

# Thomas Meitinger

## List of Publications by Year in descending order

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546  
papers

97,882  
citations

144

150  
h-index

189

301  
g-index

634  
all docs

634  
docs citations

634  
times ranked

96584  
citing authors

#	ARTICLE	IF	CITATIONS
1	Multi-trait analysis characterizes the genetics of thyroid function and identifies causal associations with clinical implications. <i>Nature Communications</i> , 2024, 15, .	14.1	12
2	Multi-trait analysis characterizes the genetics of thyroid function and identifies causal associations with clinical implications. <i>Nature Communications</i> , 2024, 15, .	14.1	7
3	Understanding the genetic complexity of puberty timing across the allele frequency spectrum. <i>Nature Genetics</i> , 2024, 56, 1397-1411.	16.3	3
4	Next-generation phenotyping integrated in a national framework for patients with ultrarare disorders improves genetic diagnostics and yields new molecular findings. <i>Nature Genetics</i> , 2024, 56, 1644-1653.	16.3	5
5	The constitutional gain-of-function variant p.<sc>Glu1099Lys</sc> in <i>NSD2</i> is associated with a novel syndrome. <i>Clinical Genetics</i> , 2023, 103, 226-230.	2.2	1
6	Genetic insights into resting heart rate and its role in cardiovascular disease. <i>Nature Communications</i> , 2023, 14, .	14.1	13
7	Stimulation of soluble guanylyl cyclase (sGC) by riociguat attenuates heart failure and pathological cardiac remodelling. <i>British Journal of Pharmacology</i> , 2022, 179, 2430-2442.	6.5	22
8	<i>Cis</i>-epistasis at the <i>LPA</i> locus and risk of cardiovascular diseases. <i>Cardiovascular Research</i> , 2022, 118, 1088-1102.	5.6	14
9	Blood DNA methylation provides an accurate biomarker of <i>KMT2B</i>-related dystonia and predicts onset. <i>Brain</i> , 2022, 145, 644-654.	8.9	23
10	Genetic variation influencing DNA methylation provides insights into molecular mechanisms regulating genomic function. <i>Nature Genetics</i> , 2022, 54, 18-29.	16.3	69
11	Population-based screening in children for early diagnosis and treatment of familial hypercholesterolemia: design of the VRONI study. <i>European Journal of Public Health</i> , 2022, 32, 422-428.	0.3	13
12	Association of circulating MR-proADM with all-cause and cardiovascular mortality in the general population: Results from the KORA F4 cohort study. <i>PLoS ONE</i> , 2022, 17, e0262330.	2.5	5
13	MicroRNA-365 regulates human cardiac action potential duration. <i>Nature Communications</i> , 2022, 13, .	14.1	20
14	Genome-wide meta-analysis of phytosterols reveals five novel loci and a detrimental effect on coronary atherosclerosis. <i>Nature Communications</i> , 2022, 13, .	14.1	21
15	Common Genetic Variants Contribute to Risk of Transposition of the Great Arteries. <i>Circulation Research</i> , 2022, 130, 166-180.	12.8	25
16	Rare coding variants in 35 genes associate with circulating lipid levelsâ€”A multi-ancestry analysis of 170,000 exomes. <i>American Journal of Human Genetics</i> , 2022, 109, 81-96.	6.8	23
17	Elucidating the relationship between migraine risk and brain structure using genetic data. <i>Brain</i> , 2022, 145, 3214-3224.	8.9	18
18	Clonal hematopoiesis as a pitfall in germline variant interpretation in the context of Mendelian disorders. <i>Human Molecular Genetics</i> , 2022, 31, 2386-2395.	3.1	3

