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List of Publications by Year in Descending Order

440 papers	26,390 citations	81 h-index	146 g-index
474 ext. papers	30,226 ext. citations	7.9 avg, IF	5.94 L-index

#	Paper	IF	Citations
440	SRD5A3-CDG: Twins with an intragenic tandem duplication.. *European Journal of Medical Genetics*, **2022**, 65, 104492	2.6	3
439	Astrogenesis in the murine dentate gyrus is a life-long and dynamic process.. *EMBO Journal*, **2022**, e110409	13	2
438	Biallelic ANKS6 mutations cause late onset ciliopathy with chronic kidney disease through YAP dysregulation. *Human Molecular Genetics*, **2021**,	5.6	1
437	Clinical and molecular delineation of spondylocostal dysostosis type 3. *Clinical Genetics*, **2021**, 99, 851-852	4	0
436	EIF3F-related neurodevelopmental disorder: refining the phenotypic and expanding the molecular spectrum. *Orphanet Journal of Rare Diseases*, **2021**, 16, 136	4.2	1
435	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. *Genetics in Medicine*, **2021**, 23, 1028-1040	8.1	7
434	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. *Genome Medicine*, **2021**, 13, 63	14.4	9
433	Urine Metabolite Levels, Adverse Kidney Outcomes, and Mortality in CKD Patients: A Metabolome-wide Association Study. *American Journal of Kidney Diseases*, **2021**, 78, 669-677.e1	7.4	1
432	Mutations in and Other Panel Genes in Patients With Metastatic Breast Cancer -Association With Patient and Disease Characteristics and Effect on Prognosis. *Journal of Clinical Oncology*, **2021**, 39, 1619-1630	2.2	11
431	A noninvasive diagnostic approach to retrospective donor HLA typing in kidney transplant patients using urine. *Transplant International*, **2021**, 34, 1226-1238	3	0
430	Genome sequencing in families with congenital limb malformations. *Human Genetics*, **2021**, 140, 1229-1239	3.9	0
429	QRICH1 variants in Ververi-Brady syndrome-delineation of the genotypic and phenotypic spectrum. *Clinical Genetics*, **2021**, 99, 199-207	4	1
428	De novo variants in MED12 cause X-linked syndromic neurodevelopmental disorders in 18 females. *Genetics in Medicine*, **2021**, 23, 645-652	8.1	8
427	Early-onset parkinsonism in PPP2R5D-related neurodevelopmental disorder. *European Journal of Medical Genetics*, **2021**, 64, 104123	2.6	5
426	DLG4-related synaptopathy: a new rare brain disorder. *Genetics in Medicine*, **2021**, 23, 888-899	8.1	0
425	Association of Rare CYP39A1 Variants With Exfoliation Syndrome Involving the Anterior Chamber of the Eye. *JAMA - Journal of the American Medical Association*, **2021**, 325, 753-764	27.4	6
424	ZMYND11 variants are a novel cause of centrotemporal and generalised epilepsies with neurodevelopmental disorder. *Clinical Genetics*, **2021**, 100, 412-429	4	0

423	Frequent LPA KIV-2 Variants Lower Lipoprotein(a) Concentrations and Protect Against Coronary Artery Disease. *Journal of the American College of Cardiology*, **2021**, 78, 437-449	15.1	15
422	Genetic Analysis of MPO Variants in Four Psoriasis Subtypes in Patients from Germany. *Journal of Investigative Dermatology*, **2021**, 141, 2079-2083	4.3	2
421	BDV Syndrome: An Emerging Syndrome With Profound Obesity and Neurodevelopmental Delay Resembling Prader-Willi Syndrome. *Journal of Clinical Endocrinology and Metabolism*, **2021**, 106, 3413-3427	5.6	3
420	The recurrent missense mutation p.(Arg367Trp) in YARS1 causes a distinct neurodevelopmental phenotype. *Journal of Molecular Medicine*, **2021**, 99, 1755-1768	5.5	0
419	Manifestation of epilepsy in a patient with EED-related overgrowth (Cohen-Gibson syndrome). *American Journal of Medical Genetics, Part A*, **2021**,	2.5	0
418	A case of severe autosomal recessive spinocerebellar ataxia type 18 with a novel nonsense variant in GRID2. *European Journal of Medical Genetics*, **2020**, 63, 103998	2.6	3
417	Sox11 is an Activity-Regulated Gene with Dentate-Gyrus-Specific Expression Upon General Neural Activation. *Cerebral Cortex*, **2020**, 30, 3731-3743	5.1	1
