research*, **2018**, 53, 992-998	4.3	9
248	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
247	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
246	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
245	Rare and low-frequency coding variants alter human adult height. *Nature*, **2017**, 542, 186-190	50.4	412
244	NLRC5 polymorphism is associated with susceptibility to chronic periodontitis. *Immunobiology*, **2017**, 222, 704-708	3.4	11
243	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

242	LTF and DEFB1 polymorphisms are associated with susceptibility toward chronic periodontitis development. *Oral Diseases*, **2017**, 23, 1001-1008	3.5	16
241	Targeted sequencing identifies novel variants involved in autosomal recessive hereditary hearing loss in Qatari families. *Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis*, **2017**, 800-802, 29-36	3.3	13
240	1000 Genomes-based meta-analysis identifies 10 novel loci for kidney function. *Scientific Reports*, **2017**, 7, 45040	4.9	70
239	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
238	Whole-Genome Sequencing Coupled to Imputation Discovers Genetic Signals for Anthropometric Traits. *American Journal of Human Genetics*, **2017**, 100, 865-884	11	74
237	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
236	Factors Influencing the Phenotypic Characterization of the Oral Marker, PROP. *Nutrients*, **2017**, 9,	6.7	42
235	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
234	Genetic structure in the Sherpa and neighboring Nepalese populations. *BMC Genomics*, **2017**, 18, 102	4.5	12
233	Enrichment of low-frequency functional variants revealed by whole-genome sequencing of multiple isolated European populations. *Nature Communications*, **2017**, 8, 15927	17.4	37
232	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
231	A reference panel of 64,976 haplotypes for genotype imputation. *Nature Genetics*, **2016**, 48, 1279-83	36.3	1447
230	Global diversity in the TAS2R38 bitter taste receptor: revisiting a classic evolutionary PROPosal. *Scientific Reports*, **2016**, 6, 25506	4.9	43
229	Caries and Innate Immunity: DEFB1 Gene Polymorphisms and Caries Susceptibility in Genetic Isolates from North-Eastern Italy. *Caries Research*, **2016**, 50, 589-594	4.2	15
228	52 Genetic Loci Influencing Myocardial Mass. *Journal of the American College of Cardiology*, **2016**, 68, 1435-1448	15.1	76
227	Non-additive genome-wide association scan reveals a new gene associated with habitual coffee consumption. *Scientific Reports*, **2016**, 6, 31590	4.9	19
226	Genome-wide analysis identifies 12 loci influencing human reproductive behavior. *Nature Genetics*, **2016**, 48, 1462-1472	36.3	198
225	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

224	Pharmacogenetics driving personalized medicine: analysis of genetic polymorphisms related to breast cancer medications in Italian isolated populations. *Journal of Translational Medicine*, **2016**, 14, 22	8.5	3
223	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
222	Understanding the role of personality and alexithymia in food preferences and PROP taste perception. *Physiology and Behavior*, **2016**, 157, 72-8	3.5	27
221	Genetic associations at 53 loci highlight cell types and biological pathways relevant for kidney function. *Nature Communications*, **2016**, 7, 10023	17.4	295
220	Genetic analysis of Italian patients with congenital tufting enteropathy. *World Journal of Pediatrics*, **2016**, 12, 219-24	4.6	14
219	A Genome-Wide Association Study in isolated populations reveals new genes associated to common food likings. *Reviews in Endocrine and Metabolic Disorders*, **2016**, 17, 209-19	10.5	17
218	Genome-wide association study identifies 74 loci associated with educational attainment. *Nature*, **2016**, 533, 539-42	50.4	850
217	Discovery and refinement of genetic loci associated with cardiometabolic risk using dense imputation maps. *Nature Genetics*, **2016**, 48, 1303-1312	36.3	51
216	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
215	Directional dominance on stature and cognition in diverse human populations. *Nature*, **2015**, 523, 459-462	50.4	119
214	Analysis of functional variants reveals new candidate genes associated with alexithymia. *Psychiatry Research*, **2015**, 227, 363-5	9.9	7
213	Phenotypic and genetic characterization of a family carrying two Xq21.1-21.3 interstitial deletions associated with syndromic hearing loss. *Molecular Cytogenetics*, **2015**, 8, 18	2	13
212	Genome-wide association analysis on five isolated populations identifies variants of the HLA-DOA gene associated with white wine liking. *European Journal of Human Genetics*, **2015**, 23, 1717-22	5.3	10
211	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
210	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
209	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
208	Multicohort analysis of the maternal age effect on recombination. *Nature Communications*, **2015**, 6, 7846	17.4	21
207	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

