Christian Gilissen

List of Publications by Year
in descending order

216 papers	20,505 citations	9775 73 h-index	12585 132 g-index
249 all docs	249 docs citations	249 times ranked	32137 citing authors

#	Article	IF	Citations
1	BBS1 branchpoint variant is associated with non-syndromic retinitis pigmentosa. Journal of Medical Genetics, 2022, 59, 438-444.	1.5	13
2	Genome-wide variant calling in reanalysis of exome sequencing data uncovered a pathogenic TUBB3 variant. European Journal of Medical Genetics, 2022, 65, 104402.	0.7	2
3	A de novo paradigm for male infertility. Nature Communications, 2022, 13, 154.	5.8	38
4	Generation of a patient-derived induced pluripotent cell line (SCTCi016-A) carrying a homozygous variant in RPE65. Stem Cell Research, 2022, 60, 102689.	0.3	3
5	Clinical exome sequencingâ€"Mistakes and caveats. Human Mutation, 2022, 43, 1041-1055.	1.1	20
6	Inherited variants in CHD3 show variable expressivity in Snijders Blok-Campeau syndrome. Genetics in Medicine, 2022, 24, 1283-1296.	1.1	9
7	A Common Genomic Denominator for Neuroblastoma and Differentiated Thyroid Carcinoma? A Case Series in Children. Clinical Oncology, 2022, , .	0.6	0
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.	0.7	2
9	De novo mutations in children born after medical assisted reproduction. Human Reproduction, 2022, 37, 1360-1369.	0.4	12
10	Application of metabolite set enrichment analysis on untargeted metabolomics data prioritises relevant pathways and detects novel biomarkers for inherited metabolic disorders. Journal of Inherited Metabolic Disease, 2022, 45, 682-695.	1.7	6
11	Recommendations for whole genome sequencing in diagnostics for rare diseases. European Journal of Human Genetics, 2022, 30, 1017-1021.	1.4	48
12	Scrutinizing pathogenicity of the USH2A c.2276â€‰Gâ€‰>â€‰T; p.(Cys759Phe) variant. Npj Genomic Medicine, 2022, 7, .	1.7	5
13	DeNovoCNN: a deep learning approach to de novo variant calling in next generation sequencing data. Nucleic Acids Research, 2022, 50, e97-e97.	6.5	8
14	Reanalysis of exome negative patients with rare disease: a pragmatic workflow for diagnostic applications. Genome Medicine, 2022, 14, .	3.6	17
15	Missense variants in ANKRD11 cause KBG syndrome by impairment of stability or transcriptional activity of the encoded protein. Genetics in Medicine, 2022, 24, 2051-2064.	1.1	12
16	Characterization of SETD1A haploinsufficiency in humans and Drosophila defines a novel neurodevelopmental syndrome. Molecular Psychiatry, 2021, 26, 2013-2024.	4.1	43
17	Long-read trio sequencing of individuals with unsolved intellectual disability. European Journal of Human Genetics, 2021, 29, 637-648.	1.4	27
18	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356.	2.6	30

