Alexander Hoischen

List of Publications by Year
in descending order

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176 papers	18,103 citations	13099 68 h-index	15266 126 g-index
201 all docs	201 docs citations	201 times ranked	28790 citing authors

#	Article	IF	Citations
1	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.	2.9	105
2	A wide range of protective and predisposing variants in aggrecan influence the susceptibility for otosclerosis. Human Genetics, 2022, 141, 951-963.	3.8	6
3	Exploring the missing heritability in subjects with hearing loss, enlarged vestibular aqueducts, and a single or no pathogenic SLC26A4 variant. Human Genetics, 2022, 141, 465-484.	3.8	3
4	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.	4.0	17
5	The broader phenotypic spectrum of congenital caudal abnormalities associated with mutations in the caudal type homeobox 2 gene. Clinical Genetics, 2022, 101, 183-189.	2.0	4
6	Optical genome mapping identifies rare structural variations as predisposition factors associated with severe COVID-19. IScience, 2022, 25, 103760.	4.1	15
7	Familial long-read sequencing increases yield of de novo mutations. American Journal of Human Genetics, 2022, 109, 631-646.	6.2	32
8	Solving the genetic aetiology of hereditary gastrointestinal tumour syndromesâ€“ a collaborative multicentre endeavour within the project Solve-RD. European Journal of Medical Genetics, 2022, 65, 104475.	1.3	2
9	Optical Genome Mapping: A Promising New Tool to Assess Genomic Complexity in Chronic Lymphocytic Leukemia (CLL). Cancers, 2022, 14, 3376.	3.7	18
10	National external quality assessment for next-generation sequencing-based diagnostics of primary immunodeficiencies. European Journal of Human Genetics, 2021, 29, 20-28.	2.8	5
11	Long-read trio sequencing of individuals with unsolved intellectual disability. European Journal of Human Genetics, 2021, 29, 637-648.	2.8	27
12	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.	6.5	17
13	Resequencing of candidate genes for Keratoconus reveals a role for Ehlersâ€“Danlos Syndrome genes. European Journal of Human Genetics, 2021, 29, 1745-1755.	2.8	8
14	Impact of rare and common genetic variation in the interleukin-1 pathway on human cytokine responses. Genome Medicine, 2021, 13, 94.	8.2	5
15	Solving unsolved rare neurological diseasesâ€”a Solve-RD viewpoint. European Journal of Human Genetics, 2021, 29, 1332-1336.	2.8	4
16	Pro-inflammatory Monocyte Phenotype During Acute Progression of Cerebral Small Vessel Disease. Frontiers in Cardiovascular Medicine, 2021, 8, 639361.	2.4	8
17	Overarching control of autophagy and DNA damage response by CHD6 revealed by modeling a rare human pathology. Nature Communications, 2021, 12, 3014.	12.8	16
18	Solving the unsolved rare diseases in Europe. European Journal of Human Genetics, 2021, 29, 1319-1320.	2.8	8

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19	Extending the allelic spectrum at noncoding risk loci of orofacial clefting. Human Mutation, 2021, 42, 1066-1078.	2.5	3
20	A mosaic PIK3CA variant in a young adult with diffuse gastric cancer: case report. European Journal of Human Genetics, 2021, 29, 1354-1358.	2.8	9
21	Solve-RD: systematic pan-European data sharing and collaborative analysis to solve rare diseases. European Journal of Human Genetics, 2021, 29, 1325-1331.	2.8	49
22	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.	2.8	7
23	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. European Journal of Human Genetics, 2021, 29, 1337-1347.	2.8	34
24	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.	4.5	23
25	Genome of PeÅŸtera Muierii skull shows high diversity and low mutational load in pre-glacial Europe. Current Biology, 2021, 31, 2973-2983.e9.	3.9	18
26	Genetic Screening for TLR7 Variants in Young and Previously Healthy Men With Severe COVID-19. Frontiers in Immunology, 2021, 12, 719115.	4.8	76
27	Clinical Validation of Whole Genome Sequencing for Cancer Diagnostics. Journal of Molecular Diagnostics, 2021, 23, 816-833.	2.8	47
28	Optical genome mapping enables constitutional chromosomal aberration detection. American Journal of Human Genetics, 2021, 108, 1409-1422.	6.2	108
29	Next-generation cytogenetics: Comprehensive assessment of 52 hematological malignancy genomes by optical genome mapping. American Journal of Human Genetics, 2021, 108, 1423-1435.	6.2	85
30	Long-read technologies identify a hidden inverted duplication in a family with choroideremia. Human Genetics and Genomics Advances, 2021, 2, 100046.	1.7	4
31	oxLDL-Induced Trained Immunity Is Dependent on Mitochondrial Metabolic Reprogramming. Immunometabolism, 2021, 3, e210025.	6.0	7
32	Identification of Restless Legs Syndrome Genes by Mutational Load Analysis. Annals of Neurology, 2020, 87, 184-193.	5.3	19
33	Arterial Wall Inflammation and Increased Hematopoietic Activity in Patients With Primary Aldosteronism. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e1967-e1980.	3.6	27
34	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.	2.8	4
35	Presence of Genetic Variants Among Young Men With Severe COVID-19. JAMA - Journal of the American Medical Association, 2020, 324, 663.	7.4	626
36	Novel GANAB variants associated with polycystic liver disease. Orphanet Journal of Rare Diseases, 2020, 15, 302.	2.7	11