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19	Genome-wide association analyses identify new Brugada syndrome risk loci and highlight a new mechanism of sodium channel regulation in disease susceptibility. <i>Nature Genetics</i> , 2022, 54, 232-239.	16.3	82
20	Lifetime risk of autosomal recessive neurodegeneration with brain iron accumulation (NBIA) disorders calculated from genetic databases. <i>EBioMedicine</i> , 2022, 77, 103869.	10.0	23
21	Genetic and clinical determinants of abdominal aortic diameter: genome-wide association studies, exome array data and Mendelian randomization study. <i>Human Molecular Genetics</i> , 2022, 31, 3566-3579.	3.1	6
22	Polygenic prediction of educational attainment within and between families from genome-wide association analyses in 3 million individuals. <i>Nature Genetics</i> , 2022, 54, 437-449.	16.3	272
23	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. <i>Genome Medicine</i> , 2022, 14, .	9.9	102
24	Population-based screening in children for early diagnosis and treatment of familial hypercholesterolemia: design of the VRONI study. <i>Medizinische Genetik</i> , 2022, 34, 41-51.	0.4	0
25	Exome sequencing in individuals with cardiovascular laterality defects identifies potential candidate genes. <i>European Journal of Human Genetics</i> , 2022, 30, 946-954.	3.1	5
26	Multi-ancestry genetic study of type 2 diabetes highlights the power of diverse populations for discovery and translation. <i>Nature Genetics</i> , 2022, 54, 560-572.	16.3	310
27	Suleiman-El-Hattab syndrome: a histone modification disorder caused by TASP1 deficiency. <i>Human Molecular Genetics</i> , 2022, 31, 3083-3094.	3.1	4
28	Genetic loci and prioritization of genes for kidney function decline derived from a meta-analysis of 62 longitudinal genome-wide association studies. <i>Kidney International</i> , 2022, 102, 624-639.	5.6	30
29	Genetic analyses of the electrocardiographic QT interval and its components identify additional loci and pathways. <i>Nature Communications</i> , 2022, 13, .	14.1	21
30	Serum uromodulin is inversely associated with biomarkers of subclinical inflammation in the population-based KORA F4 study. <i>CKJ: Clinical Kidney Journal</i> , 2021, 14, 1618-1625.	3.7	8
31	A homozygous truncating variant in <i>CCDC186</i> in an individual with epileptic encephalopathy. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 278-283.	3.8	4
32	Defining the genotypic and phenotypic spectrum of X-linked MSL3-related disorder. <i>Genetics in Medicine</i> , 2021, 23, 384-395.	2.3	10
33	<i>De novo</i> stop-loss variants in <i>CLDN11</i> cause hypomyelinating leukodystrophy. <i>Brain</i> , 2021, 144, 411-419.	8.9	15
34	Multi-ancestry genome-wide association study accounting for gene-psychosocial factor interactions identifies novel loci for blood pressure traits. <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100013.	1.9	4
35	Meta-analysis uncovers genome-wide significant variants for rapid kidney function decline. <i>Kidney International</i> , 2021, 99, 926-939.	5.6	60
36	Defective phosphatidylethanolamine biosynthesis leads to a broad ataxia-spasticity spectrum. <i>Brain</i> , 2021, 144, e30-e30.	8.9	10

