Thomas Meitinger

List of Publications by Year in Descending Order

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

585 papers	83,802 citations	139 h-index	280 g-index
629 ext. papers	98,147 ext. citations	12.6 avg, IF	6.43 L-index

#	Paper	IF	Citations
585	Association of circulating MR-proADM with all-cause and cardiovascular mortality in the general population: Results from the KORA F4 cohort study.. *PLoS ONE*, **2022**, 17, e0262330	3.7	1
584	MicroRNA-365 regulates human cardiac action potential duration.. *Nature Communications*, **2022**, 13, 220	17.4	3
583	Genome-wide meta-analysis of phytosterols reveals five novel loci and a detrimental effect on coronary atherosclerosis.. *Nature Communications*, **2022**, 13, 143	17.4	3
582	Genetic variation influencing DNA methylation provides insights into molecular mechanisms regulating genomic function.. *Nature Genetics*, **2022**,	36.3	6
581	Lifetime risk of autosomal recessive neurodegeneration with brain iron accumulation (NBIA) disorders calculated from genetic databases.. *EBioMedicine*, **2022**, 77, 103869	8.8	1
580	Polygenic prediction of educational attainment within and between families from genome-wide association analyses in 3 million individuals.. *Nature Genetics*, **2022**,	36.3	7
579	Clinical implementation of RNA sequencing for Mendelian disease diagnostics.. *Genome Medicine*, **2022**, 14, 38	14.4	4
578	Population-based screening in children for early diagnosis and treatment of familial hypercholesterolemia: design of the VRONI study. *Medizinische Genetik*, **2022**, 34, 41-51	0.5	
577	Multi-ancestry genetic study of type 2 diabetes highlights the power of diverse populations for discovery and translation.. *Nature Genetics*, **2022**,	36.3	7
576	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
575	Common Genetic Variants Contribute to Risk of Transposition of the Great Arteries. *Circulation Research*, **2021**,	15.7	2
574	Sequential Defects in Cardiac Lineage Commitment and Maturation Cause Hypoplastic Left Heart Syndrome. *Circulation*, **2021**, 144, 1409-1428	16.7	6
573	Bi-allelic variants in SPATA5L1 lead to intellectual disability, spastic-dystonic cerebral palsy, epilepsy, and hearing loss. *American Journal of Human Genetics*, **2021**, 108, 2006-2016	11	3
572	Genome-wide association analysis in dilated cardiomyopathy reveals two new players in systolic heart failure on chromosomes 3p25.1 and 22q11.23. *European Heart Journal*, **2021**, 42, 2000-2011	9.5	14
571	Multi-ancestry genome-wide gene-sleep interactions identify novel loci for blood pressure. *Molecular Psychiatry*, **2021**,	15.1	3
570	MicroRNA-21-Dependent Macrophage-to-Fibroblast Signaling Determines the Cardiac Response to Pressure Overload. *Circulation*, **2021**, 143, 1513-1525	16.7	14
569	Delineation of epileptic and neurodevelopmental phenotypes associated with variants in STX1B. *Seizure: the Journal of the British Epilepsy Association*, **2021**, 87, 25-29	3.2	2