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37	Transcriptional and functional insights into the host immune response against the emerging fungal pathogen Candida auris. Nature Microbiology, 2020, 5, 1516-1531.	13.3	75
38	Resolving the dark matter of ABCA4 for 1054 Stargardt disease probands through integrated genomics and transcriptomics. Genetics in Medicine, 2020, 22, 1235-1246.	2.4	92
39	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.	3.6	7
40	Postzygotic mosaicism in cerebral cavernous malformation. Journal of Medical Genetics, 2020, 57, 212-216.	3.2	13
41	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.	0.9	44
42	A Genetics-First Approach Revealed Monogenic Disorders in Patients With ARM and VACTERL Anomalies. Frontiers in Pediatrics, 2020, 8, 310.	1.9	17
43	Primary immunodeficiencies in cytosolic patternâ€recognition receptor pathways: Toward hostâ€directed treatment strategies. Immunological Reviews, 2020, 297, 247-272.	6.0	10
44	Catecholamines Induce Trained Immunity in Monocytes In Vitro and In Vivo. Circulation Research, 2020, 127, 269-283.	4.5	76
45	Exome sequencing in routine diagnostics: a generic test for 254 patients with primary immunodeficiencies. Genome Medicine, 2019, 11, 38.	8.2	49
46	Long-Read Sequencing Emerging in Medical Genetics. Frontiers in Genetics, 2019, 10, 426.	2.3	290
47	Deletions and loss-of-function variants in TP63 associated with orofacial clefting. European Journal of Human Genetics, 2019, 27, 1101-1112.	2.8	16
48	Insufficient evidence for a role of SERPINF1 in otosclerosis. Molecular Genetics and Genomics, 2019, 294, 1001-1006.	2.1	11
49	Confirmation of the role of pathogenic SMAD6 variants in bicuspid aortic valve-related aortopathy. European Journal of Human Genetics, 2019, 27, 1044-1053.	2.8	32
50	Genetic Spectrum of ABCA4-Associated Retinal Degeneration in Poland. Genes, 2019, 10, 959.	2.4	17
51	Single Molecule Molecular Inversion Probes for High Throughput Germline Screenings in Dystonia. Frontiers in Neurology, 2019, 10, 1332.	2.4	2
52	Next-generation phenotyping using computer vision algorithms in rare genomic neurodevelopmental disorders. Genetics in Medicine, 2019, 21, 1719-1725.	2.4	34
53	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.	2.4	17
54	WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. American Journal of Human Genetics, 2018, 102, 27-43.	6.2	88