416	Role of Endogenous Regulators of Hem- And Lymphangiogenesis in Corneal Transplantation. *Journal of Clinical Medicine*, **2020**, 9,	5.1	4
415	Rare Loss-of-Function Mutation in SERPINA3 in Generalized Pustular Psoriasis. *Journal of Investigative Dermatology*, **2020**, 140, 1451-1455.e13	4.3	22
414	Transcription factor Tcf4 is the preferred heterodimerization partner for Olig2 in oligodendrocytes and required for differentiation. *Nucleic Acids Research*, **2020**, 48, 4839-4857	20.1	11
413	Further delineation of the female phenotype with KDM5C disease causing variants: 19 new individuals and review of the literature. *Clinical Genetics*, **2020**, 98, 43-55	4	7
412	Genetic interaction screen for severe neurodevelopmental disorders reveals a functional link between Ube3a and Mef2 in Drosophila melanogaster. *Scientific Reports*, **2020**, 10, 1204	4.9	4
411	De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder. *Genetics in Medicine*, **2020**, 22, 538-546	8.1	14
410	Molecular diagnosis of kidney transplant failure based on urine. *American Journal of Transplantation*, **2020**, 20, 1410-1416	8.7	1
409	De novo CLTC variants are associated with a variable phenotype from mild to severe intellectual disability, microcephaly, hypoplasia of the corpus callosum, and epilepsy. *Genetics in Medicine*, **2020**, 22, 797-802	8.1	6
408	Genetic and phenotypic spectrum associated with IFIH1 gain-of-function. *Human Mutation*, **2020**, 41, 837-849	4.7	31
407	CRISPR/Cas9 mediated generation of human ARID1B heterozygous knockout hESC lines to model Coffin-Siris syndrome. *Stem Cell Research*, **2020**, 47, 101889	1.6	3
406	Targeted sequencing of FH-deficient uterine leiomyomas reveals biallelic inactivating somatic fumarase variants and allows characterization of missense variants. *Modern Pathology*, **2020**, 33, 2341-2353	6.8	4

405	Variants in SCAF4 Cause a Neurodevelopmental Disorder and Are Associated with Impaired mRNA Processing. *American Journal of Human Genetics*, **2020**, 107, 544-554	11	2
404	Myeloperoxidase Modulates Inflammation in Generalized Pustular Psoriasis and Additional Rare Pustular Skin Diseases. *American Journal of Human Genetics*, **2020**, 107, 527-538	11	16
403	Breast MRI texture analysis for prediction of BRCA-associated genetic risk. *BMC Medical Imaging*, **2020**, 20, 86	2.9	1
402	Clinical and molecular description of 19 patients with GATAD2B-Associated Neurodevelopmental Disorder (GAND). *European Journal of Medical Genetics*, **2020**, 63, 104004	2.6	3
401	A novel splice variant expands the LAMC3-associated cortical phenotype to frontal only polymicrogyria and adult-onset epilepsy. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2761-2764	2.5	1
400	7q31.2q31.31 deletion downstream of FOXP2 segregating in a family with speech and language disorder. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2737-2741	2.5	1
399	Loss of PHF6 leads to aberrant development of human neuron-like cells. *Scientific Reports*, **2020**, 10, 19030	4.9	
398	Drugs linked to plasma homoarginine in chronic kidney disease patients-a cross-sectional analysis of the German Chronic Kidney Disease cohort. *Nephrology Dialysis Transplantation*, **2020**, 35, 1187-1195	4.3	3
397	Results from the German Chronic Kidney Disease (GCKD) study support association of relative telomere length with mortality in a large cohort of patients with moderate chronic kidney disease. *Kidney International*, **2020**, 98, 488-497	9.9	7
396	Prenatal diagnosis of HNF1B-associated renal cysts: Is there a need to differentiate intragenic variants from 17q12 microdeletion syndrome?. *Prenatal Diagnosis*, **2019**, 39, 1136-1147	3.2	3
395	Cost effectiveness of bilateral risk-reducing mastectomy and salpingo-oophorectomy. *European Journal of Medical Research*, **2019**, 24, 32	4.8	3
394	TRIM28 haploinsufficiency predisposes to Wilms tumor. *International Journal of Cancer*, **2019**, 145, 941-951	7.3	26