206	Population genetic differentiation of height and body mass index across Europe. *Nature Genetics*, **2015**, 47, 1357-62	36.3	186
205	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
204	Connexin 26 variant carriers have a better gastrointestinal health: is this the heterozygote advantage?. *European Journal of Human Genetics*, **2015**, 23, 563-564	5.3	7
203	PSIP1/LEDGF: a new gene likely involved in sensorineural progressive hearing loss. *Scientific Reports*, **2015**, 5, 18568	4.9	4
202	Clinical and Molecular Cytogenetic Characterisation of Children with Developmental Delay and Dysmorphic Features. *Zdravstveno Varstvo*, **2015**, 54, 69-73	1.3	
201	Genetic testing and genomic analysis: a debate on ethical, social and legal issues in the Arab world with a focus on Qatar. *Journal of Translational Medicine*, **2015**, 13, 358	8.5	15
200	The p.Cys169Tyr variant of connexin 26 is not a polymorphism. *Human Molecular Genetics*, **2015**, 24, 2641-8	5.8	9
199	Assessment of the olfactory function in Italian patients with type 3 von Willebrand disease caused by a homozygous 253 Kb deletion involving VWF and TMEM16B/ANO2. *PLoS ONE*, **2015**, 10, e0116483	3.7	7
198	Modulation of genetic associations with serum urate levels by body-mass-index in humans. *PLoS ONE*, **2015**, 10, e0119752	3.7	31
197	Brain-derived neurotrophic factor serum levels in genetically isolated populations: gender-specific association with anxiety disorder subtypes but not with anxiety levels or Val66Met polymorphism. *PeerJ*, **2015**, 3, e1252	3.1	8
196	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
195	Parent-of-origin-specific allelic associations among 106 genomic loci for age at menarche. *Nature*, **2014**, 514, 92-97	50.4	401
194	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
193	Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization. *Nature Genetics*, **2014**, 46, 826-36	36.3	199
192	A novel deletion mutation involving TMEM38B in a patient with autosomal recessive osteogenesis imperfecta. *Gene*, **2014**, 545, 290-2	3.8	31
191	A novel P2RX2 mutation in an Italian family affected by autosomal dominant nonsyndromic hearing loss. *Gene*, **2014**, 534, 236-9	3.8	25
190	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
189	Salt-inducible kinase 3, SIK3, is a new gene associated with hearing. *Human Molecular Genetics*, **2014**, 23, 6407-18	5.6	23

188	A general approach for haplotype phasing across the full spectrum of relatedness. *PLoS Genetics*, **2014**, 10, e1004234	6	377
187	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
186	Age related hearing loss and level of education: An epidemiological study on a large cohort of isolated populations. *Hearing, Balance and Communication*, **2014**, 12, 94-98	0.7	3
185	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
184	DNA mismatch repair gene MSH6 implicated in determining age at natural menopause. *Human Molecular Genetics*, **2014**, 23, 2490-7	5.6	35
183	Consanguinity and hereditary hearing loss in Qatar. *Human Heredity*, **2014**, 77, 175-82	1.1	13
182	Genetic landscape of populations along the Silk Road: admixture and migration patterns. *BMC Genetics*, **2014**, 15, 131	2.6	15
181	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
180	Insight into genetic determinants of resting heart rate. *Gene*, **2014**, 545, 170-4	3.8	7
179	Expression and replication studies to identify new candidate genes involved in normal hearing function. *PLoS ONE*, **2014**, 9, e85352	3.7	30
178	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
177	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
176	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
175	Regulatory evaluation of Glybera in Europe - two committees, one mission. *Nature Reviews Drug Discovery*, **2013**, 12, 719	64.1	43
174	A novel CRYBB2 missense mutation causing congenital autosomal dominant cataract in an Italian family. *Ophthalmic Genetics*, **2013**, 34, 115-7	1.2	9
173	Genome-wide association analyses identify 18 new loci associated with serum urate concentrations. *Nature Genetics*, **2013**, 45, 145-54	36.3	505
172	Molecular cytogenetic characterization of 2p23.2p23.3 deletion in a child with developmental delay, hypotonia and cryptorchism. *European Journal of Medical Genetics*, **2013**, 56, 62-5	2.6	4
171	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