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55	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.	6.2	61
56	A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency. European Journal of Human Genetics, 2018, 26, 54-63.	2.8	32
57	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.	1.3	4
58	Somatic variants in autosomal dominant genes are a rare cause of sporadic Alzheimer's disease. Alzheimer's and Dementia, 2018, 14, 1632-1639.	0.8	51
59	Mutations in ATP6V1E1 or ATP6V1A Cause Autosomal-Recessive Cutis Laxa. American Journal of Human Genetics, 2017, 100, 216-227.	6.2	82
60	BRCA Testing by Single-Molecule Molecular Inversion Probes. Clinical Chemistry, 2017, 63, 503-512.	3.2	46
61	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.	6.2	156
62	Next-generation DNA sequencing identifies novel gene variants and pathways involved in specific language impairment. Scientific Reports, 2017, 7, 46105.	3.3	79
63	MST1R mutation as a genetic cause of Lady Windermere syndrome. European Respiratory Journal, 2017, 49, 1601478.	6.7	18
64	Expanding the clinical spectrum of recessive truncating mutations ofKLHL7to a Bohring-Opitz-like phenotype. Journal of Medical Genetics, 2017, 54, 830-835.	3.2	15
65	Unraveling genetic predisposition to familial or early onset gastric cancer using germline whole-exome sequencing. European Journal of Human Genetics, 2017, 25, 1246-1252.	2.8	34
66	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.	2.5	45
67	Quantification of differential gene expression by multiplexed targeted resequencing of cDNA. Nature Communications, 2017, 8, 15190.	12.8	19
68	Ultra-sensitive Sequencing Identifies High Prevalence of Clonal Hematopoiesis-Associated Mutations throughout Adult Life. American Journal of Human Genetics, 2017, 101, 50-64.	6.2	210
69	NovelBRCA1andBRCA2Tumor Test as Basis for Treatment Decisions and Referral for Genetic Counselling of Patients with Ovarian Carcinomas. Human Mutation, 2017, 38, 226-235.	2.5	55
70	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.	1.8	12
71	Overlapping SETBP1 gain-of-function mutations in Schinzel-Giedion syndrome and hematologic malignancies. PLoS Genetics, 2017, 13, e1006683.	3.5	35
72	Whole-genome sequencing of spermatocytic tumors provides insights into the mutational processes operating in the male germline. PLoS ONE, 2017, 12, e0178169.	2.5	36

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73	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.	6.2	124
74	New insights into the generation and role of de novo mutations in health and disease. Genome Biology, 2016, 17, 241.	8.8	339
75	Immunologic defects in severe mucocutaneous HSV-2 infections: Response to IFN-Î³ therapy. Journal of Allergy and Clinical Immunology, 2016, 138, 895-898.	2.9	6
76	De Novo Truncating Variants in ASXL2 Are Associated with a Unique and Recognizable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 991-999.	6.2	68
77	Progressive multifocal leukoencephalopathy in an immunocompetent patient. Annals of Clinical and Translational Neurology, 2016, 3, 226-232.	3.7	19
78	Chromosomal abnormalities in hepatic cysts point to novel polycystic liver disease genes. European Journal of Human Genetics, 2016, 24, 1707-1714.	2.8	14
79	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.	2.3	19
80	Reliable Next-Generation Sequencing of Formalin-Fixed, Paraffin-Embedded Tissue Using Single Molecule Tags. Journal of Molecular Diagnostics, 2016, 18, 851-863.	2.8	94
81	ATP6AP1 deficiency causes an immunodeficiency with hepatopathy, cognitive impairment and abnormal protein glycosylation. Nature Communications, 2016, 7, 11600.	12.8	110
82	Parent-of-origin-specific signatures of de novo mutations. Nature Genetics, 2016, 48, 935-939.	21.4	266
83	RareVariantVis: new tool for visualization of causative variants in rare monogenic disorders using whole genome sequencing data. Bioinformatics, 2016, 32, 3018-3020.	4.1	7
84	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.	2.8	34
85	TRIOloss of function is associated with mild intellectual disability and affects dendritic branching and synapse function. Human Molecular Genetics, 2016, 25, 892-902.	2.9	94
86	Novel mutations in LRP6 highlight the role of WNT signaling in tooth agenesis. Genetics in Medicine, 2016, 18, 1158-1162.	2.4	58
87	CCDC115 Deficiency Causes a Disorder of Golgi Homeostasis with Abnormal Protein Glycosylation. American Journal of Human Genetics, 2016, 98, 310-321.	6.2	88
88	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.	6.2	73
89	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.	6.2	88
90	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. American Journal of Human Genetics, 2016, 98, 541-552.	6.2	132

