Alexander Hoischen

List of Publications by Year
in descending order

176 papers	18,103 citations	13087 68 h-index	15249 126 g-index
201 all docs	201 docs citations	201 times ranked	28790 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	Refining analyses of copy number variation identifies specific genes associated with developmental delay. Nature Genetics, 2014, 46, 1063-1071.	9.4	583
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	New insights into the generation and role of de novo mutations in health and disease. Genome Biology, 2016, 17, 241.	3.8	339
10	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
11	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
12	Long-Read Sequencing Emerging in Medical Genetics. Frontiers in Genetics, 2019, 10, 426.	1.1	290
13	Parent-of-origin-specific signatures of de novo mutations. Nature Genetics, 2016, 48, 935-939.	9.4	266
14	Exome Sequencing Identifies WDR35 Variants Involved in Sensenbrenner Syndrome. American Journal of Human Genetics, 2010, 87, 418-423.	2.6	260
15	Nextâ€generation genetic testing for retinitis pigmentosa. Human Mutation, 2012, 33, 963-972.	1.1	258
16	De novo mutations in the actin genes ACTB and ACTG1 cause Baraitser-Winter syndrome. Nature Genetics, 2012, 44, 440-444.	9.4	237
17	De novo nonsense mutations in ASXL1 cause Bohring-Opitz syndrome. Nature Genetics, 2011, 43, 729-731.	9.4	236
18	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

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19	Unlocking Mendelian disease using exome sequencing. Genome Biology, 2011, 12, 228.	13.9	228
20	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
21	Human TLR10 is an anti-inflammatory pattern-recognition receptor. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, E4478-84.	3.3	211
22	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
23	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
24	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
25	Mutations in GDP-Mannose Pyrophosphorylase B Cause Congenital and Limb-Girdle Muscular Dystrophies Associated with Hypoglycosylation of Î±-Dystroglycan. American Journal of Human Genetics, 2013, 93, 29-41.	2.6	197
26	Gain-of-function mutations in the mechanically activated ion channel PIEZO2 cause a subtype of Distal Arthrogryposis. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 4667-4672.	3.3	193
27	Coffin-Siris Syndrome and the BAF Complex: Genotype-Phenotype Study in 63 Patients. Human Mutation, 2013, 34, 1519-1528.	1.1	178
28	Genome-Wide Profiling of p63 DNAâ€“Binding Sites Identifies an Element that Regulates Gene Expression during Limb Development in the 7q21 SHFM1 Locus. PLoS Genetics, 2010, 6, e1001065.	1.5	169
29	Functional genomics identifies type I interferon pathway as central for host defense against Candida albicans. Nature Communications, 2013, 4, 1342.	5.8	157
30	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
31	Prioritization of neurodevelopmental disease genes by discovery of new mutations. Nature Neuroscience, 2014, 17, 764-772.	7.1	148
32	Gene identification in the congenital disorders of glycosylation type I by whole-exome sequencing. Human Molecular Genetics, 2012, 21, 4151-4161.	1.4	147
33	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
34	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
35	CantÃº Syndrome Is Caused by Mutations in ABCC9. American Journal of Human Genetics, 2012, 90, 1094-1101.	2.6	141
36	Severe mental retardation with breathing abnormalities (Pittâ€“Hopkins syndrome) is caused by haploinsufficiency of the neuronal bHLH transcription factor TCF4. Human Molecular Genetics, 2007, 16, 1488-1494.	1.4	137