#	Article	IF	Citations
19	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
20	Lack of evidence for a role of PIWIL1 variants in human male infertility. Cell, 2021, 184, 1941-1942.	13.5	11
21	Novel Compound Heterozygous Mutation in TRAPPC9 Gene: The Relevance of Whole Genome Sequencing. Genes, 2021, 12, 557.	1.0	14
22	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
23	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
24	Cochlear supporting cells require GAS2 for cytoskeletal architecture and hearing. Developmental Cell, 2021, 56, 1526-1540.e7.	3.1	18
25	Impact of rare and common genetic variation in the interleukin-1 pathway on human cytokine responses. Genome Medicine, 2021, 13, 94.	3.6	5
26	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
27	Exome reanalysis and proteomic profiling identified TRIP4 as a novel cause of cerebellar hypoplasia and spinal muscular atrophy (PCH1). European Journal of Human Genetics, 2021, 29, 1348-1353.	1.4	10
28	Extending the allelic spectrum at noncoding risk loci of orofacial clefting. Human Mutation, 2021, 42, 1066-1078.	1.1	3
29	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
30	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
31	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. European Journal of Human Genetics, 2021, 29, 1337-1347.	1.4	34
32	Differences in the number of de novo mutations between individuals are due to small family-specific effects and stochasticity. Genome Research, 2021, 31, 1513-1518.	2.4	6
33	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	1.1	16
34	Metabolomics-Based Screening of Inborn Errors of Metabolism: Enhancing Clinical Application with a Robust Computational Pipeline. Metabolites, 2021, 11, 568.	1.3	11
35	Population sequencing data reveal a compendium of mutational processes in the human germ line. Science, 2021, 373, 1030-1035.	6.0	43
36	Long-read technologies identify a hidden inverted duplication in a family with choroideremia. Human Genetics and Genomics Advances, 2021, 2, 100046.	1.0	4

#	Article	IF	Citations
37	Whole genome sequencing and in vitro splice assays reveal genetic causes for inherited retinal diseases. Npj Genomic Medicine, 2021, 6, 97.	1.7	27
38	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
39	Accurate detection of clinically relevant uniparental disomy from exome sequencing data. Genetics in Medicine, 2020, 22, 803-808.	1.1	35
40	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
41	Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature, 2020, 586, 757-762.	13.7	343
42	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
43	Presence of Genetic Variants Among Young Men With Severe COVID-19. JAMA - Journal of the American Medical Association, 2020, 324, 663.	3.8	626
44	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
45	Sudden death in epilepsy and ectopic neurohypophysis in Joubert syndrome 23 diagnosed using SNVs/indels and structural variants pipelines on WGS data: a case report. BMC Medical Genetics, 2020, 21, 96.	2.1	5
46	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
47	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
48	BAZ2B haploinsufficiency as a cause of developmental delay, intellectual disability, and autism spectrum disorder. Human Mutation, 2020, 41, 921-925.	1.1	11
49	De Novo Variants in SPOP Cause Two Clinically Distinct Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 405-411.	2.6	8
50	Novel PXDN biallelic variants in patients with microphthalmia and anterior segment dysgenesis. Journal of Human Genetics, 2020, 65, 487-491.	1.1	5
51	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
52	In or Out? New Insights on Exon Recognition through Splice-Site Interdependency. International Journal of Molecular Sciences, 2020, 21, 2300.	1.8	8
53	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
54	Late-Onset Stargardt Disease Due to Mild, Deep-Intronic ABCA4 Alleles. , 2019, 60, 4249.		25

#	Article	IF	Citations
55	De Novo Mutations Reflect Development and Aging of the Human Germline. Trends in Genetics, 2019, 35, 828-839.	2.9	80
56	Front Cover, Volume 40, Issue 8. Human Mutation, 2019, 40, i-i.	1.1	51
57	Deregulated Adhesion Program in Palatal Keratinocytes of Orofacial Cleft Patients. Genes, 2019, 10, 836.	1.0	4
58	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
59	Exome sequencing in routine diagnostics: a generic test for 254 patients with primary immunodeficiencies. Genome Medicine, 2019, 11, 38.	3.6	49
60	Costâ€effective molecular inversion probeâ€based ABCA4 sequencing reveals deepâ€intronic variants in Stargardt disease. Human Mutation, 2019, 40, 1749-1759.	1.1	39
61	A systems genomics approach identifies SIGLEC15 as a susceptibility factor in recurrent vulvovaginal candidiasis. Science Translational Medicine, 2019, 11, .	5.8	38
62	MetaDome: Pathogenicity analysis of genetic variants through aggregation of homologous human protein domains. Human Mutation, 2019, 40, 1030-1038.	1.1	133
63	Exome sequencing in patients with chronic central serous chorioretinopathy. Scientific Reports, 2019, 9, 6598.	1.6	12
64	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
65	Deletions and loss-of-function variants in TP63 associated with orofacial clefting. European Journal of Human Genetics, 2019, 27, 1101-1112.	1.4	16
66	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
67	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
68	TBCK Encephaloneuropathy With Abnormal Lysosomal Storage: Use of a Structural Variant Bioinformatics Pipeline on Whole-Genome Sequencing Data Unravels a 20-Year-Old Clinical Mystery. Pediatric Neurology, 2019, 96, 74-75.	1.0	11
69	Genetic Spectrum of ABCA4-Associated Retinal Degeneration in Poland. Genes, 2019, 10, 959.	1.0	17
70	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
71	1 in 38 individuals at risk of a dominant medically actionable disease. European Journal of Human Genetics, 2019, 27, 325-330.	1.4	56
72	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