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91	LRP5 variants may contribute to ADPKD. European Journal of Human Genetics, 2016, 24, 237-242.	2.8	28
92	EPHB4 kinaseâ€“inactivating mutations cause autosomal dominant lymphatic-related hydrops fetalis. Journal of Clinical Investigation, 2016, 126, 3080-3088.	8.2	83
93	Deleterious Germline BLM Mutations and the Risk for Early-onset Colorectal Cancer. Scientific Reports, 2015, 5, 14060.	3.3	67
94	B56Î´-related protein phosphatase 2A dysfunction identified in patients with intellectual disability. Journal of Clinical Investigation, 2015, 125, 3051-3062.	8.2	91
95	Syndromic X-linked intellectual disability segregating with a missense variant in RLIM. European Journal of Human Genetics, 2015, 23, 1652-1656.	2.8	30
96	Post-zygotic Point Mutations Are an Underrecognized Source of De Novo Genomic Variation. American Journal of Human Genetics, 2015, 97, 67-74.	6.2	215
97	De novo mutations in PLXND1 and REV3L cause MÃ¶bius syndrome. Nature Communications, 2015, 6, 7199.	12.8	76
98	Thyroid hormone resistance syndrome due to mutations in the thyroid hormone receptor Î± gene (THRA). Journal of Medical Genetics, 2015, 52, 312-316.	3.2	80
99	Homozygous mutation of STXBP5L explains an autosomal recessive infantile-onset neurodegenerative disorder. Human Molecular Genetics, 2015, 24, 2000-2010.	2.9	25
100	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.	6.2	230
101	Recurrent Mutations in the Basic Domain of TWIST2 Cause Ablepharon Macrostomia and Barber-Say Syndromes. American Journal of Human Genetics, 2015, 97, 99-110.	6.2	61
102	Exome Sequencing in an Admixed Isolated Population Indicates NFXL1 Variants Confer a Risk for Specific Language Impairment. PLoS Genetics, 2015, 11, e1004925.	3.5	50
103	A germline homozygous mutation in the base-excision repair gene NTHL1 causes adenomatous polyposis and colorectal cancer. Nature Genetics, 2015, 47, 668-671.	21.4	311
104	DVL1 Frameshift Mutations Clustering in the Penultimate Exon Cause Autosomal-Dominant Robinow Syndrome. American Journal of Human Genetics, 2015, 96, 612-622.	6.2	110
105	Refinement of the critical 2p25.3 deletion region: the role of MYT1L in intellectual disability and obesity. Genetics in Medicine, 2015, 17, 460-466.	2.4	45
106	Baraitserâ€“Winter cerebrofrontofacial syndrome: delineation of the spectrum in 42 cases. European Journal of Human Genetics, 2015, 23, 292-301.	2.8	115
107	Accurate Detection of Low Mosaic Mutations Associated with Therapy Resistance in Pediatric Acute Lymphoblastic Leukemia Using Single Molecule Tagging and Deep-Sequencing. Blood, 2015, 126, 2608-2608.	1.4	0
108	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.	3.5	41

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109	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.	7.1	79
110	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.	2.8	45
111	CantÃº Syndrome Resulting from Activating Mutation in theKCNJ8Gene. Human Mutation, 2014, 35, 809-813.	2.5	92
112	Early presentation of cystic kidneys in a family with a homozygous INVS mutation. American Journal of Medical Genetics, Part A, 2014, 164, 1627-1634.	1.2	7
113	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.	9.6	101
114	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.	5.1	58
115	Prioritization of neurodevelopmental disease genes by discovery of new mutations. Nature Neuroscience, 2014, 17, 764-772.	14.8	148
116	Diagnostic serum glycosylation profile in patients with intellectual disability as a result of MAN1B1 deficiency. Brain, 2014, 137, 1030-1038.	7.6	41
117	Refining analyses of copy number variation identifies specific genes associated with developmental delay. Nature Genetics, 2014, 46, 1063-1071.	21.4	583
118	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.	7.1	211
119	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.	6.2	110
120	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.	6.2	59
121	Genome sequencing identifies major causes of severe intellectual disability. Nature, 2014, 511, 344-347.	27.8	996
122	<scp>MLL2</scp> mutation detection in 86 patients with Kabuki syndrome: a genotypeâ€“phenotype study. Clinical Genetics, 2013, 84, 539-545.	2.0	85
123	Coffin-Siris Syndrome and the BAF Complex: Genotype-Phenotype Study in 63 Patients. Human Mutation, 2013, 34, 1519-1528.	2.5	178
124	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.	1.2	70
125	Functional genomics identifies type I interferon pathway as central for host defense against Candida albicans. Nature Communications, 2013, 4, 1342.	12.8	157
126	Mutations in MED12 Cause X-Linked Ohdo Syndrome. American Journal of Human Genetics, 2013, 92, 401-406.	6.2	78