568	Rescue of STAT3 Function in Hyper-IgE Syndrome Using Adenine Base Editing. *CRISPR Journal*, **2021**, 4, 178-190	2.5	2
567	Cis-epistasis at the LPA locus and risk of cardiovascular diseases. *Cardiovascular Research*, **2021**,	9.9	6
566	A comprehensive phenotypic characterization of a whole-body Wdr45 knock-out mouse. *Mammalian Genome*, **2021**, 32, 332-349	3.2	1
565	A multi-ethnic epigenome-wide association study of leukocyte DNA methylation and blood lipids. *Nature Communications*, **2021**, 12, 3987	17.4	3
564	MATR3 haploinsufficiency and early-onset neurodegeneration. *Brain*, **2021**, 144, e72	11.2	
563	Determinants of penetrance and variable expressivity in monogenic metabolic conditions across 77,184 exomes. *Nature Communications*, **2021**, 12, 3505	17.4	5
562	Serum uromodulin is inversely associated with biomarkers of subclinical inflammation in the population-based KORA F4 study. *CKJ: Clinical Kidney Journal*, **2021**, 14, 1618-1625	4.5	2
561	A homozygous truncating variant in CCDC186 in an individual with epileptic encephalopathy. *Annals of Clinical and Translational Neurology*, **2021**, 8, 278-283	5.3	1
560	TP53 germline mutations in the context of families with hereditary breast and ovarian cancer: a clinical challenge. *Archives of Gynecology and Obstetrics*, **2021**, 303, 1557-1567	2.5	2
559	Defining the genotypic and phenotypic spectrum of X-linked MSL3-related disorder. *Genetics in Medicine*, **2021**, 23, 384-395	8.1	0
558	De novo stop-loss variants in CLDN11 cause hypomyelinating leukodystrophy. *Brain*, **2021**, 144, 411-419	11.2	0
557	Meta-analysis uncovers genome-wide significant variants for rapid kidney function decline. *Kidney International*, **2021**, 99, 926-939	9.9	6
556	Identification of disease-causing variants by comprehensive genetic testing with exome sequencing in adults with suspicion of hereditary FSGS. *European Journal of Human Genetics*, **2021**, 29, 262-270	5.3	2
555	Defective phosphatidylethanolamine biosynthesis leads to a broad ataxia-spasticity spectrum. *Brain*, **2021**, 144, e30	11.2	2
554	Congenital heart disease risk loci identified by genome-wide association study in European patients. *Journal of Clinical Investigation*, **2021**, 131,	15.9	9
553	DNA methylation and lipid metabolism: an EWAS of 226 metabolic measures. *Clinical Epigenetics*, **2021**, 13, 7	7.7	11
552	De novo variants in neurodevelopmental disorders-experiences from a tertiary care center. *Clinical Genetics*, **2021**, 100, 14-28	4	11
551	Identification of a Functional Variant at the Chromosome 4q27 Coronary Artery Disease Locus in an Extended Myocardial Infarction Family. *Circulation*, **2021**, 144, 662-665	16.7	1

550	Genetic insights into biological mechanisms governing human ovarian ageing. *Nature*, **2021**, 596, 393-397	50.4	28
549	Shared genetic pathways contribute to risk of hypertrophic and dilated cardiomyopathies with opposite directions of effect. *Nature Genetics*, **2021**, 53, 128-134	36.3	35
548	Bi-allelic truncating mutations in VWA1 cause neuromyopathy. *Brain*, **2021**, 144, 574-583	11.2	5
547	Mutation Type and a Genetic Risk Score Associate Variably With Brugada Syndrome Phenotype in Families. *Circulation Genomic and Precision Medicine*, **2020**, 13, e002911	5.2	13
546	Habitual sleep disturbances and migraine: a Mendelian randomization study. *Annals of Clinical and Translational Neurology*, **2020**, 7, 2370-2380	5.3	6
545	Transethnic Genome-Wide Association Study Provides Insights in the Genetic Architecture and Heritability of Long QT Syndrome. *Circulation*, **2020**, 142, 324-338	16.7	27
544	Multi-ancestry GWAS of the electrocardiographic PR interval identifies 202 loci underlying cardiac conduction. *Nature Communications*, **2020**, 11, 2542	17.4	16
543	A genome-wide cross-phenotype meta-analysis of the association of blood pressure with migraine. *Nature Communications*, **2020**, 11, 3368	17.4	22
542	Common and rare susceptibility genetic variants predisposing to Brugada syndrome in Thailand. *Heart Rhythm*, **2020**, 17, 2145-2153	6.7	8
541	DeepWAS: Multivariate genotype-phenotype associations by directly integrating regulatory information using deep learning. *PLoS Computational Biology*, **2020**, 16, e1007616	5	14
540	Biallelic loss-of-function variants in RBL2 in siblings with a neurodevelopmental disorder. *Annals of Clinical and Translational Neurology*, **2020**, 7, 390-396	5.3	8
539	Diagnostic exome sequencing in non-acquired focal epilepsies highlights a major role of GATOR1 complex genes. *Journal of Medical Genetics*, **2020**, 57, 624-633	5.8	9
538	A genome-wide analysis of DNA methylation identifies a novel association signal for Lp(a) concentrations in the LPA promoter. *PLoS ONE*, **2020**, 15, e0232073	3.7	7
537	A novel pathogenic variant in MYO18B associating early-onset muscular hypotonia, and characteristic dysmorphic features, delineation of the phenotypic spectrum of MYO18B-related conditions. *Gene*, **2020**, 742, 144542	3.8	3
536	Bi-allelic Variants in RALGAPA1 Cause Profound Neurodevelopmental Disability, Muscular Hypotonia, Infantile Spasms, and Feeding Abnormalities. *American Journal of Human Genetics*, **2020**, 106, 246-255	11	6
535	SQSTM1/p62 variants in 486 patients with familial ALS from Germany and Sweden. *Neurobiology of Aging*, **2020**, 87, 139.e9-139.e15	5.6	16
534	Epigenetics meets proteomics in an epigenome-wide association study with circulating blood plasma protein traits. *Nature Communications*, **2020**, 11, 15	17.4	34
533	Stimulation of soluble guanylyl cyclase (sGC) by riociguat attenuates heart failure and pathological cardiac remodelling. *British Journal of Pharmacology*, **2020**,	8.6	5

