Christian Gilissen

List of Publications by Year
in descending order

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216	20,505	11235	14386
papers	citations	73 h-index	132 g-index

249	249	249	34782
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Diagnostic Exome Sequencing in Persons with Severe Intellectual Disability. New England Journal of Medicine, 2012, 367, 1921-1929.	13.9	1,367
2	Genome sequencing identifies major causes of severe intellectual disability. Nature, 2014, 511, 344-347.	13.7	996
3	A de novo paradigm for mental retardation. Nature Genetics, 2010, 42, 1109-1112.	9.4	751
4	Presence of Genetic Variants Among Young Men With Severe COVID-19. JAMA - Journal of the American Medical Association, 2020, 324, 663.	3.8	626
5	STAT1Mutations in Autosomal Dominant Chronic Mucocutaneous Candidiasis. New England Journal of Medicine, 2011, 365, 54-61.	13.9	614
6	Genetic studies in intellectual disability and related disorders. Nature Reviews Genetics, 2016, 17, 9-18.	7.7	614
7	De novo mutations of SETBP1 cause Schinzel-Giedion syndrome. Nature Genetics, 2010, 42, 483-485.	9.4	417
8	Disease gene identification strategies for exome sequencing. European Journal of Human Genetics, 2012, 20, 490-497.	1.4	412
9	Meta-analysis of 2,104 trios provides support for 10 new genes for intellectual disability. Nature Neuroscience, 2016, 19, 1194-1196.	7.1	407
10	A recent bottleneck of Y chromosome diversity coincides with a global change in culture. Genome Research, 2015, 25, 459-466.	2.4	348
11	Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature, 2020, 586, 757-762.	13.7	343
12	Exome Sequencing Identifies Truncating Mutations in Human SERPINF1 in Autosomal-Recessive Osteogenesis Imperfecta. American Journal of Human Genetics, 2011, 88, 362-371.	2.6	316
13	Mutations in SWI/SNF chromatin remodeling complex gene ARID1B cause Coffin-Siris syndrome. Nature Genetics, 2012, 44, 379-380.	9.4	312
14	A germline homozygous mutation in the base-excision repair gene NTHL1 causes adenomatous polyposis and colorectal cancer. Nature Genetics, 2015, 47, 668-671.	9.4	311
15	A Post-Hoc Comparison of the Utility of Sanger Sequencing and Exome Sequencing for the Diagnosis of Heterogeneous Diseases. Human Mutation, 2013, 34, 1721-1726.	1.1	303
16	Improved exome prioritization of disease genes through cross-species phenotype comparison. Genome Research, 2014, 24, 340-348.	2.4	300
17	Whole-genome sequencing reveals important role for TBK1 and OPTN mutations in frontotemporal lobar degeneration without motor neuron disease. Acta Neuropathologica, 2015, 130, 77-92.	3.9	267
18	Parent-of-origin-specific signatures of de novo mutations. Nature Genetics, 2016, 48, 935-939.	9.4	266

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19	Exome Sequencing Identifies WDR35 Variants Involved in Sensenbrenner Syndrome. American Journal of Human Genetics, 2010, 87, 418-423.	2.6	260
20	Nextâ€generation genetic testing for retinitis pigmentosa. Human Mutation, 2012, 33, 963-972.	1.1	258
21	De novo mutations in the actin genes ACTB and ACTG1 cause Baraitser-Winter syndrome. Nature Genetics, 2012, 44, 440-444.	9.4	237
22	De novo nonsense mutations in ASXL1 cause Bohring-Opitz syndrome. Nature Genetics, 2011, 43, 729-731.	9.4	236
23	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. American Journal of Human Genetics, 2015, 97, 343-352.	2.6	230
24	Unlocking Mendelian disease using exome sequencing. Genome Biology, 2011, 12, 228.	13.9	228
25	Post-zygotic Point Mutations Are an Underrecognized Source of De Novo Genomic Variation. American Journal of Human Genetics, 2015, 97, 67-74.	2.6	215
26	Disruption of an EHMT1-Associated Chromatin-Modification Module Causes Intellectual Disability. American Journal of Human Genetics, 2012, 91, 73-82.	2.6	214
27	Ciliopathies with Skeletal Anomalies and Renal Insufficiency due to Mutations in the IFT-A Gene WDR19. American Journal of Human Genetics, 2011, 89, 634-643.	2.6	210