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37	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. American Journal of Human Genetics, 2016, 98, 541-552.	2.6	132
38	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
39	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
40	Heterozygous Loss-of-Function SEC61A1 Mutations Cause Autosomal-Dominant Tubulo-Interstitial and Glomerulocystic Kidney Disease with Anemia. American Journal of Human Genetics, 2016, 99, 174-187.	2.6	124
41	Baraitserâ€“Winter cerebrofrontofacial syndrome: delineation of the spectrum in 42 cases. European Journal of Human Genetics, 2015, 23, 292-301.	1.4	115
42	Angiocentric Glioma. American Journal of Surgical Pathology, 2007, 31, 1709-1718.	2.1	110
43	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
44	DVL1 Frameshift Mutations Clustering in the Penultimate Exon Cause Autosomal-Dominant Robinow Syndrome. American Journal of Human Genetics, 2015, 96, 612-622.	2.6	110
45	ATP6AP1 deficiency causes an immunodeficiency with hepatopathy, cognitive impairment and abnormal protein glycosylation. Nature Communications, 2016, 7, 11600.	5.8	110
46	Optical genome mapping enables constitutional chromosomal aberration detection. American Journal of Human Genetics, 2021, 108, 1409-1422.	2.6	108
47	Adult-onset autoinflammation caused by somatic mutations in UBA1: AÂDutch case series of patients with VEXAS. Journal of Allergy and Clinical Immunology, 2022, 149, 432-439.e4.	1.5	105
48	STAT1 Hyperphosphorylation and Defective IL12R/IL23R Signaling Underlie Defective Immunity in Autosomal Dominant Chronic Mucocutaneous Candidiasis. PLoS ONE, 2011, 6, e29248.	1.1	101
49	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
50	Frequent loss of chromosome 9, homozygous CDKN2A/p14ARF/CDKN2B deletion and low TSC1 mRNA expression in pleomorphic xanthoastrocytomas. Oncogene, 2007, 26, 1088-1097.	2.6	98
51	Intragenic deletion in DYRK1A leads to mental retardation and primary microcephaly. Clinical Genetics, 2011, 79, 296-299.	1.0	94
52	Reliable Next-Generation Sequencing of Formalin-Fixed, Paraffin-Embedded Tissue Using Single Molecule Tags. Journal of Molecular Diagnostics, 2016, 18, 851-863.	1.2	94
53	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
54	Trisomy for Synaptojanin1 in Down syndrome is functionally linked to the enlargement of early endosomes. Human Molecular Genetics, 2012, 21, 3156-3172.	1.4	92

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55	CantÃº Syndrome Resulting from Activating Mutation in theKCNJ8Gene. Human Mutation, 2014, 35, 809-813.	1.1	92
56	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
57	B56Î´-related protein phosphatase 2A dysfunction identified in patients with intellectual disability. Journal of Clinical Investigation, 2015, 125, 3051-3062.	3.9	91
58	CCDC115 Deficiency Causes a Disorder of Golgi Homeostasis with Abnormal Protein Glycosylation. American Journal of Human Genetics, 2016, 98, 310-321.	2.6	88
59	DVL3 Alleles Resulting in a â˜1 Frameshift of the Last Exon Mediate Autosomal-Dominant Robinow Syndrome. American Journal of Human Genetics, 2016, 98, 553-561.	2.6	88
60	WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. American Journal of Human Genetics, 2018, 102, 27-43.	2.6	88
61	Massively parallel sequencing of ataxia genes after array-based enrichment. Human Mutation, 2010, 31, 494-499.	1.1	86
62	<scp>MLL2</scp> mutation detection in 86 patients with Kabuki syndrome: a genotypeâ€“phenotype study. Clinical Genetics, 2013, 84, 539-545.	1.0	85
63	Next-generation cytogenetics: Comprehensive assessment of 52 hematological malignancy genomes by optical genome mapping. American Journal of Human Genetics, 2021, 108, 1423-1435.	2.6	85
64	EPHB4 kinaseâ€“inactivating mutations cause autosomal dominant lymphatic-related hydrops fetalis. Journal of Clinical Investigation, 2016, 126, 3080-3088.	3.9	83
65	Mutations in ATP6V1E1 or ATP6V1A Cause Autosomal-Recessive Cutis Laxa. American Journal of Human Genetics, 2017, 100, 216-227.	2.6	82
66	A complex V ATP5A1 defect causes fatal neonatal mitochondrial encephalopathy. Brain, 2013, 136, 1544-1554.	3.7	80
67	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
68	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
69	Next-generation DNA sequencing identifies novel gene variants and pathways involved in specific language impairment. Scientific Reports, 2017, 7, 46105.	1.6	79
70	Mutations in MED12 Cause X-Linked Ohdo Syndrome. American Journal of Human Genetics, 2013, 92, 401-406.	2.6	78
71	De novo mutations in PLXND1 and REV3L cause MÃ¶bius syndrome. Nature Communications, 2015, 6, 7199.	5.8	76
72	Catecholamines Induce Trained Immunity in Monocytes In Vitro and In Vivo. Circulation Research, 2020, 127, 269-283.	2.0	76