532	Bi-allelic HPDL Variants Cause a Neurodegenerative Disease Ranging from Neonatal Encephalopathy to Adolescent-Onset Spastic Paraplegia. *American Journal of Human Genetics*, **2020**, 107, 364-373	11	19
531	Biotinidase deficiency: A treatable cause of hereditary spastic paraparesis. *Neurology: Genetics*, **2020**, 6, e525	3.8	1
530	Low-frequency variation near common germline susceptibility loci are associated with risk of Ewing sarcoma. *PLoS ONE*, **2020**, 15, e0237792	3.7	2
529	Variants associated with expression have sex-differential effects on lung function. *Wellcome Open Research*, **2020**, 5, 111	4.8	0
528	Fatal metabolic decompensation in carbonic anhydrase VA deficiency despite early treatment and control of hyperammonemia. *Genetics in Medicine*, **2020**, 22, 654-655	8.1	5
527	Lifetime risk of autosomal recessive mitochondrial disorders calculated from genetic databases. *EBioMedicine*, **2020**, 54, 102730	8.8	22
526	Cross-trait analyses with migraine reveal widespread pleiotropy and suggest a vascular component to migraine headache. *International Journal of Epidemiology*, **2020**, 49, 1022-1031	7.8	15
525	Low-frequency variation near common germline susceptibility loci are associated with risk of Ewing sarcoma **2020**, 15, e0237792		
524	Low-frequency variation near common germline susceptibility loci are associated with risk of Ewing sarcoma **2020**, 15, e0237792		
523	Low-frequency variation near common germline susceptibility loci are associated with risk of Ewing sarcoma **2020**, 15, e0237792		
522	Low-frequency variation near common germline susceptibility loci are associated with risk of Ewing sarcoma **2020**, 15, e0237792		
521	Predicting cardiac electrical response to sodium-channel blockade and Brugada syndrome using polygenic risk scores. *European Heart Journal*, **2019**, 40, 3097-3107	9.5	33
520	Genome-wide association meta-analyses and fine-mapping elucidate pathways influencing albuminuria. *Nature Communications*, **2019**, 10, 4130	17.4	43
519	Target genes, variants, tissues and transcriptional pathways influencing human serum urate levels. *Nature Genetics*, **2019**, 51, 1459-1474	36.3	122
518	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
517	Multiancestry Genome-Wide Association Study of Lipid Levels Incorporating Gene-Alcohol Interactions. *American Journal of Epidemiology*, **2019**, 188, 1033-1054	3.8	39
516	Multi-ancestry study of blood lipid levels identifies four loci interacting with physical activity. *Nature Communications*, **2019**, 10, 376	17.4	41
515	A biallelic mutation links MYORG to autosomal-recessive primary familial brain calcification. *Brain*, **2019**, 142, e4	11.2	12