28	Ultra-sensitive Sequencing Identifies High Prevalence of Clonal Hematopoiesis-Associated Mutations throughout Adult Life. American Journal of Human Genetics, 2017, 101, 50-64.	2.6	210
29	Next-Generation Sequencing of a 40 Mb Linkage Interval Reveals TSPAN12 Mutations in Patients with Familial Exudative Vitreoretinopathy. American Journal of Human Genetics, 2010, 86, 240-247.	2.6	202
30	Mutations in the chromatin modifier gene KANSL1 cause the 17q21.31 microdeletion syndrome. Nature Genetics, 2012, 44, 639-641.	9.4	194
31	Mutations in ISPD cause Walker-Warburg syndrome and defective glycosylation of Î±-dystroglycan. Nature Genetics, 2012, 44, 581-585.	9.4	191
32	Mutations in the phospholipid remodeling gene SERAC1 impair mitochondrial function and intracellular cholesterol trafficking and cause dystonia and deafness. Nature Genetics, 2012, 44, 797-802.	9.4	175
33	Mutations in DDHD2, Encoding an Intracellular Phospholipase A1, Cause a Recessive Form of Complex Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2012, 91, 1073-1081.	2.6	159
34	Comparison of Exome and Genome Sequencing Technologies for the Complete Capture of Proteinâ€Coding Regions. Human Mutation, 2015, 36, 815-822.	1.1	156
35	Neurocalcin Delta Suppression Protects against Spinal Muscular Atrophy in Humans and across Species by Restoring Impaired Endocytosis. American Journal of Human Genetics, 2017, 100, 297-315.	2.6	156
36	Mutations in BICD2, which Encodes a Golgin and Important Motor Adaptor, Cause Congenital Autosomal-Dominant Spinal Muscular Atrophy. American Journal of Human Genetics, 2013, 92, 946-954.	2.6	150

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37	Dystrophin Gene Mutation Location and the Risk of Cognitive Impairment in Duchenne Muscular Dystrophy. PLoS ONE, 2010, 5, e8803.	1.1	147
38	Gene identification in the congenital disorders of glycosylation type I by whole-exome sequencing. Human Molecular Genetics, 2012, 21, 4151-4161.	1.4	147
39	Nextâ€generation metabolic screening: targeted and untargeted metabolomics for the diagnosis of inborn errors of metabolism in individual patients. Journal of Inherited Metabolic Disease, 2018, 41, 337-353.	1.7	145
40	ZNF408 is mutated in familial exudative vitreoretinopathy and is crucial for the development of zebrafish retinal vasculature. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 9856-9861.	3.3	144
41	Rare pathogenic microdeletions and tandem duplications are microhomology-mediated and stimulated by local genomic architecture. Human Molecular Genetics, 2009, 18, 3579-3593.	1.4	143
42	Detection of clinically relevant copy-number variants by exome sequencing in a large cohort of genetic disorders. Genetics in Medicine, 2017, 19, 667-675.	1.1	143
43	CantÃº Syndrome Is Caused by Mutations in ABCC9. American Journal of Human Genetics, 2012, 90, 1094-1101.	2.6	141
44	Homozygous and heterozygous disruptions of ANK3: at the crossroads of neurodevelopmental and psychiatric disorders. Human Molecular Genetics, 2013, 22, 1960-1970.	1.4	137
45	MetaDome: Pathogenicity analysis of genetic variants through aggregation of homologous human protein domains. Human Mutation, 2019, 40, 1030-1038.	1.1	133
46	Exome sequencing identifiesDYNC2H1mutations as a common cause of asphyxiating thoracic dystrophy (Jeune syndrome) without major polydactyly, renal or retinal involvement. Journal of Medical Genetics, 2013, 50, 309-323.	1.5	127
47	Novel genetic causes for cerebral visual impairment. European Journal of Human Genetics, 2016, 24, 660-665.	1.4	127
48	Targeted Next-Generation Sequencing of a 12.5 Mb Homozygous Region Reveals ANO10 Mutations in Patients with Autosomal-Recessive Cerebellar Ataxia. American Journal of Human Genetics, 2010, 87, 813-819.	2.6	125
49	YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction. American Journal of Human Genetics, 2017, 100, 907-925.	2.6	125