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73	Genetic Screening for TLR7 Variants in Young and Previously Healthy Men With Severe COVID-19. Frontiers in Immunology, 2021, 12, 719115.	2.2	76
74	Transcriptional and functional insights into the host immune response against the emerging fungal pathogen Candida auris. Nature Microbiology, 2020, 5, 1516-1531.	5.9	75
75	Mutations in ANTXR1 Cause GAPO Syndrome. American Journal of Human Genetics, 2013, 92, 792-799.	2.6	73
76	TMEM199 Deficiency Is a Disorder of Golgi Homeostasis Characterized by Elevated Aminotransferases, Alkaline Phosphatase, and Cholesterol and Abnormal Glycosylation. American Journal of Human Genetics, 2016, 98, 322-330.	2.6	73
77	Exome sequencing identifies putative drivers of progression of transient myeloproliferative disorder to AMKL in infants with Down syndrome. Blood, 2013, 122, 554-561.	0.6	72
78	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
79	De Novo Truncating Variants in ASXL2 Are Associated with a Unique and Recognizable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 991-999.	2.6	68
80	Deleterious Germline BLM Mutations and the Risk for Early-onset Colorectal Cancer. Scientific Reports, 2015, 5, 14060.	1.6	67
81	Recurrent Mutations in the Basic Domain of TWIST2 Cause Ablepharon Macrostomia and Barber-Say Syndromes. American Journal of Human Genetics, 2015, 97, 99-110.	2.6	61
82	Bi-allelic Mutations in the Mitochondrial Ribosomal Protein MRPS2 Cause Sensorineural Hearing Loss, Hypoglycemia, and Multiple OXPHOS Complex Deficiencies. American Journal of Human Genetics, 2018, 102, 685-695.	2.6	61
83	Mapping candidate regions and genes for congenital anomalies of the kidneys and urinary tract (CAKUT) by array-based comparative genomic hybridization. Nephrology Dialysis Transplantation, 2011, 26, 136-143.	0.4	60
84	The phenotype of Floating-Harbor syndrome: clinical characterization of 52 individuals with mutations in exon 34 of SRCAP. Orphanet Journal of Rare Diseases, 2013, 8, 63.	1.2	60
85	Mutations Affecting the SAND Domain of DEAF1 Cause Intellectual Disability with Severe Speech Impairment and Behavioral Problems. American Journal of Human Genetics, 2014, 94, 649-661.	2.6	59
86	Comprehensive Characterization of Genomic Aberrations in Gangliogliomas by CGH, Arrayâ€based CGH and Interphase FISH. Brain Pathology, 2008, 18, 326-337.	2.1	58
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	NovelBRCA1andBRCA2Tumor Test as Basis for Treatment Decisions and Referral for Genetic Counselling of Patients with Ovarian Carcinomas. Human Mutation, 2017, 38, 226-235.	1.1	55
90	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