#	Article	IF	Citations
73	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
74	Mutations in SELENBP1, encoding a novel human methanethiol oxidase, cause extraoral halitosis. Nature Genetics, 2018, 50, 120-129.	9.4	86
75	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
76	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
77	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
78	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
79	OP0285â€…Identification of rare coding variants in il-1-related pathways in patients with adult-onset stillâ€™s disease. , 2018, , .		0
80	Identification of C12orf4 as a gene for autosomal recessive intellectual disability. Clinical Genetics, 2017, 91, 100-105.	1.0	15
81	Quantification of Phenotype Information Aids the Identification of Novel Disease Genes. Human Mutation, 2017, 38, 594-599.	1.1	3
82	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
83	Upstream SLC2A1 translation initiation causes GLUT1 deficiency syndrome. European Journal of Human Genetics, 2017, 25, 771-774.	1.4	15
84	Rare NOX3 Variants Confer Susceptibility to Agranulocytosis During Thyrostatic Treatment of Graves' Disease. Clinical Pharmacology and Therapeutics, 2017, 102, 1017-1024.	2.3	12
85	YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction. American Journal of Human Genetics, 2017, 100, 907-925.	2.6	125
86	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
87	MST1R mutation as a genetic cause of Lady Windermere syndrome. European Respiratory Journal, 2017, 49, 1601478.	3.1	18
88	Variation in a range of mTOR-related genes associates with intracranial volume and intellectual disability. Nature Communications, 2017, 8, 1052.	5.8	63
89	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
90	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

#	Article	IF	Citations
91	Candidate Genes for Nonsyndromic Cleft Palate Detected by Exome Sequencing. Journal of Dental Research, 2017, 96, 1314-1321.	2.5	27
92	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
93	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
94	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
95	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
96	NovelIRF6Mutations Detected in Orofacial Cleft Patients by Targeted Massively Parallel Sequencing. Journal of Dental Research, 2017, 96, 179-185.	2.5	12
97	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
98	Exome sequencing of Pakistani consanguineous families identifies 30 novel candidate genes for recessive intellectual disability. Molecular Psychiatry, 2017, 22, 1604-1614.	4.1	118
99	Overlapping SETBP1 gain-of-function mutations in Schinzel-Giedion syndrome and hematologic malignancies. PLoS Genetics, 2017, 13, e1006683.	1.5	35
100	Apoptosis-Related Gene Expression Profiling in Hematopoietic Cell Fractions of MDS Patients. PLoS ONE, 2016, 11, e0165582.	1.1	16
101	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
102	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
103	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
104	Panel-based NGS Reveals Novel Pathogenic Mutations in Autosomal Recessive Retinitis Pigmentosa. Scientific Reports, 2016, 6, 19531.	1.6	48
105	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
106	Novel bioinformatic developments for exome sequencing. Human Genetics, 2016, 135, 603-614.	1.8	37
107	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
108	Meta-analysis of 2,104 trios provides support for 10 new genes for intellectual disability. Nature Neuroscience, 2016, 19, 1194-1196.	7.1	407