50	Functional Differences Between Mesenchymal Stem Cell Populations Are Reflected by Their Transcriptome. Stem Cells and Development, 2010, 19, 481-490.	1.1	124
51	Exome sequencing of Pakistani consanguineous families identifies 30 novel candidate genes for recessive intellectual disability. Molecular Psychiatry, 2017, 22, 1604-1614.	4.1	118
52	Neu-Laxova Syndrome Is a Heterogeneous Metabolic Disorder Caused by Defects in Enzymes of the L-Serine Biosynthesis Pathway. American Journal of Human Genetics, 2014, 95, 285-293.	2.6	110
53	ATP6AP1 deficiency causes an immunodeficiency with hepatopathy, cognitive impairment and abnormal protein glycosylation. Nature Communications, 2016, 7, 11600.	5.8	110
54	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. Genome Biology, 2014, 15, R53.	13.9	101

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55	Recurrent De Novo Mutations in PACS1 Cause Defective Cranial-Neural-Crest Migration and Define a Recognizable Intellectual-Disability Syndrome. American Journal of Human Genetics, 2012, 91, 1122-1127.	2.6	96
56	De Novo Loss-of-Function Mutations in USP9X Cause a Female-Specific Recognizable Syndrome with Developmental Delay and Congenital Malformations. American Journal of Human Genetics, 2016, 98, 373-381.	2.6	95
57	TRIOloss of function is associated with mild intellectual disability and affects dendritic branching and synapse function. Human Molecular Genetics, 2016, 25, 892-902.	1.4	94
58	Identification of pathogenic gene variants in small families with intellectually disabled siblings by exome sequencing. Journal of Medical Genetics, 2013, 50, 802-811.	1.5	93
59	Resolving the dark matter of ABCA4 for 1054 Stargardt disease probands through integrated genomics and transcriptomics. Genetics in Medicine, 2020, 22, 1235-1246.	1.1	92
60	MicroRNA hsa-miR-135b Regulates Mineralization in Osteogenic Differentiation of Human Unrestricted Somatic Stem Cells. Stem Cells and Development, 2010, 19, 877-885.	1.1	90
61	The diagnostic yield of whole-exome sequencing targeting a gene panel for hearing impairment in The Netherlands. European Journal of Human Genetics, 2017, 25, 308-314.	1.4	90
62	Clinical exome sequencing for cerebellar ataxia and spastic paraplegia uncovers novel geneâ€“disease associations and unanticipated rare disorders. European Journal of Human Genetics, 2016, 24, 1460-1466.	1.4	89
63	Osteo-transcriptomics of human mesenchymal stem cells: Accelerated gene expression and osteoblast differentiation induced by vitamin D reveals c-MYC as an enhancer of BMP2-induced osteogenesis. Bone, 2010, 46, 613-627.	1.4	88
64	Chondrodysplasia and Abnormal Joint Development Associated with Mutations in IMPAD1, Encoding the Golgi-Resident Nucleotide Phosphatase, gPAPP. American Journal of Human Genetics, 2011, 88, 608-615.	2.6	88
65	Massively parallel sequencing of ataxia genes after array-based enrichment. Human Mutation, 2010, 31, 494-499.	1.1	86
66	The heat shock response restricts virus infection in Drosophila. Scientific Reports, 2015, 5, 12758.	1.6	86
67	Mutations in SELENBP1, encoding a novel human methanethiol oxidase, cause extraoral halitosis. Nature Genetics, 2018, 50, 120-129.	9.4	86
68	<scp>MLL2</scp> mutation detection in 86 patients with Kabuki syndrome: a genotypeâ€“phenotype study. Clinical Genetics, 2013, 84, 539-545.	1.0	85
69	Spatial Clustering of de Novo Missense Mutations Identifies Candidate Neurodevelopmental Disorder-Associated Genes. American Journal of Human Genetics, 2017, 101, 478-484.	2.6	84
70	EPHB4 kinaseâ€“inactivating mutations cause autosomal dominant lymphatic-related hydrops fetalis. Journal of Clinical Investigation, 2016, 126, 3080-3088.	3.9	83
71	A complex V ATP5A1 defect causes fatal neonatal mitochondrial encephalopathy. Brain, 2013, 136, 1544-1554.	3.7	80