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91	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
92	Exome sequencing in routine diagnostics: a generic test for 254 patients with primary immunodeficiencies. Genome Medicine, 2019, 11, 38.	3.6	49
93	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
94	KIAA1797/FOCAD encodes a novel focal adhesion protein with tumour suppressor function in gliomas. Brain, 2012, 135, 1027-1041.	3.7	47
95	Clinical Validation of Whole Genome Sequencing for Cancer Diagnostics. Journal of Molecular Diagnostics, 2021, 23, 816-833.	1.2	47
96	Trisomic dose of several chromosome 21 genes perturbs haematopoietic stem and progenitor cell differentiation in Down's syndrome. Oncogene, 2010, 29, 6102-6114.	2.6	46
97	BRCA Testing by Single-Molecule Molecular Inversion Probes. Clinical Chemistry, 2017, 63, 503-512.	1.5	46
98	A de novo non-sense mutation in ZBTB18 in a patient with features of the 1q43q44 microdeletion syndrome. European Journal of Human Genetics, 2014, 22, 844-846.	1.4	45
99	Refinement of the critical 2p25.3 deletion region: the role of MYT1L in intellectual disability and obesity. Genetics in Medicine, 2015, 17, 460-466.	1.1	45
100	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
101	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
102	Towards mapping phenotypical traits in 18pâ˜ syndrome by array-based comparative genomic hybridisation and fluorescent in situ hybridisation. European Journal of Human Genetics, 2007, 15, 35-44.	1.4	42
103	Shotgun metagenomic data reveals significant abundance but low diversity of â€œCandidatus Scalinduaâ€• marine anammox bacteria in the Arabian Sea oxygen minimum zone. Frontiers in Microbiology, 2014, 5, 31.	1.5	41
104	Diagnostic serum glycosylation profile in patients with intellectual disability as a result of MAN1B1 deficiency. Brain, 2014, 137, 1030-1038.	3.7	41
105	Whole-genome sequencing of spermatocytic tumors provides insights into the mutational processes operating in the male germline. PLoS ONE, 2017, 12, e0178169.	1.1	36
106	Overlapping SETBP1 gain-of-function mutations in Schinzel-Giedion syndrome and hematologic malignancies. PLoS Genetics, 2017, 13, e1006683.	1.5	35
107	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
108	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

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109	Next-generation phenotyping using computer vision algorithms in rare genomic neurodevelopmental disorders. Genetics in Medicine, 2019, 21, 1719-1725.	1.1	34
110	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. European Journal of Human Genetics, 2021, 29, 1337-1347.	1.4	34
111	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
112	Confirmation of the role of pathogenic SMAD6 variants in bicuspid aortic valve-related aortopathy. European Journal of Human Genetics, 2019, 27, 1044-1053.	1.4	32
113	Familial long-read sequencing increases yield of de novo mutations. American Journal of Human Genetics, 2022, 109, 631-646.	2.6	32
114	A novel marine nitrite-oxidizing Nitrospira species from Dutch coastal North Sea water. Frontiers in Microbiology, 2013, 4, 60.	1.5	30
115	Syndromic X-linked intellectual disability segregating with a missense variant in RLIM. European Journal of Human Genetics, 2015, 23, 1652-1656.	1.4	30
116	LRP5 variants may contribute to ADPKD. European Journal of Human Genetics, 2016, 24, 237-242.	1.4	28
117	Arterial Wall Inflammation and Increased Hematopoietic Activity in Patients With Primary Aldosteronism. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e1967-e1980.	1.8	27
118	Long-read trio sequencing of individuals with unsolved intellectual disability. European Journal of Human Genetics, 2021, 29, 637-648.	1.4	27
119	Homozygous mutation of STXBP5L explains an autosomal recessive infantile-onset neurodegenerative disorder. Human Molecular Genetics, 2015, 24, 2000-2010.	1.4	25
120	Optical genome mapping identifies a germline retrotransposon insertion in <scp>SMARCB1</scp> in two siblings with atypical teratoid rhabdoid tumors. Journal of Pathology, 2021, 255, 202-211.	2.1	23
121	Further case of CantÃº syndrome: Exclusion of cryptic subtelomeric chromosome aberrations. American Journal of Medical Genetics Part A, 2002, 111, 205-209.	2.4	22
122	Diagnostic Exome Sequencing in Persons With Severe Intellectual Disability. Obstetrical and Gynecological Survey, 2013, 68, 191-193.	0.2	22
123	Progressive multifocal leukoencephalopathy in an immunocompetent patient. Annals of Clinical and Translational Neurology, 2016, 3, 226-232.	1.7	19
124	Whole exome sequencing and arrayâ€based molecular karyotyping as aids to prenatal diagnosis in fetuses with suspected Simpsonâ€“Golabiâ€“Behmel syndrome. Prenatal Diagnosis, 2016, 36, 961-965.	1.1	19
125	Quantification of differential gene expression by multiplexed targeted resequencing of cDNA. Nature Communications, 2017, 8, 15190.	5.8	19
126	Identification of Restless Legs Syndrome Genes by Mutational Load Analysis. Annals of Neurology, 2020, 87, 184-193.	2.8	19