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37	Congenital heart disease risk loci identified by genome-wide association study in European patients. <i>Journal of Clinical Investigation</i> , 2021, 131, .	9.1	50
38	DNA methylation and lipid metabolism: an EWAS of 226 metabolic measures. <i>Clinical Epigenetics</i> , 2021, 13, .	4.3	47
39	<i>De novo</i> variants in neurodevelopmental disordersâ€™ experiences from a tertiary care center. <i>Clinical Genetics</i> , 2021, 100, 14-28.	2.2	67
40	Genome-wide association analysis in dilated cardiomyopathy reveals two new players in systolic heart failure on chromosomes 3p25.1 and 22q11.23. <i>European Heart Journal</i> , 2021, 42, 2000-2011.	2.2	51
41	Multi-ancestry genome-wide geneâ€™sleep interactions identify novel loci for blood pressure. <i>Molecular Psychiatry</i> , 2021, 26, 6293-6304.	8.3	12
42	MicroRNA-21â€™Dependent Macrophage-to-Fibroblast Signaling Determines the Cardiac Response to Pressure Overload. <i>Circulation</i> , 2021, 143, 1513-1525.	19.4	88
43	Delineation of epileptic and neurodevelopmental phenotypes associated with variants in STX1B. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2021, 87, 25-29.	2.3	6
44	Rescue of STAT3 Function in Hyper-IgE Syndrome Using Adenine Base Editing. <i>CRISPR Journal</i> , 2021, 4, 178-190.	3.7	11
45	Variants associated with HHIP expression have sex-differential effects on lung function. <i>Wellcome Open Research</i> , 2021, 5, 111.	1.0	2
46	A comprehensive phenotypic characterization of a whole-body Wdr45 knock-out mouse. <i>Mammalian Genome</i> , 2021, 32, 332-349.	2.4	8
47	A multi-ethnic epigenome-wide association study of leukocyte DNA methylation and blood lipids. <i>Nature Communications</i> , 2021, 12, .	14.1	24
48	<i>MATR3</i> haploinsufficiency and early-onset neurodegeneration. <i>Brain</i> , 2021, 144, e72-e72.	8.9	5
49	Determinants of penetrance and variable expressivity in monogenic metabolic conditions across 77,184 exomes. <i>Nature Communications</i> , 2021, 12, .	14.1	48
50	Identification of a Functional <i>PDE5A</i> Variant at the Chromosome 4q27 Coronary Artery Disease Locus in an Extended Myocardial Infarction Family. <i>Circulation</i> , 2021, 144, 662-665.	19.4	7
51	Genetic insights into biological mechanisms governing human ovarian ageing. <i>Nature</i> , 2021, 596, 393-397.	40.1	219
52	Shared genetic pathways contribute to risk of hypertrophic and dilated cardiomyopathies with opposite directions of effect. <i>Nature Genetics</i> , 2021, 53, 128-134.	16.3	172
53	Bi-allelic truncating mutations in <i>VWA1</i> cause neuromyopathy. <i>Brain</i> , 2021, 144, 574-583.	8.9	20
54	Sequential Defects in Cardiac Lineage Commitment and Maturation Cause Hypoplastic Left Heart Syndrome. <i>Circulation</i> , 2021, 144, 1409-1428.	19.4	33

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55	Fatal metabolic decompensation in carbonic anhydrase VA deficiency despite early treatment and control of hyperammonemia. <i>Genetics in Medicine</i> , 2020, 22, 654-655.	2.3	9
56	SQSTM1/p62 variants in 486 patients with familial ALS from Germany and Sweden. <i>Neurobiology of Aging</i> , 2020, 87, 139.e9-139.e15.	3.4	21
57	Epigenetics meets proteomics in an epigenome-wide association study with circulating blood plasma protein traits. <i>Nature Communications</i> , 2020, 11, .	14.1	56
58	Biotinidase deficiency. <i>Neurology: Genetics</i> , 2020, 6, .	2.7	6
59	Low-frequency variation near common germline susceptibility loci are associated with risk of Ewing sarcoma. <i>PLoS ONE</i> , 2020, 15, e0237792.	2.5	7
60	<i>SCN5A</i> Mutation Type and a Genetic Risk Score Associate Variably With Brugada Syndrome Phenotype in <i>SCN5A</i> Families. <i>Circulation Genomic and Precision Medicine</i> , 2020, 13, .	3.2	47
61	Habitual sleep disturbances and migraine: a Mendelian randomization study. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 2370-2380.	3.8	21
62	Gene-educational attainment interactions in a multi-ancestry genome-wide meta-analysis identify novel blood pressure loci. <i>Molecular Psychiatry</i> , 2020, 26, 2111-2125.	8.3	20
63	Transethnic Genome-Wide Association Study Provides Insights in the Genetic Architecture and Heritability of Long QT Syndrome. <i>Circulation</i> , 2020, 142, 324-338.	19.4	101
64	Multi-ancestry GWAS of the electrocardiographic PR interval identifies 202 loci underlying cardiac conduction. <i>Nature Communications</i> , 2020, 11, .	14.1	65
65	A genome-wide cross-phenotype meta-analysis of the association of blood pressure with migraine. <i>Nature Communications</i> , 2020, 11, .	14.1	57
66	Common and rare susceptibility genetic variants predisposing to Brugada syndrome in Thailand. <i>Heart Rhythm</i> , 2020, 17, 2145-2153.	0.8	27
67	DeepWAS: Multivariate genotype-phenotype associations by directly integrating regulatory information using deep learning. <i>PLoS Computational Biology</i> , 2020, 16, e1007616.	3.3	49
68	Biallelic loss-of-function variants in <i>RBL2</i> in siblings with a neurodevelopmental disorder. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 390-396.	3.8	10
69	Diagnostic exome sequencing in non-acquired focal epilepsies highlights a major role of GATOR1 complex genes. <i>Journal of Medical Genetics</i> , 2020, 57, 624-633.	3.7	18
70	A genome-wide analysis of DNA methylation identifies a novel association signal for Lp(a) concentrations in the LPA promoter. <i>PLoS ONE</i> , 2020, 15, e0232073.	2.5	7
71	A novel pathogenic variant in MYO18B associating early-onset muscular hypotonia, and characteristic dysmorphic features, delineation of the phenotypic spectrum of MYO18B-related conditions. <i>Gene</i> , 2020, 742, 144542.	2.4	6
72	Lifetime risk of autosomal recessive mitochondrial disorders calculated from genetic databases. <i>EBioMedicine</i> , 2020, 54, 102730.	10.0	42