72	Thyroid hormone resistance syndrome due to mutations in the thyroid hormone receptor Î± gene (THRA). Journal of Medical Genetics, 2015, 52, 312-316.	1.5	80

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73	De Novo Mutations Reflect Development and Aging of the Human Germline. Trends in Genetics, 2019, 35, 828-839.	2.9	80
74	Whole-exome sequencing reveals LRP5 mutations and canonical Wnt signaling associated with hepatic cystogenesis. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 5343-5348.	3.3	79
75	Mutations in MED12 Cause X-Linked Ohdo Syndrome. American Journal of Human Genetics, 2013, 92, 401-406.	2.6	78
76	De novo mutations in PLXND1 and REV3L cause MÃ¶bius syndrome. Nature Communications, 2015, 6, 7199.	5.8	76
77	A mutation in the FAM36A gene, the human ortholog of COX20, impairs cytochrome c oxidase assembly and is associated with ataxia and muscle hypotonia. Human Molecular Genetics, 2013, 22, 656-667.	1.4	75
78	Structural Variants Create New Topological-Associated Domains and Ectopic Retinal Enhancer-Gene Contact in Dominant Retinitis Pigmentosa. American Journal of Human Genetics, 2020, 107, 802-814.	2.6	75
79	Mutations in the interleukin receptor <scp>IL</scp>11<scp>RA</scp> cause autosomal recessive Crouzonâ€like craniosynostosis. Molecular Genetics & Genomic Medicine, 2013, 1, 223-237.	0.6	70
80	BTG1 regulates glucocorticoid receptor autoinduction in acute lymphoblastic leukemia. Blood, 2010, 115, 4810-4819.	0.6	69
81	Identification and functional characterization ofde novo FOXP1variants provides novel insights into the etiology of neurodevelopmental disorder. Human Molecular Genetics, 2016, 25, 546-557.	1.4	69
82	Germline de novo mutation clusters arise during oocyte aging in genomic regions with high double-strand-break incidence. Nature Genetics, 2018, 50, 487-492.	9.4	68
83	Deleterious Germline BLM Mutations and the Risk for Early-onset Colorectal Cancer. Scientific Reports, 2015, 5, 14060.	1.6	67
84	Haploinsufficiency of MeCP2-interacting transcriptional co-repressor SIN3A causes mild intellectual disability by affecting the development of cortical integrity. Nature Genetics, 2016, 48, 877-887.	9.4	67
85	Variation in a range of mTOR-related genes associates with intracranial volume and intellectual disability. Nature Communications, 2017, 8, 1052.	5.8	63
86	Validation Study of Existing Gene Expression Signatures for Anti-TNF Treatment in Patients with Rheumatoid Arthritis. PLoS ONE, 2012, 7, e33199.	1.1	61
87	Exome sequencing identifies a de novo <scp>SCN</scp>2<scp>A</scp> mutation in a patient with intractable seizures, severe intellectual disability, optic atrophy, muscular hypotonia, and brain abnormalities. Epilepsia, 2014, 55, e25-9.	2.6	58
88	Novel mutations in LRP6 highlight the role of WNT signaling in tooth agenesis. Genetics in Medicine, 2016, 18, 1158-1162.	1.1	58
89	Nuclear ReceptorsNur77andNurr1Modulate Mesenchymal Stromal Cell Migration. Stem Cells and Development, 2012, 21, 228-238.	1.1	56
90	1 in 38 individuals at risk of a dominant medically actionable disease. European Journal of Human Genetics, 2019, 27, 325-330.	1.4	56

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91	Identification of Novel Candidate Genes for Early-Onset Colorectal Cancer Susceptibility. PLoS Genetics, 2016, 12, e1005880.	1.5	52
92	Somatic variants in autosomal dominant genes are a rare cause of sporadic Alzheimer's disease. Alzheimer's and Dementia, 2018, 14, 1632-1639.	0.4	51
93	Front Cover, Volume 40, Issue 8. Human Mutation, 2019, 40, i-i.	1.1	51
94	Hematopoietic stem cells exhibit a specific ABC transporter gene expression profile clearly distinct from other stem cells. BMC Pharmacology, 2010, 10, 12.	0.4	50
95	Exome Sequencing in an Admixed Isolated Population Indicates NFXL1 Variants Confer a Risk for Specific Language Impairment. PLoS Genetics, 2015, 11, e1004925.	1.5	50