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127	MST1R mutation as a genetic cause of Lady Windermere syndrome. European Respiratory Journal, 2017, 49, 1601478.	3.1	18
128	Genome of PeÅŸtera Muierii skull shows high diversity and low mutational load in pre-glacial Europe. Current Biology, 2021, 31, 2973-2983.e9.	1.8	18
129	Optical Genome Mapping: A Promising New Tool to Assess Genomic Complexity in Chronic Lymphocytic Leukemia (CLL). Cancers, 2022, 14, 3376.	1.7	18
130	Genetic Spectrum of ABCA4-Associated Retinal Degeneration in Poland. Genes, 2019, 10, 959.	1.0	17
131	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
132	A Genetics-First Approach Revealed Monogenic Disorders in Patients With ARM and VACTERL Anomalies. Frontiers in Pediatrics, 2020, 8, 310.	0.9	17
133	Next-Generation Sequencing in the Field of Primary Immunodeficiencies: Current Yield, Challenges, and Future Perspectives. Clinical Reviews in Allergy and Immunology, 2021, 61, 212-225.	2.9	17
134	Clonal Hematopoiesis Is Associated With Low CD4 Nadir and Increased Residual HIV Transcriptional Activity in Virally Suppressed Individuals With HIV. Journal of Infectious Diseases, 2022, 225, 1339-1347.	1.9	17
135	Deletions and loss-of-function variants in TP63 associated with orofacial clefting. European Journal of Human Genetics, 2019, 27, 1101-1112.	1.4	16
136	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
137	BDNF and DYRK1A Are Variable and Inversely Correlated in Lymphoblastoid Cell Lines from Down Syndrome Patients. Molecular Neurobiology, 2012, 46, 297-303.	1.9	15
138	Expanding the clinical spectrum of recessive truncating mutations ofKLHL7to a Bohring-Opitz-like phenotype. Journal of Medical Genetics, 2017, 54, 830-835.	1.5	15
139	Optical genome mapping identifies rare structural variations as predisposition factors associated with severe COVID-19. IScience, 2022, 25, 103760.	1.9	15
140	A phenotype map for 14q32.3 terminal deletions. American Journal of Medical Genetics, Part A, 2012, 158A, 695-706.	0.7	14
141	Chromosomal abnormalities in hepatic cysts point to novel polycystic liver disease genes. European Journal of Human Genetics, 2016, 24, 1707-1714.	1.4	14
142	Postzygotic mosaicism in cerebral cavernous malformation. Journal of Medical Genetics, 2020, 57, 212-216.	1.5	13
143	Genome-wide analysis for micro-aberrations in familial exstrophy of the bladder using array-based comparative genomic hybridization. BJU International, 2007, 100, 646-650.	1.3	12
144	A molecular inversion probe-based next-generation sequencing panel to detect germline mutations in Chinese early-onset colorectal cancer patients. Oncotarget, 2017, 8, 24533-24547.	0.8	12