393	Inflammation-induced glycolytic switch controls suppressivity of mesenchymal stem cells via STAT1 glycosylation. *Leukemia*, **2019**, 33, 1783-1796	10.7	25
392	Diagnosis and management in Pitt-Hopkins syndrome: First international consensus statement. *Clinical Genetics*, **2019**, 95, 462-478	4	34
391	Prevalence of FOXC1 Variants in Individuals With a Suspected Diagnosis of Primary Congenital Glaucoma. *JAMA Ophthalmology*, **2019**, 137, 348-355	3.9	16
390	Intellectual disability and autism spectrum disorders 'on the fly': insights from. *DMM Disease Models and Mechanisms*, **2019**, 12,	4.1	22
389	Dissecting TSC2-mutated renal and hepatic angiomyolipomas in an individual with ARID1B-associated intellectual disability. *BMC Cancer*, **2019**, 19, 435	4.8	1
388	Mutant RAMP2 causes primary open-angle glaucoma via the CRLR-cAMP axis. *Genetics in Medicine*, **2019**, 21, 2345-2354	8.1	9

387	A novel human stem cell model for Coffin-Siris syndrome-like syndrome reveals the importance of SOX11 dosage for neuronal differentiation and survival. *Human Molecular Genetics*, **2019**, 28, 2589-2599	5.6	8
386	Mutations in PIK3C2A cause syndromic short stature, skeletal abnormalities, and cataracts associated with ciliary dysfunction. *PLoS Genetics*, **2019**, 15, e1008088	6	27
385	The protective variant rs7173049 at LOXL1 locus impacts on retinoic acid signaling pathway in pseudoexfoliation syndrome. *Human Molecular Genetics*, **2019**, 28, 2531-2548	5.6	10
384	TCF4 (E2-2) harbors tumor suppressive functions in SHH medulloblastoma. *Acta Neuropathologica*, **2019**, 137, 657-673	14.3	12
383	Analyses of association of psoriatic arthritis and psoriasis vulgaris with functional NCF1 variants. *Rheumatology*, **2019**, 58, 915-917	3.9	4
382	The mutational and phenotypic spectrum of TUBA1A-associated tubulinopathy. *Orphanet Journal of Rare Diseases*, **2019**, 14, 38	4.2	23
381	Encephalopathies with KCNC1 variants: genotype-phenotype-functional correlations. *Annals of Clinical and Translational Neurology*, **2019**, 6, 1263-1272	5.3	12
380	Mitochondrial DNA copy number is associated with mortality and infections in a large cohort of patients with chronic kidney disease. *Kidney International*, **2019**, 96, 480-488	9.9	29
379	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. *Genetics in Medicine*, **2019**, 21, 2723-2733	8.1	18
378	Diagnostik seltener Erkrankungen mit „next generation sequencing" angekommen oder abgewehrt?. *Medizinische Genetik*, **2019**, 31, 335-343	0.5	
377	Evolutionary conserved networks of human height identify multiple Mendelian causes of short stature. *European Journal of Human Genetics*, **2019**, 27, 1061-1071	5.3	7
376	Estimating the effect size of the 15Q11.2 BP1-BP2 deletion and its contribution to neurodevelopmental symptoms: recommendations for practice. *Journal of Medical Genetics*, **2019**, 56, 701-710	5.8	22
375	Loss of function of SVBP leads to autosomal recessive intellectual disability, microcephaly, ataxia, and hypotonia. *Genetics in Medicine*, **2019**, 21, 1790-1796	8.1	13
374	Tyrosinase Is a Novel Endogenous Regulator of Developmental and Inflammatory Lymphangiogenesis. *American Journal of Pathology*, **2019**, 189, 440-448	5.8	7
373	A biallelic truncating AEBP1 variant causes connective tissue disorder in two siblings. *American Journal of Medical Genetics, Part A*, **2019**, 179, 50-56	2.5	6
372	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. *Genetics in Medicine*, **2018**, 20, 1175-1185	8.1	80
371	Biallelic intragenic deletion in MASP1 in an adult female with 3MC syndrome. *European Journal of Medical Genetics*, **2018**, 61, 363-368	2.6	15
370	Mutations in the BAF-Complex Subunit DPF2 Are Associated with Coffin-Siris Syndrome. *American Journal of Human Genetics*, **2018**, 102, 468-479	11	37