514	Biallelic mutations in cause developmental and epileptic encephalopathy. *Annals of Clinical and Translational Neurology*, **2019**, 6, 968-973	5.3	5
513	A catalog of genetic loci associated with kidney function from analyses of a million individuals. *Nature Genetics*, **2019**, 51, 957-972	36.3	217
512	Exome sequencing of 20,791´cases of type 2 diabetes and 24,440´controls. *Nature*, **2019**, 570, 71-76	50.4	129
511	Mendelian randomization evaluation of causal effects of fibrinogen on incident coronary heart disease. *PLoS ONE*, **2019**, 14, e0216222	3.7	11
510	Mitochondrial DNA mutation analysis from exome sequencing-A more holistic approach in diagnostics of suspected mitochondrial disease. *Journal of Inherited Metabolic Disease*, **2019**, 42, 909-917	5.4	38
509	Lessons from exome sequencing in prenatally diagnosed heart defects: A basis for prenatal testing. *Clinical Genetics*, **2019**, 95, 582-589	4	15
508	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
507	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
506	Point mutations in the PDX1 transactivation domain impair human Ecell development and function. *Molecular Metabolism*, **2019**, 24, 80-97	8.8	27
505	Whole-exome sequencing revealed a nonsense mutation in STKLD1 causing non-syndromic pre-axial polydactyly type A affecting only upper limb. *Clinical Genetics*, **2019**, 96, 134-139	4	4
504	Generation of a human induced pluripotent stem cell line (HMGUi002-A) from a healthy male individual. *Stem Cell Research*, **2019**, 39, 101531	1.6	1
503	Effects of Calcium, Magnesium, and Potassium Concentrations on Ventricular Repolarization in Unselected Individuals. *Journal of the American College of Cardiology*, **2019**, 73, 3118-3131	15.1	12
502	Disentangling the genetics of lean mass. *American Journal of Clinical Nutrition*, **2019**, 109, 276-287	7	24
501	Adult-onset variant ataxia-telangiectasia diagnosed by exome and cDNA sequencing. *Neurology: Genetics*, **2019**, 5, e346	3.8	4
500	Associations of autozygosity with a broad range of human phenotypes. *Nature Communications*, **2019**, 10, 4957	17.4	40
499	Multi-ancestry sleep-by-SNP interaction analysis in 126,926 individuals reveals lipid loci stratified by sleep duration. *Nature Communications*, **2019**, 10, 5121	17.4	31
498	Phenotypic variability of GABRA1-related epilepsy in monozygotic twins. *Annals of Clinical and Translational Neurology*, **2019**, 6, 2317-2322	5.3	3
497	Reply: Adult-onset distal spinal muscular atrophy: a new phenotype associated with KIF5A mutations. *Brain*, **2019**, 142, e67	11.2	0

496	Exome Sequencing in Children. *Deutsches Ärzteblatt International*, **2019**, 116, 197-204	2.5	20
495	Association of alcohol consumption with allergic disease and asthma: a multi-centre Mendelian randomization analysis. *Addiction*, **2019**, 114, 216-225	4.6	7
494	Association of the PHACTR1/EDN1 Genetic Locus With Spontaneous Coronary Artery Dissection. *Journal of the American College of Cardiology*, **2019**, 73, 58-66	15.1	86
493	A Homozygous Splice Site Mutation in SLC25A42, Encoding the Mitochondrial Transporter of Coenzyme A, Causes Metabolic Crises and Epileptic Encephalopathy. *JIMD Reports*, **2019**, 44, 1-7	1.9	12
492	Hot-spot KIF5A mutations cause familial ALS. *Brain*, **2018**, 141, 688-697	11.2	105
491	Refining the accuracy of validated target identification through coding variant fine-mapping in type 2 diabetes. *Nature Genetics*, **2018**, 50, 559-571	36.3	221
490	Comprehensive analysis of the mutation spectrum in 301 German ALS families. *Journal of Neurology, Neurosurgery and Psychiatry*, **2018**, 89, 817-827	5.5	43
489	Genome-wide analysis of PDX1 target genes in human pancreatic progenitors. *Molecular Metabolism*, **2018**, 9, 57-68	8.8	40
488	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
487	Compound heterozygous SPATA5 variants in four families and functional studies of SPATA5 deficiency. *European Journal of Human Genetics*, **2018**, 26, 407-419	5.3	15
486	SCYL1 variants cause a syndrome with low □-glutamyl-transferase cholestasis, acute liver failure, and neurodegeneration (CALFAN). *Genetics in Medicine*, **2018**, 20, 1255-1265	8.1	29
485	Determinants of occurrence and survival after sudden cardiac arrest-A European perspective: The ESCAPE-NET project. *Resuscitation*, **2018**, 124, 7-13	4	22
484	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. *Neuron*, **2018**, 97, 1268-1283.e6	13.9	296
483	Genetic Factors Explain a Major Fraction of the 50% Lower Lipoprotein(a) Concentrations in Finns. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2018**, 38, 1230-1241	9.4	17
482	Biallelic Mutations in SLC1A2; an Additional Mode of Inheritance for SLC1A2-Related Epilepsy. *Neuropediatrics*, **2018**, 49, 59-62	1.6	9
481	SERAC1 deficiency causes complicated HSP: evidence from a novel splice mutation in a large family. *Journal of Medical Genetics*, **2018**, 55, 39-47	5.8	21
480	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. *Orphanet Journal of Rare Diseases*, **2018**, 13, 120	4.2	41
479	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