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73	Cross-trait analyses with migraine reveal widespread pleiotropy and suggest a vascular component to migraine headache. <i>International Journal of Epidemiology</i> , 2020, 49, 1022-1031.	5.2	37
74	TP53 germline mutations in the context of families with hereditary breast and ovarian cancer: a clinical challenge. <i>Archives of Gynecology and Obstetrics</i> , 2020, 303, 1557-1567.	2.0	4
75	Identification of disease-causing variants by comprehensive genetic testing with exome sequencing in adults with suspicion of hereditary FSGS. <i>European Journal of Human Genetics</i> , 2020, 29, 262-270.	3.1	14
76	Generation of a human induced pluripotent stem cell line (HMGUi002-A) from a healthy male individual. <i>Stem Cell Research</i> , 2019, 39, 101531.	0.6	1
77	Effects of Calcium, Magnesium, and Potassium Concentrations on Ventricular Repolarization in Unselected Individuals. <i>Journal of the American College of Cardiology</i> , 2019, 73, 3118-3131.	2.6	27
78	Disentangling the genetics of lean mass. <i>American Journal of Clinical Nutrition</i> , 2019, 109, 276-287.	5.1	34
79	Adult-onset variant ataxia-telangiectasia diagnosed by exome and cDNA sequencing. <i>Neurology: Genetics</i> , 2019, 5, .	2.7	4
80	Associations of autozygosity with a broad range of human phenotypes. <i>Nature Communications</i> , 2019, 10, .	14.1	76
81	Multi-ancestry sleep-by-SNP interaction analysis in 126,926 individuals reveals lipid loci stratified by sleep duration. <i>Nature Communications</i> , 2019, 10, .	14.1	54
82	Phenotypic variability of <i>GABRA1</i> -related epilepsy in monozygotic twins. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 2317-2322.	3.8	4
83	Reply: Adult-onset distal spinal muscular atrophy: a new phenotype associated with KIF5A mutations. <i>Brain</i> , 2019, 142, e67-e67.	8.9	1
84	Predicting cardiac electrical response to sodium-channel blockade and Brugada syndrome using polygenic risk scores. <i>European Heart Journal</i> , 2019, 40, 3097-3107.	2.2	61
85	Genome-wide association meta-analyses and fine-mapping elucidate pathways influencing albuminuria. <i>Nature Communications</i> , 2019, 10, .	14.1	143
86	Target genes, variants, tissues and transcriptional pathways influencing human serum urate levels. <i>Nature Genetics</i> , 2019, 51, 1459-1474.	16.3	261
87	Genome-wide association meta-analysis of 30,000 samples identifies seven novel loci for quantitative ECG traits. <i>European Journal of Human Genetics</i> , 2019, 27, 952-962.	3.1	25
88	Multiancestry Genome-Wide Association Study of Lipid Levels Incorporating Gene-Alcohol Interactions. <i>American Journal of Epidemiology</i> , 2019, 188, 1033-1054.	3.6	71
89	Multi-ancestry study of blood lipid levels identifies four loci interacting with physical activity. <i>Nature Communications</i> , 2019, 10, .	14.1	62
90	A biallelic mutation links <i>MYORG</i> to autosomal-recessive primary familial brain calcification. <i>Brain</i> , 2019, 142, e4-e4.	8.9	19