369	Missense Variants in RHOBTB2 Cause a Developmental and Epileptic Encephalopathy in Humans, and Altered Levels Cause Neurological Defects in Drosophila. *American Journal of Human Genetics*, **2018**, 102, 44-57	11	34
368	BRCA mutations and their influence on pathological complete response and prognosis in a clinical cohort of neoadjuvantly treated breast cancer patients. *Breast Cancer Research and Treatment*, **2018**, 171, 85-94	4.4	40
367	SWI/SNF protein expression status in fumarate hydratase-deficient renal cell carcinoma: immunohistochemical analysis of 32 tumors from 28 patients. *Human Pathology*, **2018**, 77, 139-146	3.7	13
366	Single molecule real time sequencing in ADTKD-MUC1 allows complete assembly of the VNTR and exact positioning of causative mutations. *Scientific Reports*, **2018**, 8, 4170	4.9	24
365	Clinical relevance of systematic phenotyping and exome sequencing in patients with short stature. *Genetics in Medicine*, **2018**, 20, 630-638	8.1	68
364	De Novo Variants in the F-Box Protein FBXO11 in 20 Individuals with a Variable Neurodevelopmental Disorder. *American Journal of Human Genetics*, **2018**, 103, 305-316	11	21
363	Biallelic Expression of Mucin-1 in Autosomal Dominant Tubulointerstitial Kidney Disease: Implications for Nongenetic Disease Recognition. *Journal of the American Society of Nephrology: JASN*, **2018**, 29, 2298-2309	12.7	15
362	□Synuclein oligomers induce early axonal dysfunction in human iPSC-based models of synucleinopathies. *Proceedings of the National Academy of Sciences of the United States of America*, **2018**, 115, 7813-7818	11.5	111
361	Analysis of the expression pattern of the schizophrenia-risk and intellectual disability gene TCF4 in the developing and adult brain suggests a role in development and plasticity of cortical and hippocampal neurons. *Molecular Autism*, **2018**, 9, 20	6.5	27
360	Integrative bioinformatics analysis characterizing the role of EDC3 in mRNA decay and its association to intellectual disability. *BMC Medical Genomics*, **2018**, 11, 41	3.7	5
359	The polynucleotide kinase 3'-phosphatase gene (PNKP) is involved in Charcot-Marie-Tooth disease (CMT2B2) previously related to MED25. *Neurogenetics*, **2018**, 19, 215-225	3	23
358	Risk, Prediction and Prevention of Hereditary Breast Cancer - Large-Scale Genomic Studies in Times of Big and Smart Data. *Geburtshilfe Und Frauenheilkunde*, **2018**, 78, 481-492	2	27
357	Novel STRA6 null mutations in the original family described with Matthew-Wood syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 134-138	2.5	6
356	Evidence for genetic overlap between adult onset Still's disease and hereditary periodic fever syndromes. *Rheumatology International*, **2018**, 38, 111-120	3.6	8
355	Genetik der allgemeinen kognitiven F□higkeit. *Medizinische Genetik*, **2018**, 30, 306-317	0.5	
354	Microphthalmia is not a mandatory finding in X-linked recessive syndromic microphthalmia caused by the recurrent BCOR variant p.Pro85Leu. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2872-2876	2.5	1
353	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. *Nature Communications*, **2018**, 9, 4619	17.4	39
352	X-chromosomale Entwicklungsst□rungen im weiblichen Geschlecht. *Medizinische Genetik*, **2018**, 30, 334-341	0.5	

351	Need for high-resolution Genetic Analysis in iPSC: Results and Lessons from the ForIPS Consortium. *Scientific Reports*, **2018**, 8, 17201	4.9	35
350	Variants in PUS7 Cause Intellectual Disability with Speech Delay, Microcephaly, Short Stature, and Aggressive Behavior. *American Journal of Human Genetics*, **2018**, 103, 1045-1052	11	51
349	Addition of triple negativity of breast cancer as an indicator for germline mutations in predisposing genes increases sensitivity of clinical selection criteria. *BMC Cancer*, **2018**, 18, 926	4.8	4
348	Novel truncating mutation in CACNA1F in a young male patient diagnosed with optic atrophy. *Ophthalmic Genetics*, **2018**, 39, 741-748	1.2	4
347	De novo mutations in MSL3 cause an X-linked syndrome marked by impaired histone H4 lysine 16 acetylation. *Nature Genetics*, **2018**, 50, 1442-1451	36.3	19