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145	Insufficient evidence for a role of SERPINF1 in otosclerosis. Molecular Genetics and Genomics, 2019, 294, 1001-1006.	1.0	11
146	Novel GANAB variants associated with polycystic liver disease. Orphanet Journal of Rare Diseases, 2020, 15, 302.	1.2	11
147	Rosetted glioneuronal tumor of the spine with overtly anaplastic histological features. Acta Neuropathologica, 2009, 117, 591-593.	3.9	10
148	Amplified segment in the â€˜Down Syndrome critical regionâ€™ on HSA21 shared between Down syndrome and euploid AML-M0 excludes RUNX1, ERG and ETS2. British Journal of Haematology, 2012, 157, 197-200.	1.2	10
149	Primary immunodeficiencies in cytosolic patternâ€recognition receptor pathways: Toward hostâ€directed treatment strategies. Immunological Reviews, 2020, 297, 247-272.	2.8	10
150	A mosaic PIK3CA variant in a young adult with diffuse gastric cancer: case report. European Journal of Human Genetics, 2021, 29, 1354-1358.	1.4	9
151	Resequencing of candidate genes for Keratoconus reveals a role for Ehlersâ€“Danlos Syndrome genes. European Journal of Human Genetics, 2021, 29, 1745-1755.	1.4	8
152	Pro-inflammatory Monocyte Phenotype During Acute Progression of Cerebral Small Vessel Disease. Frontiers in Cardiovascular Medicine, 2021, 8, 639361.	1.1	8
153	Solving the unsolved rare diseases in Europe. European Journal of Human Genetics, 2021, 29, 1319-1320.	1.4	8
154	Array-CGH in unclear syndromic nephropathies identifies a microdeletion in Xq22.3-q23. Pediatric Nephrology, 2009, 24, 1673-1681.	0.9	7
155	Early presentation of cystic kidneys in a family with a homozygous INVS mutation. American Journal of Medical Genetics, Part A, 2014, 164, 1627-1634.	0.7	7
156	RareVariantVis: new tool for visualization of causative variants in rare monogenic disorders using whole genome sequencing data. Bioinformatics, 2016, 32, 3018-3020.	1.8	7
157	Novel defect in phosphatidylinositol 4â€kinase type 2â€alpha (<scp>PI4K2A</scp>) at the membraneâ€enzyme interface is associated with metabolic cutis laxa. Journal of Inherited Metabolic Disease, 2020, 43, 1382-1391.	1.7	7
158	A MT-TL1 variant identified by whole exome sequencing in an individual with intellectual disability, epilepsy, and spastic tetraparesis. European Journal of Human Genetics, 2021, 29, 1359-1368.	1.4	7
159	oxLDL-Induced Trained Immunity Is Dependent on Mitochondrial Metabolic Reprogramming. Immunometabolism, 2021, 3, e210025.	6.0	7
160	Immunologic defects in severe mucocutaneous HSV-2 infections: Response to IFN-Î³ therapy. Journal of Allergy and Clinical Immunology, 2016, 138, 895-898.	1.5	6
161	A wide range of protective and predisposing variants in aggrecan influence the susceptibility for otosclerosis. Human Genetics, 2022, 141, 951-963.	1.8	6
162	National external quality assessment for next-generation sequencing-based diagnostics of primary immunodeficiencies. European Journal of Human Genetics, 2021, 29, 20-28.	1.4	5

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163	Impact of rare and common genetic variation in the interleukin-1 pathway on human cytokine responses. Genome Medicine, 2021, 13, 94.	3.6	5
164	Circulating interleukin-37 declines with aging in healthy humans: relations to healthspan indicators and IL37 gene SNPs. GeroScience, 0, , .	2.1	5
165	Accurate detection of low-level mosaic mutations in pediatric acute lymphoblastic leukemia using single molecule tagging and deep-sequencing. Leukemia and Lymphoma, 2018, 59, 1690-1699.	0.6	4
166	Overrepresentation of genetic variation in the AnkyrinG interactome is related to a range of neurodevelopmental disorders. European Journal of Human Genetics, 2020, 28, 1726-1733.	1.4	4
167	Solving unsolved rare neurological diseasesâ€"a Solve-RD viewpoint. European Journal of Human Genetics, 2021, 29, 1332-1336.	1.4	4
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