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91	Biallelic mutations in <i>PIGP</i> cause developmental and epileptic encephalopathy. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 968-973.	3.8	8
92	A catalog of genetic loci associated with kidney function from analyses of a million individuals. <i>Nature Genetics</i> , 2019, 51, 957-972.	16.3	532
93	Exome sequencing of 20,791 cases of type 2 diabetes and 24,440 controls. <i>Nature</i> , 2019, 570, 71-76.	40.1	212
94	Mendelian randomization evaluation of causal effects of fibrinogen on incident coronary heart disease. <i>PLoS ONE</i> , 2019, 14, e0216222.	2.5	19
95	Mitochondrial DNA mutation analysis from exome sequencing – A more holistic approach in diagnostics of suspected mitochondrial disease. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 909-917.	3.4	62
96	Lessons from exome sequencing in prenatally diagnosed heart defects: A basis for prenatal testing. <i>Clinical Genetics</i> , 2019, 95, 582-589.	2.2	23
97	A multi-ancestry genome-wide study incorporating gene-smoking interactions identifies multiple new loci for pulse pressure and mean arterial pressure. <i>Human Molecular Genetics</i> , 2019, 28, 2615-2633.	3.1	31
98	Multi-ancestry genome-wide gene-smoking interaction study of 387,272 individuals identifies new loci associated with serum lipids. <i>Nature Genetics</i> , 2019, 51, 636-648.	16.3	86
99	Point mutations in the PDX1 transactivation domain impair human $\beta$ -cell development and function. <i>Molecular Metabolism</i> , 2019, 24, 80-97.	6.1	63
100	Whole-exome sequencing revealed a nonsense mutation in <i>STKLD1</i> causing non-syndromic preaxial polydactyly type A affecting only upper limb. <i>Clinical Genetics</i> , 2019, 96, 134-139.	2.2	9
101	Association of alcohol consumption with allergic disease and asthma: a multi-centre Mendelian randomization analysis. <i>Addiction</i> , 2019, 114, 216-225.	6.1	16
102	Association of the PHACTR1/EDN1 Genetic Locus With Spontaneous Coronary Artery Dissection. <i>Journal of the American College of Cardiology</i> , 2019, 73, 58-66.	2.6	159
103	Hot-spot KIF5A mutations cause familial ALS. <i>Brain</i> , 2018, 141, 688-697.	8.9	167
104	Refining the accuracy of validated target identification through coding variant fine-mapping in type 2 diabetes. <i>Nature Genetics</i> , 2018, 50, 559-571.	16.3	305
105	Comprehensive analysis of the mutation spectrum in 301 German ALS families. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 817-827.	2.0	90
106	Genome-wide analysis of PDX1 target genes in human pancreatic progenitors. <i>Molecular Metabolism</i> , 2018, 9, 57-68.	6.1	65
107	A Large-Scale Multi-ancestry Genome-wide Study Accounting for Smoking Behavior Identifies Multiple Significant Loci for Blood Pressure. <i>American Journal of Human Genetics</i> , 2018, 102, 375-400.	6.8	104
108	Compound heterozygous SPATA5 variants in four families and functional studies of SPATA5 deficiency. <i>European Journal of Human Genetics</i> , 2018, 26, 407-419.	3.1	23