346	Diagnostic Yield and Novel Candidate Genes by Exome Sequencing in 152 Consanguineous Families With Neurodevelopmental Disorders. *JAMA Psychiatry*, **2017**, 74, 293-299	14.5	116
345	A Homozygous Mutation in GPT2 Associated with Nonsyndromic Intellectual Disability in a Consanguineous Family from Costa Rica. *JIMD Reports*, **2017**, 36, 59-66	1.9	5
344	Cost-effectiveness of risk-reducing surgeries in preventing hereditary breast and ovarian cancer. *Breast*, **2017**, 32, 186-191	3.6	18
343	Association Between Telomere Length and Risk of Cancer and Non-Neoplastic Diseases: A Mendelian Randomization Study. *JAMA Oncology*, **2017**, 3, 636-651	13.4	236
342	FOXP2 variants in 14 individuals with developmental speech and language disorders broaden the mutational and clinical spectrum. *Journal of Medical Genetics*, **2017**, 54, 64-72	5.8	43
341	Hypomorphic Pathogenic Variants in TAF13 Are Associated with Autosomal-Recessive Intellectual Disability and Microcephaly. *American Journal of Human Genetics*, **2017**, 100, 555-561	11	13
340	mutations cause a novel cohesinopathy characterised by unspecific syndromic intellectual disability. *Journal of Medical Genetics*, **2017**, 54, 479-488	5.8	25
339	Pseudoexfoliation syndrome-associated genetic variants affect transcription factor binding and alternative splicing of LOXL1. *Nature Communications*, **2017**, 8, 15466	17.4	28
338	Haploinsufficiency of NR4A2 is associated with a neurodevelopmental phenotype with prominent language impairment. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2231-2234	2.5	17
337	Large scale meta-analysis characterizes genetic architecture for common psoriasis associated variants. *Nature Communications*, **2017**, 8, 15382	17.4	136
336	Genetic association study of exfoliation syndrome identifies a protective rare variant at LOXL1 and five new susceptibility loci. *Nature Genetics*, **2017**, 49, 993-1004	36.3	72
335	DDX3X mutations in two girls with a phenotype overlapping Toriello-Carey syndrome. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1369-1373	2.5	23
334	Central nervous system anomalies in two females with Borjeson-Forssman-Lehmann syndrome. *Epilepsy and Behavior*, **2017**, 69, 104-109	3.2	4

333	Bainbridge-Ropers syndrome caused by loss-of-function variants in ASXL3: a recognizable condition. *European Journal of Human Genetics*, **2017**, 25, 183-191	5.3	28
332	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. *Nature Genetics*, **2017**, 49, 223-237	36.3	116
331	Haploinsufficiency of the Chromatin Remodeler BPTF Causes Syndromic Developmental and Speech Delay, Postnatal Microcephaly, and Dysmorphic Features. *American Journal of Human Genetics*, **2017**, 101, 503-515	11	37
330	Genome-wide association and targeted analysis of copy number variants with psoriatic arthritis in German patients. *BMC Medical Genetics*, **2017**, 18, 92	2.1	6
329	Posttranscriptional Regulation of LOXL1 Expression Via Alternative Splicing and Nonsense-Mediated mRNA Decay as an Adaptive Stress Response **2017**, 58, 5930-5940		14
328	Genetic screening confirms heterozygous mutations in ACAN as a major cause of idiopathic short stature. *Scientific Reports*, **2017**, 7, 12225	4.9	37
327	Genetic Breast Cancer Susceptibility Variants and Prognosis in the Prospectively Randomized SUCCESS A Study. *Geburtshilfe Und Frauenheilkunde*, **2017**, 77, 651-659	2	12
326	Exome Pool-Seq in neurodevelopmental disorders. *European Journal of Human Genetics*, **2017**, 25, 1364-1376	5.3	52
325	AMPA-receptor specific biogenesis complexes control synaptic transmission and intellectual ability. *Nature Communications*, **2017**, 8, 15910	17.4	39
324	Gene panel sequencing in familial breast/ovarian cancer patients identifies multiple novel mutations also in genes others than BRCA1/2. *International Journal of Cancer*, **2017**, 140, 95-102	7.5	71