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127	Mutations in ANTXR1 Cause GAPO Syndrome. American Journal of Human Genetics, 2013, 92, 792-799.	6.2	73
128	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.	6.2	197
129	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.	2.7	60
130	A complex V ATP5A1 defect causes fatal neonatal mitochondrial encephalopathy. Brain, 2013, 136, 1544-1554.	7.6	80
131	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.	3.2	127
132	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.	7.1	193
133	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.	7.1	144
134	Exome sequencing identifies putative drivers of progression of transient myeloproliferative disorder to AMKL in infants with Down syndrome. Blood, 2013, 122, 554-561.	1.4	72
135	Diagnostic Exome Sequencing in Persons With Severe Intellectual Disability. Obstetrical and Gynecological Survey, 2013, 68, 191-193.	0.4	22
136	A novel marine nitrite-oxidizing Nitrospira species from Dutch coastal North Sea water. Frontiers in Microbiology, 2013, 4, 60.	3.5	30
137	Novel PI3KÎ³ Mutation in a 44-Year-Old Man with Chronic Infections and Chronic Pelvic Pain. PLoS ONE, 2013, 8, e68118.	2.5	2
138	Gene identification in the congenital disorders of glycosylation type I by whole-exome sequencing. Human Molecular Genetics, 2012, 21, 4151-4161.	2.9	147
139	KIAA1797/FOCAD encodes a novel focal adhesion protein with tumour suppressor function in gliomas. Brain, 2012, 135, 1027-1041.	7.6	47
140	Diagnostic Exome Sequencing in Persons with Severe Intellectual Disability. New England Journal of Medicine, 2012, 367, 1921-1929.	27.0	1,367
141	Nextâ€generation genetic testing for retinitis pigmentosa. Human Mutation, 2012, 33, 963-972.	2.5	258
142	De novo mutations in the actin genes ACTB and ACTG1 cause Baraitser-Winter syndrome. Nature Genetics, 2012, 44, 440-444.	21.4	237
143	BDNF and DYRK1A Are Variable and Inversely Correlated in Lymphoblastoid Cell Lines from Down Syndrome Patients. Molecular Neurobiology, 2012, 46, 297-303.	4.0	15
144	Trisomy for Synaptojanin1 in Down syndrome is functionally linked to the enlargement of early endosomes. Human Molecular Genetics, 2012, 21, 3156-3172.	2.9	92

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145	Disease gene identification strategies for exome sequencing. European Journal of Human Genetics, 2012, 20, 490-497.	2.8	412
146	A phenotype map for 14q32.3 terminal deletions. American Journal of Medical Genetics, Part A, 2012, 158A, 695-706.	1.2	14
147	CantÃº Syndrome Is Caused by Mutations in ABCC9. American Journal of Human Genetics, 2012, 90, 1094-1101.	6.2	141
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.	2.5	10
149	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.	0.7	60
150	STAT1Mutations in Autosomal Dominant Chronic Mucocutaneous Candidiasis. New England Journal of Medicine, 2011, 365, 54-61.	27.0	614
151	De novo nonsense mutations in ASXL1 cause Bohring-Opitz syndrome. Nature Genetics, 2011, 43, 729-731.	21.4	236
152	Unlocking Mendelian disease using exome sequencing. Genome Biology, 2011, 12, 228.	9.6	228
153	STAT1 Hyperphosphorylation and Defective IL12R/IL23R Signaling Underlie Defective Immunity in Autosomal Dominant Chronic Mucocutaneous Candidiasis. PLoS ONE, 2011, 6, e29248.	2.5	101
154	Intragenic deletion in DYRK1A leads to mental retardation and primary microcephaly. Clinical Genetics, 2011, 79, 296-299.	2.0	94
155	Exome Sequencing Identifies Truncating Mutations in Human SERPINF1 in Autosomal-Recessive Osteogenesis Imperfecta. American Journal of Human Genetics, 2011, 88, 362-371.	6.2	316
156	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.	6.2	210
157	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.	6.2	202
158	Exome Sequencing Identifies WDR35 Variants Involved in Sensenbrenner Syndrome. American Journal of Human Genetics, 2010, 87, 418-423.	6.2	260
159	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.	6.2	125
160	Massively parallel sequencing of ataxia genes after array-based enrichment. Human Mutation, 2010, 31, 494-499.	2.5	86
161	Trisomic dose of several chromosome 21 genes perturbs haematopoietic stem and progenitor cell differentiation in Down's syndrome. Oncogene, 2010, 29, 6102-6114.	5.9	46
162	An adult female patient with ring chromosome 21: behavioural phenotype and results of high-resolution molecular characterisation. Acta Neuropsychiatrica, 2010, 22, 188-194.	2.1	1

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163	De novo mutations of SETBP1 cause Schinzel-Giedion syndrome. Nature Genetics, 2010, 42, 483-485.	21.4	417
164	A de novo paradigm for mental retardation. Nature Genetics, 2010, 42, 1109-1112.	21.4	751
165	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.	3.5	169
166	Rare pathogenic microdeletions and tandem duplications are microhomology-mediated and stimulated by local genomic architecture. Human Molecular Genetics, 2009, 18, 3579-3593.	2.9	143
167	Array-CGH in unclear syndromic nephropathies identifies a microdeletion in Xq22.3-q23. Pediatric Nephrology, 2009, 24, 1673-1681.	1.7	7
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