478	- A Candidate Gene for Epilepsy, Developmental Delay and Behavioral Abnormalities in a Patient With Microdeletion 2q34. *Frontiers in Genetics*, **2018**, 9, 99	4.5	3
477	Genome-Wide Association Study on Immunoglobulin G Glycosylation Patterns. *Frontiers in Immunology*, **2018**, 9, 277	8.4	36
476	Mutations in PPCS, Encoding Phosphopantothenoylcysteine Synthetase, Cause Autosomal-Recessive Dilated Cardiomyopathy. *American Journal of Human Genetics*, **2018**, 102, 1018-1030	11	29
475	Genome-wide association study identifies multiple new loci associated with Ewing sarcoma susceptibility. *Nature Communications*, **2018**, 9, 3184	17.4	25
474	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
473	A comprehensive evaluation of the genetic architecture of sudden cardiac arrest. *European Heart Journal*, **2018**, 39, 3961-3969	9.5	31
472	Somatic alterations compromised molecular diagnosis of DOCK8 hyper-IgE syndrome caused by a novel intronic splice site mutation. *Scientific Reports*, **2018**, 8, 16719	4.9	4
471	Fine-mapping type 2 diabetes loci to single-variant resolution using high-density imputation and islet-specific epigenome maps. *Nature Genetics*, **2018**, 50, 1505-1513	36.3	675
470	A De Novo Missense Variant in POU3F2 Identified in a Child with Global Developmental Delay. *Neuropediatrics*, **2018**, 49, 401-404	1.6	9
469	ExomeChip-Wide Analysis of 95 626 Individuals Identifies 10 Novel Loci Associated With QT and JT Intervals. *Circulation Genomic and Precision Medicine*, **2018**, 11, e001758	5.2	14
468	Bi-allelic ADPRHL2 Mutations Cause Neurodegeneration with Developmental Delay, Ataxia, and Axonal Neuropathy. *American Journal of Human Genetics*, **2018**, 103, 817-825	11	24
467	Genome Analyses of >200,000 Individuals Identify 58 Loci for Chronic Inflammation and Highlight Pathways that Link Inflammation and Complex Disorders. *American Journal of Human Genetics*, **2018**, 103, 691-706	11	151
466	Genome-wide association meta-analysis highlights light-induced signaling as a driver for refractive error. *Nature Genetics*, **2018**, 50, 834-848	36.3	135
465	Common and Rare Coding Genetic Variation Underlying the Electrocardiographic PR Interval. *Circulation Genomic and Precision Medicine*, **2018**, 11, e002037	5.2	11
464	Interplay of cell-cell contacts and RhoA/MRTF-A signaling regulates cardiomyocyte identity. *EMBO Journal*, **2018**, 37,	13	46
463	PRUNE1 Deficiency: Expanding the Clinical and Genetic Spectrum. *Neuropediatrics*, **2018**, 49, 330-338	1.6	8
462	Mutations in ATP6V1E1 or ATP6V1A Cause Autosomal-Recessive Cutis Laxa. *American Journal of Human Genetics*, **2017**, 100, 216-227	11	58
461	A homozygous splice variant in AP4S1 mimicking neurodegeneration with brain iron accumulation. *Movement Disorders*, **2017**, 32, 797-799	7	11