96	Clinical, molecular, and cellular immunologic findings in patients with SP110-associated veno-occlusive disease with immunodeficiency syndrome. Journal of Allergy and Clinical Immunology, 2012, 130, 735-742.e6.	1.5	49
97	Exome sequencing in routine diagnostics: a generic test for 254 patients with primary immunodeficiencies. Genome Medicine, 2019, 11, 38.	3.6	49
98	Rapid whole exome sequencing in pregnancies to identify the underlying genetic cause in fetuses with congenital anomalies detected by ultrasound imaging. Prenatal Diagnosis, 2020, 40, 972-983.	1.1	49
99	Solve-RD: systematic pan-European data sharing and collaborative analysis to solve rare diseases. European Journal of Human Genetics, 2021, 29, 1325-1331.	1.4	49
100	Panel-based NGS Reveals Novel Pathogenic Mutations in Autosomal Recessive Retinitis Pigmentosa. Scientific Reports, 2016, 6, 19531.	1.6	48
101	Recommendations for whole genome sequencing in diagnostics for rare diseases. European Journal of Human Genetics, 2022, 30, 1017-1021.	1.4	48
102	Accurate Distinction of Pathogenic from Benign CNVs in Mental Retardation. PLoS Computational Biology, 2010, 6, e1000752.	1.5	46
103	Validation and application of a novel integrated genetic screening method to a cohort of 1,112 men with idiopathic azoospermia or severe oligozoospermia. Human Mutation, 2017, 38, 1592-1605.	1.1	45
104	Dominant variants in the splicing factor PUF60 cause a recognizable syndrome with intellectual disability, heart defects and short stature. European Journal of Human Genetics, 2017, 25, 43-51.	1.4	44
105	Rare genetic variants in interleukin-37 link this anti-inflammatory cytokine to the pathogenesis and treatment of gout. Annals of the Rheumatic Diseases, 2020, 79, 536-544.	0.5	44
106	Characterization of SETD1A haploinsufficiency in humans and Drosophila defines a novel neurodevelopmental syndrome. Molecular Psychiatry, 2021, 26, 2013-2024.	4.1	43
107	Population sequencing data reveal a compendium of mutational processes in the human germ line. Science, 2021, 373, 1030-1035.	6.0	43
108	The Discovery of a LEMD2-Associated Nuclear Envelopathy with Early Progeroid Appearance Suggests Advanced Applications for AI-Driven Facial Phenotyping. American Journal of Human Genetics, 2019, 104, 749-757.	2.6	41

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109	Costâ€effective molecular inversion probeâ€based ABCA4 sequencing reveals deepâ€intronic variants in Stargardt disease. Human Mutation, 2019, 40, 1749-1759.	1.1	39
110	A systems genomics approach identifies SIGLEC15 as a susceptibility factor in recurrent vulvovaginal candidiasis. Science Translational Medicine, 2019, 11, .	5.8	38
111	A de novo paradigm for male infertility. Nature Communications, 2022, 13, 154.	5.8	38
112	APR-246/PRIMA-1MET rescues epidermal differentiation in skin keratinocytes derived from EEC syndrome patients with p63 mutations. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 2157-2162.	3.3	37
113	Variants inCUL4Bare Associated with Cerebral Malformations. Human Mutation, 2015, 36, 106-117.	1.1	37
114	Novel bioinformatic developments for exome sequencing. Human Genetics, 2016, 135, 603-614.	1.8	37
115	Aggregation of populationâ€based genetic variation over protein domain homologues and its potential use in genetic diagnostics. Human Mutation, 2017, 38, 1454-1463.	1.1	36
116	The landscape of autosomal-recessive pathogenic variants in European populations reveals phenotype-specific effects. American Journal of Human Genetics, 2021, 108, 608-619.	2.6	36
117	Overlapping SETBP1 gain-of-function mutations in Schinzel-Giedion syndrome and hematologic malignancies. PLoS Genetics, 2017, 13, e1006683.	1.5	35
118	De Novo Variants Disturbing the Transactivation Capacity of POU3F3 Cause a Characteristic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 105, 403-412.	2.6	35
119	Accurate detection of clinically relevant uniparental disomy from exome sequencing data. Genetics in Medicine, 2020, 22, 803-808.	1.1	35