#	Article	IF	Citations
109	Towards embryonic-like scaffolds for skin tissue engineering: identification of effector molecules and construction of scaffolds. Journal of Tissue Engineering and Regenerative Medicine, 2016, 10, E34-E44.	1.3	8
110	FAM222B Is Not a Likely Novel Candidate Gene for Cerebral Cavernous Malformations. Molecular Syndromology, 2016, 7, 144-152.	0.3	5
111	ATP6AP1 deficiency causes an immunodeficiency with hepatopathy, cognitive impairment and abnormal protein glycosylation. Nature Communications, 2016, 7, 11600.	5.8	110
112	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
113	Parent-of-origin-specific signatures of de novo mutations. Nature Genetics, 2016, 48, 935-939.	9.4	266
114	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
115	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
116	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
117	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
118	Novel mutations in LRP6 highlight the role of WNT signaling in tooth agenesis. Genetics in Medicine, 2016, 18, 1158-1162.	1.1	58
119	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
120	Genetic studies in intellectual disability and related disorders. Nature Reviews Genetics, 2016, 17, 9-18.	7.7	614
121	Novel genetic causes for cerebral visual impairment. European Journal of Human Genetics, 2016, 24, 660-665.	1.4	127
122	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
123	LRP5 variants may contribute to ADPKD. European Journal of Human Genetics, 2016, 24, 237-242.	1.4	28
124	EPHB4 kinaseâ€“inactivating mutations cause autosomal dominant lymphatic-related hydrops fetalis. Journal of Clinical Investigation, 2016, 126, 3080-3088.	3.9	83
125	Identification of Novel Candidate Genes for Early-Onset Colorectal Cancer Susceptibility. PLoS Genetics, 2016, 12, e1005880.	1.5	52
126	Differential effects of Wnt signaling in adult and fetal bone marrow-derived MSCs. Cytotherapy, 2015, 17, S34.	0.3	0

#	Article	IF	Citations
127	Comparison of Exome and Genome Sequencing Technologies for the Complete Capture of Proteinâ€Coding Regions. Human Mutation, 2015, 36, 815-822.	1.1	156
128	Deleterious Germline BLM Mutations and the Risk for Early-onset Colorectal Cancer. Scientific Reports, 2015, 5, 14060.	1.6	67
129	Syndromic X-linked intellectual disability segregating with a missense variant in RLIM. European Journal of Human Genetics, 2015, 23, 1652-1656.	1.4	30
130	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
131	De novo mutations in PLXND1 and REV3L cause MÃ¶bius syndrome. Nature Communications, 2015, 6, 7199.	5.8	76
132	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
133	A missense mutation underlies defective <scp>SOCS</scp>4 function in a family with autoimmunity. Journal of Internal Medicine, 2015, 278, 203-210.	2.7	6
134	Variants inCUL4Bare Associated with Cerebral Malformations. Human Mutation, 2015, 36, 106-117.	1.1	37
135	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
136	The heat shock response restricts virus infection in Drosophila. Scientific Reports, 2015, 5, 12758.	1.6	86
137	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
138	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
139	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
140	A recent bottleneck of Y chromosome diversity coincides with a global change in culture. Genome Research, 2015, 25, 459-466.	2.4	348
141	Immunoglobulin rearrangement analysis from multiple lesions in the same patient using nextâ€generation sequencing. Histopathology, 2015, 67, 843-858.	1.6	5
142	Exome Sequencing Identifies Three Novel Candidate Genes Implicated in Intellectual Disability. PLoS ONE, 2014, 9, e112687.	1.1	23
143	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
144	Improved exome prioritization of disease genes through cross-species phenotype comparison. Genome Research, 2014, 24, 340-348.	2.4	300