460	Immune-Array Analysis in Sporadic Inclusion Body Myositis Reveals HLA-DRB1 Amino Acid Heterogeneity Across the Myositis Spectrum. *Arthritis and Rheumatology*, **2017**, 69, 1090-1099	9.5	24
459	Genome-wide association study in takotsubo syndrome - Preliminary results and future directions. *International Journal of Cardiology*, **2017**, 236, 335-339	3.2	22
458	Biallelic Mutations in DNAJC12 Cause Hyperphenylalaninemia, Dystonia, and Intellectual Disability. *American Journal of Human Genetics*, **2017**, 100, 257-266	11	81
457	Systematic Evaluation of Pleiotropy Identifies 6 Further Loci Associated With ´Coronary Artery ´Disease. *Journal of the American College of Cardiology*, **2017**, 69, 823-836	15.1	146
456	Genomic analyses identify hundreds of variants associated with age at menarche and support a role for puberty timing in cancer risk. *Nature Genetics*, **2017**, 49, 834-841	36.3	257
455	Response to Comment on Adam et al. Metformin Effect on Nontargeted Metabolite Profiles in Patients With Type 2 Diabetes and in Multiple Murine Tissues. Diabetes 2016;65:3776-3785. *Diabetes*, **2017**, 66, e3-e4	0.9	1
454	Exome sequencing revealed a splice site variant in the IQCE gene underlying post-axial polydactyly type A restricted to lower limb. *European Journal of Human Genetics*, **2017**, 25, 960-965	5.3	26
453	1000 Genomes-based meta-analysis identifies 10 novel loci for kidney function. *Scientific Reports*, **2017**, 7, 45040	4.9	70
452	A genome-wide association meta-analysis on lipoprotein (a) concentrations adjusted for apolipoprotein (a) isoforms. *Journal of Lipid Research*, **2017**, 58, 1834-1844	6.3	74
451	Coexisting variants in OSTM1 and MANEAL cause a complex neurodegenerative disorder with NBIA-like brain abnormalities. *European Journal of Human Genetics*, **2017**, 25, 1092-1095	5.3	7
450	Elevated glutaric acid levels in Dhtkd1-/Gcdh- double knockout mice challenge our current understanding of lysine metabolism. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2017** , 1863, 2220-2228	6.9	23
449	An Expanded Genome-Wide Association Study of Type 2 Diabetes in Europeans. *Diabetes*, **2017**, 66, 2888-2902	9.3	414
448	Genetic diagnosis of Mendelian disorders via RNA sequencing. *Nature Communications*, **2017**, 8, 15824	17.4	277
447	and Loci Associate with Plasma Osmolality. *Journal of the American Society of Nephrology: JASN*, **2017**, 28, 2311-2321	12.7	14
446	A Low-Frequency Inactivating Variant Enriched in the Finnish Population Is Associated With Fasting Insulin Levels and Type 2 Diabetes Risk. *Diabetes*, **2017**, 66, 2019-2032	0.9	29
445	Identification of a de novo microdeletion 1q44 in a patient with hypogenesis of the corpus callosum, seizures and microcephaly - A case report. *Gene*, **2017**, 616, 41-44	3.8	6
444	CAD mutations and uridine-responsive epileptic encephalopathy. *Brain*, **2017**, 140, 279-286	11.2	77
443	Mutations in MDH2, Encoding a Krebs Cycle Enzyme, Cause Early-Onset Severe Encephalopathy. *American Journal of Human Genetics*, **2017**, 100, 151-159	11	38