120	De novo loss-of-function mutations in WAC cause a recognizable intellectual disability syndrome and learning deficits in Drosophila. European Journal of Human Genetics, 2016, 24, 1145-1153.	1.4	34
121	Unraveling genetic predisposition to familial or early onset gastric cancer using germline whole-exome sequencing. European Journal of Human Genetics, 2017, 25, 1246-1252.	1.4	34
122	De Novo and Inherited Pathogenic Variants in KDM3B Cause Intellectual Disability, Short Stature, and Facial Dysmorphism. American Journal of Human Genetics, 2019, 104, 758-766.	2.6	34
123	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. European Journal of Human Genetics, 2021, 29, 1337-1347.	1.4	34
124	High BRE expression predicts favorable outcome in adult acute myeloid leukemia, in particular among MLL-AF9â€“positive patients. Blood, 2011, 118, 5613-5621.	0.6	32
125	De novo lossâ€ofâ€function mutations in Xâ€linked <scp>SMC1A</scp> cause severe <scp>ID</scp> and therapyâ€resistant epilepsy in females: expanding the phenotypic spectrum. Clinical Genetics, 2016, 90, 413-419.	1.0	32
126	A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency. European Journal of Human Genetics, 2018, 26, 54-63.	1.4	32

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127	Mutations in the Vâ€ATPase Assembly Factor VMA21 Cause a Congenital Disorder of Glycosylation With Autophagic Liver Disease. Hepatology, 2020, 72, 1968-1986.	3.6	32
128	Syndromic X-linked intellectual disability segregating with a missense variant in RLIM. European Journal of Human Genetics, 2015, 23, 1652-1656.	1.4	30
129	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	2.6	30
130	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356.	2.6	30
131	Non-syndromic Tooth Agenesis Associated with a Nonsense Mutation in Ectodysplasin-A (EDA). Journal of Dental Research, 2013, 92, 507-511.	2.5	29
132	LRP5 variants may contribute to ADPKD. European Journal of Human Genetics, 2016, 24, 237-242.	1.4	28
133	De novo mutations in MSL3 cause an X-linked syndrome marked by impaired histone H4 lysine 16 acetylation. Nature Genetics, 2018, 50, 1442-1451.	9.4	28
134	Candidate Genes for Nonsyndromic Cleft Palate Detected by Exome Sequencing. Journal of Dental Research, 2017, 96, 1314-1321.	2.5	27
135	Long-read trio sequencing of individuals with unsolved intellectual disability. European Journal of Human Genetics, 2021, 29, 637-648.	1.4	27
136	Whole genome sequencing and in vitro splice assays reveal genetic causes for inherited retinal diseases. Npj Genomic Medicine, 2021, 6, 97.	1.7	27
137	Exome sequencing identifies a novel and a recurrent BBS1 mutation in Pakistani families with Bardet-Biedl syndrome. Molecular Vision, 2013, 19, 644-53.	1.1	26
138	A compound heterozygous mutation in DPAGT1 results in a congenital disorder of glycosylation with a relatively mild phenotype. European Journal of Human Genetics, 2013, 21, 844-849.	1.4	25
139	Late-Onset Stargardt Disease Due to Mild, Deep-Intronic ABCA4 Alleles. , 2019, 60, 4249.		25
140	Truncating de novo mutations in the KrÃ¼ppel-type zinc-finger gene ZNF148 in patients with corpus callosum defects, developmental delay, short stature, and dysmorphisms. Genome Medicine, 2016, 8, 131.	3.6	24
141	Exome Sequencing Identifies Three Novel Candidate Genes Implicated in Intellectual Disability. PLoS ONE, 2014, 9, e112687.	1.1	23
142	Diagnostic Exome Sequencing in Persons With Severe Intellectual Disability. Obstetrical and Gynecological Survey, 2013, 68, 191-193.	0.2	22
143	Systematic analysis of short tandem repeats in 38,095 exomes provides an additional diagnostic yield. Genetics in Medicine, 2021, 23, 1569-1573.	1.1	21
144	Diagnostic exome-based preconception carrier testing in consanguineous couples: results from the first 100 couples in clinical practice. Genetics in Medicine, 2021, 23, 1125-1136.	1.1	20