170	Genetics of eye colours in different rural populations on the Silk Road. *European Journal of Human Genetics*, **2013**, 21, 1320-3	5.3	6
169	Congenital hyperinsulinism: clinical and molecular analysis of a large Italian cohort. *Gene*, **2013**, 521, 160-3	3.8	20
168	Co-inheritance of two ABCC8 mutations causing an unresponsive congenital hyperinsulinism: clinical and functional characterization of two novel ABCC8 mutations. *Gene*, **2013**, 516, 122-5	3.8	10
167	Alagille Syndrome: A New Missense Mutation Detected by Whole-Exome Sequencing in a Case Previously Found to Be Negative by DHPLC and MLPA. *Molecular Syndromology*, **2013**, 4, 207-10	1.5	5
166	Meta-analysis of genome-wide association studies identifies six new Loci for serum calcium concentrations. *PLoS Genetics*, **2013**, 9, e1003796	6	100
165	Lifestyle and normal hearing function in Italy and Central Asia: The potential role of coffee. *Hearing, Balance and Communication*, **2013**, 11, 218-223	0.7	5
164	Age- and sex-related variations in platelet count in Italy: a proposal of reference ranges based on 40987 subjectsQdata. *PLoS ONE*, **2013**, 8, e54289	3.7	146
163	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
162	Genome wide association analysis of a founder population identified TAF3 as a gene for MCHC in humans. *PLoS ONE*, **2013**, 8, e69206	3.7	9
161	Linkage study and exome sequencing identify a BDP1 mutation associated with hereditary hearing loss. *PLoS ONE*, **2013**, 8, e80323	3.7	43
160	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
159	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
158	Identification of a New Mutation (L46P) in the Human NOG Gene in an Italian Patient with Symphalangism Syndrome. *Molecular Syndromology*, **2012**, 3, 21-24	1.5	4
157	Two Novel COH1 Mutations in an Italian Patient with Cohen Syndrome. *Molecular Syndromology*, **2012**, 3, 30-33	1.5	3
156	Genetics of food preferences: a first view from silk road populations. *Journal of Food Science*, **2012**, 77, S413-8	3.4	37
155	Seventy-five genetic loci influencing the human red blood cell. *Nature*, **2012**, 492, 369-75	50.4	257
154	De novo 911 Kb interstitial deletion on chromosome 1q43 in a boy with mental retardation and short stature. *European Journal of Medical Genetics*, **2012**, 55, 117-9	2.6	13
153	Contribution of SNP arrays in diagnosis of deletion 2p11.2-p12. *Gene*, **2012**, 492, 315-8	3.8	4

152	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
151	A meta-analysis of genome-wide association studies of the electrocardiographic early repolarization pattern. *Heart Rhythm*, **2012**, 9, 1627-34	6.7	53
150	De novo 6.9 Mb interstitial deletion on chromosome 4q31.1-q32.1 in a girl with severe speech delay and dysmorphic features. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 882-7	2.5	4
149	Evidence of inbreeding depression on human height. *PLoS Genetics*, **2012**, 8, e1002655	6	62
148	Genome-wide association and functional follow-up reveals new loci for kidney function. *PLoS Genetics*, **2012**, 8, e1002584	6	143
147	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
146	Delayed diagnosis of glycogen storage disease type III. *Journal of Pediatric Gastroenterology and Nutrition*, **2012**, 54, 122-4	2.8	1
145	Phylloid pattern of hypomelanosis closely related to chromosomal abnormalities in the 13q detected by SNP array analysis. *Dermatology*, **2012**, 225, 294-7	4.4	14
144	Molecular diagnosis of Usher syndrome: application of two different next generation sequencing-based procedures. *PLoS ONE*, **2012**, 7, e43799	3.7	28
143	New gene functions in megakaryopoiesis and platelet formation. *Nature*, **2011**, 480, 201-8	50.4	330
142	Organo-silane coated substrates for DNA purification. *Applied Surface Science*, **2011**, 257, 10821-10827	6.7	14
141	D184E mutation in aquaporin-4 gene impairs water permeability and links to deafness. *Neuroscience*, **2011**, 197, 80-8	3.9	26
140	Association of a variant in the CHRNA5-A3-B4 gene cluster region to heavy smoking in the Italian population. *European Journal of Human Genetics*, **2011**, 19, 593-6	5.3	13
139	A new case of duplication of the MDS region identified by high-density SNP arrays and a review of the literature. *Journal of Applied Genetics*, **2011**, 52, 77-80	2.5	2
138	Frequency of hearing loss in a series of rural communities of five developing countries located along the Silk Road. *Audiological Medicine*, **2011**, 9, 135-140		12
137	Vertebral defects in patients with Peters plus syndrome and mutations in B3GALTL. *Ophthalmic Genetics*, **2011**, 32, 256-8	1.2	10
136	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
135	Horizontal gaze palsy and progressive scoliosis without ROBO3 mutations. *Ophthalmic Genetics*, **2011**, 32, 212-6	1.2	9

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