442	Epigenome-wide association study of body mass index, and the adverse outcomes of adiposity. *Nature*, **2017**, 541, 81-86	50.4	511
441	CNV-association meta-analysis in 191,161 European adults reveals new loci associated with anthropometric traits. *Nature Communications*, **2017**, 8, 744	17.4	37
440	Biallelic C1QBP Mutations Cause Severe Neonatal-, Childhood-, or Later-Onset Cardiomyopathy Associated with Combined Respiratory-Chain Deficiencies. *American Journal of Human Genetics*, **2017**, 101, 525-538	11	44
439	Impact of common genetic determinants of Hemoglobin A1c on type 2 diabetes risk and diagnosis in ancestrally diverse populations: A transethnic genome-wide meta-analysis. *PLoS Medicine*, **2017**, 14, e1002383	11.6	223
438	A genomic exploration identifies mechanisms that may explain adverse cardiovascular effects of COX-2 inhibitors. *Scientific Reports*, **2017**, 7, 10252	4.9	10
437	Biallelic Mutations in LIPT2 Cause a Mitochondrial Lipoylation Defect Associated with Severe Neonatal Encephalopathy. *American Journal of Human Genetics*, **2017**, 101, 283-290	11	31
436	Large meta-analysis of genome-wide association studies identifies five loci for lean body mass. *Nature Communications*, **2017**, 8, 80	17.4	88
435	Fifteen Genetic Loci Associated With the Electrocardiographic P Wave. *Circulation: Cardiovascular Genetics*, **2017**, 10,		24
434	Transcriptome-Wide Analysis Identifies Novel Associations With Blood Pressure. *Hypertension*, **2017** , 70, 743-750	8.5	21
433	Network inference from glycoproteomics data reveals new reactions in the IgG glycosylation pathway. *Nature Communications*, **2017**, 8, 1483	17.4	26
432	Investigating the causal effect of smoking on hay fever and asthma: a Mendelian randomization meta-analysis in the CARTA consortium. *Scientific Reports*, **2017**, 7, 2224	4.9	24
431	Genome-wide methylation data mirror ancestry information. *Epigenetics and Chromatin*, **2017**, 10, 1	5.8	48
430	Exome sequencing reveals a novel homozygous splice site variant in the WNT1 gene underlying osteogenesis imperfecta type 3. *Pediatric Research*, **2017**, 82, 753-758	3.2	17
429	A new missense mutation in UMOD gene leads to severely reduced serum uromodulin concentrations - A tool for the diagnosis of uromodulin-associated kidney disease. *Clinical Biochemistry*, **2017**, 50, 155-158	3.5	9
428	Neonatal encephalocardiomyopathy caused by mutations in VARS2. *Metabolic Brain Disease*, **2017**, 32, 267-270	3.9	21
427	Ellis-van Creveld syndrome and profound deafness resulted by sequence variants in the EVC/EVC2 and TMC1 genes. *Journal of Genetics*, **2017**, 96, 1005-1014	1.2	8
426	Sequence data and association statistics from 12,940 type 2 diabetes cases and controls. *Scientific Data*, **2017**, 4, 170179	8.2	22
425	pulver: an R package for parallel ultra-rapid p-value computation for linear regression interaction terms. *BMC Bioinformatics*, **2017**, 18, 429	3.6	0

424	Identification of a Novel Heterozygous 7-bp Frameshift Deletion in by Whole-Exome Sequencing Causing a Multi-Organ Syndrome Including Bilateral Dysplastic Kidneys and Hypoplastic Clavicles. *Frontiers in Pediatrics*, **2017**, 5, 251	3.4	9
423	Combined Respiratory Chain Deficiency and Mutations in Neonatal Encephalomyopathy: Defective Supercomplex Assembly in Complex III Deficiencies. *Oxidative Medicine and Cellular Longevity*, **2017**, 2017, 7202589	6.7	27
422	Genomic correlates of glatiramer acetate adverse cardiovascular effects lead to a novel locus mediating coronary risk. *PLoS ONE*, **2017**, 12, e0182999	3.7	3
421	Bayesian and frequentist analysis of an Austrian genome-wide association study of colorectal cancer and advanced adenomas. *Oncotarget*, **2017**, 8, 98623-98634	3.3	12
420	Sequence variants in four genes underlying Bardet-Biedl syndrome in consanguineous families. *Molecular Vision*, **2017**, 23, 482-494	2.3	19
419	Fatal neonatal encephalopathy and lactic acidosis caused by a homozygous loss-of-function variant in COQ9. *European Journal of Human Genetics*, **2016**, 24, 450-4	5.3	31
418	Genetic Modifiers for the Long-QT Syndrome: How Important Is the Role of Variants in the 3O Untranslated Region of KCNQ1?. *Circulation: Cardiovascular Genetics*, **2016**, 9, 330-9		16
417	A reference panel of 64,976 haplotypes for genotype imputation. *Nature Genetics*, **2016**, 48, 1279-83	36.3	1447
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415	Biallelic Variants in UBA5 Reveal that Disruption of the UFM1 Cascade Can Result in Early-Onset Encephalopathy. *American Journal of Human Genetics*, **2016**, 99, 695-703	11	47
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8	Genetic discovery and translational decision support from exome sequencing of 20,791 type 2 diabetes cases and 24,440 controls from five ancestries	2
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4	Integration of proteomics with genomics and transcriptomics increases the diagnostic rate of Mendelian disorders	8
3	Clinical implementation of RNA sequencing for Mendelian disease diagnostics	4
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1	Discovery and systematic characterization of risk variants and genes for coronary artery disease in over a million participants	5