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145	Clinical exome sequencingâ€"Mistakes and caveats. Human Mutation, 2022, 43, 1041-1055.	1.1	20
146	High density gene expression microarrays and gene ontology analysis for identifying processes in implanted tissue engineering constructs. Biomaterials, 2010, 31, 8299-8312.	5.7	19
147	Analysis of genes regulated by the transcription factor LUMAN identifies ApoA4 as a target gene in dendritic cells. Molecular Immunology, 2012, 50, 66-73.	1.0	18
148	MST1R mutation as a genetic cause of Lady Windermere syndrome. European Respiratory Journal, 2017, 49, 1601478.	3.1	18
149	Genome-wide investigation of an ID cohort reveals de novo 3â€²UTR variants affecting gene expression. Human Genetics, 2018, 137, 717-721.	1.8	18
150	Cochlear supporting cells require GAS2 for cytoskeletal architecture and hearing. Developmental Cell, 2021, 56, 1526-1540.e7.	3.1	18
151	The effect of enamel matrix derivative (EmdogainÂ®) on gene expression profiles of human primary alveolar bone cells. Journal of Tissue Engineering and Regenerative Medicine, 2014, 8, 463-472.	1.3	17
152	Missense variants in AIMP1 gene are implicated in autosomal recessive intellectual disability without neurodegeneration. European Journal of Human Genetics, 2016, 24, 392-399.	1.4	17
153	Genetic Spectrum of ABCA4-Associated Retinal Degeneration in Poland. Genes, 2019, 10, 959.	1.0	17
154	Variants affecting diverse domains of MEPE are associated with two distinct bone disorders, a craniofacial bone defect and otosclerosis. Genetics in Medicine, 2019, 21, 1199-1208.	1.1	17
155	Reanalysis of exome negative patients with rare disease: a pragmatic workflow for diagnostic applications. Genome Medicine, 2022, 14, .	3.6	17
156	Cord Blood Mesenchymal Stem Cells Suppress DC-T Cell Proliferation via Prostaglandin B2. Stem Cells and Development, 2014, 23, 1582-1593.	1.1	16
157	Apoptosis-Related Gene Expression Profiling in Hematopoietic Cell Fractions of MDS Patients. PLoS ONE, 2016, 11, e0165582.	1.1	16
158	Deletions and loss-of-function variants in TP63 associated with orofacial clefting. European Journal of Human Genetics, 2019, 27, 1101-1112.	1.4	16
159	Overarching control of autophagy and DNA damage response by CHD6 revealed by modeling a rare human pathology. Nature Communications, 2021, 12, 3014.	5.8	16
160	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	1.1	16
161	Reliable noninvasive prenatal testing by massively parallel sequencing of circulating cell-free DNA from maternal plasma processed up to 24h after venipuncture. Clinical Biochemistry, 2013, 46, 1783-1786.	0.8	15
162	Identification of C12orf4 as a gene for autosomal recessive intellectual disability. Clinical Genetics, 2017, 91, 100-105.	1.0	15

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163	Upstream SLC2A1 translation initiation causes GLUT1 deficiency syndrome. European Journal of Human Genetics, 2017, 25, 771-774.	1.4	15
164	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.	1.1	15
165	Different Balance of Wnt Signaling in Adult and Fetal Bone Marrow-Derived Mesenchymal Stromal Cells. Stem Cells and Development, 2016, 25, 934-947.	1.1	14
166	Novel Compound Heterozygous Mutation in TRAPPC9 Gene: The Relevance of Whole Genome Sequencing. Genes, 2021, 12, 557.	1.0	14
167	Missense variants in NOX1 and p22phox in a case of very-early-onset inflammatory bowel disease are functionally linked to NOD2. Journal of Physical Education and Sports Management, 2019, 5, a002428.	0.5	13
168	BBS1 branchpoint variant is associated with non-syndromic retinitis pigmentosa. Journal of Medical Genetics, 2022, 59, 438-444.	1.5	13
169	Rare NOX3 Variants Confer Susceptibility to Agranulocytosis During Thyrostatic Treatment of Graves' Disease. Clinical Pharmacology and Therapeutics, 2017, 102, 1017-1024.	2.3	12
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