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109	SCYL1 variants cause a syndrome with low <sup>γ</sup> -glutamyl-transferase cholestasis, acute liver failure, and neurodegeneration (CALFAN). <i>Genetics in Medicine</i> , 2018, 20, 1255-1265.	2.3	50
110	Determinants of occurrence and survival after sudden cardiac arrest—A European perspective: The ESCAPE-NET project. <i>Resuscitation</i> , 2018, 124, 7-13.	3.0	36
111	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. <i>Neuron</i> , 2018, 97, 1267-1288.	12.8	482
112	Genetic Factors Explain a Major Fraction of the 50% Lower Lipoprotein(a) Concentrations in Finns. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 2018, 38, 1230-1241.	6.2	33
113	Biallelic Mutations in SLC1A2; an Additional Mode of Inheritance for SLC1A2-Related Epilepsy. <i>Neuropediatrics</i> , 2018, 49, 059-062.	1.2	13
114	SERAC1 deficiency causes complicated HSP: evidence from a novel splice mutation in a large family. <i>Journal of Medical Genetics</i> , 2018, 55, 39-47.	3.7	30
115	A comprehensive evaluation of the genetic architecture of sudden cardiac arrest. <i>European Heart Journal</i> , 2018, 39, 3961-3969.	2.2	65
116	Somatic alterations compromised molecular diagnosis of DOCK8 hyper-IgE syndrome caused by a novel intronic splice site mutation. <i>Scientific Reports</i> , 2018, 8, .	3.7	9
117	Fine-mapping type 2 diabetes loci to single-variant resolution using high-density imputation and islet-specific epigenome maps. <i>Nature Genetics</i> , 2018, 50, 1505-1513.	16.3	1,185
118	A De Novo Missense Variant in POU3F2 Identified in a Child with Global Developmental Delay. <i>Neuropediatrics</i> , 2018, 49, 401-404.	1.2	10
119	ExomeChip-Wide Analysis of 95 626 Individuals Identifies 10 Novel Loci Associated With QT and JT Intervals. <i>Circulation Genomic and Precision Medicine</i> , 2018, 11, .	3.2	30
120	Genome Analyses of >200,000 Individuals Identify 58 Loci for Chronic Inflammation and Highlight Pathways that Link Inflammation and Complex Disorders. <i>American Journal of Human Genetics</i> , 2018, 103, 691-706.	6.8	296
121	Genome-wide association meta-analysis highlights light-induced signaling as a driver for refractive error. <i>Nature Genetics</i> , 2018, 50, 834-848.	16.3	240
122	Common and Rare Coding Genetic Variation Underlying the Electrocardiographic PR Interval. <i>Circulation Genomic and Precision Medicine</i> , 2018, 11, .	3.2	18
123	Interplay of cell-cell contacts and RhoA/ MRF signaling regulates cardiomyocyte identity. <i>EMBO Journal</i> , 2018, 37, .	7.4	63
124	PRUNE1 Deficiency: Expanding the Clinical and Genetic Spectrum. <i>Neuropediatrics</i> , 2018, 49, 330-338.	1.2	10
125	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, .	3.3	62
126	PR interval genome-wide association meta-analysis identifies 50 loci associated with atrial and atrioventricular electrical activity. <i>Nature Communications</i> , 2018, 9, .	14.1	67