323	Exome-wide association study reveals novel psoriasis susceptibility locus at TNFSF15 and rare protective alleles in genes contributing to type I IFN signalling. *Human Molecular Genetics*, **2017**, 26, 4301-4313	5.6	25
322	Clinical validation of genetic variants associated with chemotherapy-related lymphoblastoid cell toxicity. *Oncotarget*, **2017**, 8, 78133-78143	3.3	4
321	and are disease-specific biomarkers for psoriatic arthritis susceptibility. *Oncotarget*, **2017**, 8, 95401-95413	3.3	8
320	A recessive form of extreme macrocephaly and mild intellectual disability complements the spectrum of PTEN hamartoma tumour syndrome. *European Journal of Human Genetics*, **2016**, 24, 889-94	5.3	5
319	Mutations in MBOAT7, Encoding Lysophosphatidylinositol Acyltransferase I, Lead to Intellectual Disability Accompanied by Epilepsy and Autistic Features. *American Journal of Human Genetics*, **2016**, 99, 912-916	11	48
318	SPATA5 mutations cause a distinct autosomal recessive phenotype of intellectual disability, hypotonia and hearing loss. *Orphanet Journal of Rare Diseases*, **2016**, 11, 130	4.2	15
317	Replication of a distinct psoriatic arthritis risk variant at the IL23R locus. *Annals of the Rheumatic Diseases*, **2016**, 75, 1417-8	2.4	5
316	Expanding the Phenotype Associated with NAA10-Related N-Terminal Acetylation Deficiency. *Human Mutation*, **2016**, 37, 755-64	4.7	49

315	Systematic Phenomics Analysis Deconvolutes Genes Mutated in Intellectual Disability into Biologically Coherent Modules. *American Journal of Human Genetics*, **2016**, 98, 149-64	11	154
314	Glycaemic control and antidiabetic therapy in patients with diabetes mellitus and chronic kidney disease - cross-sectional data from the German Chronic Kidney Disease (GCKD) cohort. *BMC Nephrology*, **2016**, 17, 59	2.7	12
313	The Slavic NBN Founder Mutation: A Role for Reproductive Fitness?. *PLoS ONE*, **2016**, 11, e0167984	3.7	10
312	Autosomal-Recessive Mutations in the tRNA Splicing Endonuclease Subunit TSEN15 Cause Pontocerebellar Hypoplasia and Progressive Microcephaly. *American Journal of Human Genetics*, **2016**, 99, 228-35	11	26
311	Eight further individuals with intellectual disability and epilepsy carrying bi-allelic CNTNAP2 aberrations allow delineation of the mutational and phenotypic spectrum. *Journal of Medical Genetics*, **2016**, 53, 820-827	5.8	28
310	Genetic and neurodevelopmental spectrum of SYNGAP1-associated intellectual disability and epilepsy. *Journal of Medical Genetics*, **2016**, 53, 511-22	5.8	76
309	SAT0011 Replication of A Distinct Psoriatic Arthritis Risk Variant at IL23R. *Annals of the Rheumatic Diseases*, **2016**, 75, 667.3-668	2.4	
308	A new missense mutation in PLA2G6 gene among a family with infantile neuroaxonal dystrophy INAD. *The Gazette of the Egyptian Paediatric Association*, **2016**, 64, 171-176	1.4	3
307	The Clinical Data Intelligence Project. *Informatik-Spektrum*, **2016**, 39, 290-300	0.3	5
306	Synuclein-induced myelination deficit defines a novel interventional target for multiple system atrophy. *Acta Neuropathologica*, **2016**, 132, 59-75	14.3	42
305	Expanding the clinical spectrum of COL1A1 mutations in different forms of glaucoma. *Orphanet Journal of Rare Diseases*, **2016**, 11, 108	4.2	18
304	Do the exome: A case of Williams-Beuren syndrome with severe epilepsy due to a truncating de novo variant in GABRA1. *European Journal of Medical Genetics*, **2016**, 59, 549-53	2.6	10
303	Association analysis of psoriasis vulgaris and psoriatic arthritis with loss-of-function mutations in IL36RN in German patients. *British Journal of Dermatology*, **2016**, 175, 639-41	4	4
302	GSK3dependent dysregulation of neurodevelopment in SPG11-patient induced pluripotent stem cell model. *Annals of Neurology*, **2016**, 79, 826-840	9.4	32
301	Comprehensive screening for mutations associated with colorectal cancer in unselected cases reveals penetrant and nonpenetrant mutations. *International Journal of Cancer*, **2015**, 136, E559-68	7.5	16