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127	MAP2 â€“ A Candidate Gene for Epilepsy, Developmental Delay and Behavioral Abnormalities in a Patient With Microdeletion 2q34. <i>Frontiers in Genetics</i> , 2018, 9, .	2.4	12
128	Genome-Wide Association Study on Immunoglobulin G Glycosylation Patterns. <i>Frontiers in Immunology</i> , 2018, 9, .	5.0	59
129	Mutations in PPCS, Encoding Phosphopantothenoylcysteine Synthetase, Cause Autosomal-Recessive Dilated Cardiomyopathy. <i>American Journal of Human Genetics</i> , 2018, 102, 1018-1030.	6.8	46
130	Genome-wide association study identifies multiple new loci associated with Ewing sarcoma susceptibility. <i>Nature Communications</i> , 2018, 9, .	14.1	48
131	Novel genetic associations for blood pressure identified via gene-alcohol interaction in up to 570K individuals across multiple ancestries. <i>PLoS ONE</i> , 2018, 13, e0198166.	2.5	77
132	A Homozygous Splice Site Mutation in SLC25A42, Encoding the Mitochondrial Transporter of Coenzyme A, Causes Metabolic Crises and Epileptic Encephalopathy. <i>JIMD Reports</i> , 2018, , 1-7.	1.3	15
133	Mutations in ATP6V1E1 or ATP6V1A Cause Autosomal-Recessive Cutis Laxa. <i>American Journal of Human Genetics</i> , 2017, 100, 216-227.	6.8	84
134	A homozygous splice variant in <i>AP4S1</i> mimicking neurodegeneration with brain iron accumulation. <i>Movement Disorders</i> , 2017, 32, 797-799.	5.3	14
135	Immuneâ€“Array Analysis in Sporadic Inclusion Body Myositis Reveals HLAâ€“DRB1 Amino Acid Heterogeneity Across the Myositis Spectrum. <i>Arthritis and Rheumatology</i> , 2017, 69, 1090-1099.	6.2	49
136	Genome-wide association study in takotsubo syndrome â€” Preliminary results and future directions. <i>International Journal of Cardiology</i> , 2017, 236, 335-339.	2.2	34
137	Biallelic Mutations in DNAJC12 Cause Hyperphenylalaninemia, Dystonia, and Intellectual Disability. <i>American Journal of Human Genetics</i> , 2017, 100, 257-266.	6.8	125
138	Systematic Evaluation of Pleiotropy Identifies 6 Further Loci Associated Withâ€“Coronary Arteryâ€“Disease. <i>Journal of the American College of Cardiology</i> , 2017, 69, 823-836.	2.6	199
139	Genomic analyses identify hundreds of variants associated with age at menarche and support a role for puberty timing in cancer risk. <i>Nature Genetics</i> , 2017, 49, 834-841.	16.3	396
140	Response to Comment on Adam et al. Metformin Effect on Nontargeted Metabolite Profiles in Patients With Type 2 Diabetes and in Multiple Murine Tissues. <i>Diabetes</i> 2016;65:3776â€“3785. <i>Diabetes</i> , 2017, 66, e3-e4.	0.5	1
141	Exome sequencing revealed a splice site variant in the IQCE gene underlying post-axial polydactyly type A restricted to lower limb. <i>European Journal of Human Genetics</i> , 2017, 25, 960-965.	3.1	41
142	1000 Genomes-based meta-analysis identifies 10 novel loci for kidney function. <i>Scientific Reports</i> , 2017, 7, .	3.7	89
143	A genome-wide association meta-analysis on lipoprotein (a) concentrations adjusted for apolipoprotein (a) isoforms. <i>Journal of Lipid Research</i> , 2017, 58, 1834-1844.	3.7	119
144	Coexisting variants in OSTM1 and MANEAL cause a complex neurodegenerative disorder with NBIA-like brain abnormalities. <i>European Journal of Human Genetics</i> , 2017, 25, 1092-1095.	3.1	11

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145	Elevated glutaric acid levels in Dhtkd1-/Gcdh- double knockout mice challenge our current understanding of lysine metabolism. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2017, 1863, 2220-2228.	4.1	36
146	An Expanded Genome-Wide Association Study of Type 2 Diabetes in Europeans. <i>Diabetes</i> , 2017, 66, 2888-2902.	0.5	506
147	Genetic diagnosis of Mendelian disorders via RNA sequencing. <i>Nature Communications</i> , 2017, 8, .	14.1	404
148	NFAT5 and SLC4A10 Loci Associate with Plasma Osmolality. <i>Journal of the American Society of Nephrology: JASN</i> , 2017, 28, 2311-2321.	0.4	21
149	A Low-Frequency Inactivating <i>AKT2</i> Variant Enriched in the Finnish Population Is Associated With Fasting Insulin Levels and Type 2 Diabetes Risk. <i>Diabetes</i> , 2017, 66, 2019-2032.	0.5	41
150	Identification of a de novo microdeletion 1q44 in a patient with hypogenesis of the corpus callosum, seizures and microcephaly – A case report. <i>Gene</i> , 2017, 616, 41-44.	2.4	7
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