300	A common variant mapping to CACNA1A is associated with susceptibility to exfoliation syndrome. *Nature Genetics*, **2015**, 47, 387-92	36.3	70
299	MAP4-dependent regulation of microtubule formation affects centrosome, cilia, and Golgi architecture as a central mechanism in growth regulation. *Human Mutation*, **2015**, 36, 87-97	4.7	18
298	Inhibition of RAS activation due to a homozygous ezrin variant in patients with profound intellectual disability. *Human Mutation*, **2015**, 36, 270-8	4.7	12

297	De novo missense mutations in the NAA10 gene cause severe non-syndromic developmental delay in males and females. *European Journal of Human Genetics*, **2015**, 23, 602-9	5.3	55
296	Disease burden and risk profile in referred patients with moderate chronic kidney disease: composition of the German Chronic Kidney Disease (GCKD) cohort. *Nephrology Dialysis Transplantation*, **2015**, 30, 441-51	4.3	93
295	Prevalence and correlates of gout in a large cohort of patients with chronic kidney disease: the German Chronic Kidney Disease (GCKD) study. *Nephrology Dialysis Transplantation*, **2015**, 30, 613-21	4.3	55
294	Do telomeres have a higher plasticity than thought? Results from the German Chronic Kidney Disease (GCKD) study as a high-risk population. *Experimental Gerontology*, **2015**, 72, 162-6	4.5	13
293	Genetic variants and cellular stressors associated with exfoliation syndrome modulate promoter activity of a lncRNA within the LOXL1 locus. *Human Molecular Genetics*, **2015**, 24, 6552-63	5.6	61
292	Chromatin-Remodeling-Factor ARID1B Represses Wnt/□Catenin Signaling. *American Journal of Human Genetics*, **2015**, 97, 445-56	11	51
291	Genome-wide Association Analysis of Psoriatic Arthritis and Cutaneous Psoriasis Reveals Differences in Their Genetic Architecture. *American Journal of Human Genetics*, **2015**, 97, 816-36	11	185
290	Dense genotyping of immune-related susceptibility loci reveals new insights into the genetics of psoriatic arthritis. *Nature Communications*, **2015**, 6, 6046	17.4	103
289	De novo mutations in beta-catenin (CTNNB1) appear to be a frequent cause of intellectual disability: expanding the mutational and clinical spectrum. *Human Genetics*, **2015**, 134, 97-109	6.3	62
288	A de novo microdeletion in NRXN1 in a Dutch patient with mild intellectual disability, microcephaly and gonadal dysgenesis. *Genetical Research*, **2015**, 97, e19	1.1	
287	DYNC2LI1 mutations broaden the clinical spectrum of dynein-2 defects. *Scientific Reports*, **2015**, 5, 11649	4.9	25
286	Mikrozephalie bei psychomotorischen Entwicklungsst□ungen und geistiger Behinderung. *Medizinische Genetik*, **2015**, 27, 362-368	0.5	1
285	50 Jahre Humangenetik in Erlangen. *Medizinische Genetik*, **2015**, 27, 388-392	0.5	
284	OP0128 PTPN22 is Associated with Susceptibility to Psoriatic Arthritis but not Psoriasis: Evidence for a Further PSA-Specific Risk Locus. *Annals of the Rheumatic Diseases*, **2015**, 74, 116.3-117	2.4	0
283	MAN1B1 Mutation Leads to a Recognizable Phenotype: A Case Report and Future Prospects. *Molecular Syndromology*, **2015**, 6, 58-62	1.5	10
282	Enhanced meta-analysis and replication studies identify five new psoriasis susceptibility loci. *Nature Communications*, **2015**, 6, 7001	17.4	122
281	Whole exome sequencing reveals a novel de novo FOXC1 mutation in a patient with unrecognized Axenfeld-Rieger syndrome and glaucoma. *Gene*, **2015**, 568, 76-80	3.8	8
280	Mutations in DCPS and EDC3 in autosomal recessive intellectual disability indicate a crucial role for mRNA decapping in neurodevelopment. *Human Molecular Genetics*, **2015**, 24, 3172-80	5.6	27

279	Autosomal-Recessive Intellectual Disability with Cerebellar Atrophy Syndrome Caused by Mutation of the Manganese and Zinc Transporter Gene SLC39A8. *American Journal of Human Genetics*, **2015**, 97, 886-93	11	125
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