#	Article	IF	Citations
145	Cord Blood Mesenchymal Stem Cells Suppress DC-T Cell Proliferation via Prostaglandin B2. Stem Cells and Development, 2014, 23, 1582-1593.	1.1	16
146	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
147	Use of animal models for exome prioritization of rare disease genes. Orphanet Journal of Rare Diseases, 2014, 9, O19.	1.2	0
148	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
149	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
150	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
151	Genome sequencing identifies major causes of severe intellectual disability. Nature, 2014, 511, 344-347.	13.7	996
152	Differential Effects of Wnt Signaling on Proliferation and Hematopoietic Support of Adult and Fetal Bone Marrow-Derived MSCs. Blood, 2014, 124, 5137-5137.	0.6	0
153	A different balance in wnt-signaling in adult and fetal bone marrow-derived MSC. Experimental Hematology, 2013, 41, S75.	0.2	0
154	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
155	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
156	Homozygous and heterozygous disruptions of ANK3: at the crossroads of neurodevelopmental and psychiatric disorders. Human Molecular Genetics, 2013, 22, 1960-1970.	1.4	137
157	<scp>MLL2</scp> mutation detection in 86 patients with Kabuki syndrome: a genotypeâ€“phenotype study. Clinical Genetics, 2013, 84, 539-545.	1.0	85
158	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
159	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
160	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
161	Mutations in MED12 Cause X-Linked Ohdo Syndrome. American Journal of Human Genetics, 2013, 92, 401-406.	2.6	78
162	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

#	Article	IF	Citations
163	1387 IDENTIFICATION OF A NOVEL GENE ASSOCIATED WITH POLYCYSTIC LIVER AND KIDNEY DISEASES. Journal of Hepatology, 2013, 58, S557-S558.	1.8	1
164	A complex V ATP5A1 defect causes fatal neonatal mitochondrial encephalopathy. Brain, 2013, 136, 1544-1554.	3.7	80
165	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
166	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
167	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
168	Non-syndromic Tooth Agenesis Associated with a Nonsense Mutation in Ectodysplasin-A (EDA). Journal of Dental Research, 2013, 92, 507-511.	2.5	29
169	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
170	Diagnostic Exome Sequencing in Persons With Severe Intellectual Disability. Obstetrical and Gynecological Survey, 2013, 68, 191-193.	0.2	22
171	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
172	Gene identification in the congenital disorders of glycosylation type I by whole-exome sequencing. Human Molecular Genetics, 2012, 21, 4151-4161.	1.4	147
173	Nuclear ReceptorsNur77andNurr1Modulate Mesenchymal Stromal Cell Migration. Stem Cells and Development, 2012, 21, 228-238.	1.1	56
174	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
175	Mutations in SWI/SNF chromatin remodeling complex gene ARID1B cause Coffin-Siris syndrome. Nature Genetics, 2012, 44, 379-380.	9.4	312
176	Mutations in ISPD cause Walker-Warburg syndrome and defective glycosylation of Î±-dystroglycan. Nature Genetics, 2012, 44, 581-585.	9.4	191
177	Diagnostic Exome Sequencing in Persons with Severe Intellectual Disability. New England Journal of Medicine, 2012, 367, 1921-1929.	13.9	1,367
178	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
179	Nextâ€generation genetic testing for retinitis pigmentosa. Human Mutation, 2012, 33, 963-972.	1.1	258
180	Targeted Next Generation Sequencing Reveals a Novel Intragenic Deletion of the TPO Gene in a Family with Intellectual Disability. Archives of Medical Research, 2012, 43, 312-316.	1.5	8

#	Article	IF	Citations
181	Mutations in the chromatin modifier gene KANSL1 cause the 17q21.31 microdeletion syndrome. Nature Genetics, 2012, 44, 639-641.	9.4	194
182	De novo mutations in the actin genes ACTB and ACTG1 cause Baraitser-Winter syndrome. Nature Genetics, 2012, 44, 440-444.	9.4	237
183	Disruption of an EHMT1-Associated Chromatin-Modification Module Causes Intellectual Disability. American Journal of Human Genetics, 2012, 91, 73-82.	2.6	214
184	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
185	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
186	Validation Study of Existing Gene Expression Signatures for Anti-TNF Treatment in Patients with Rheumatoid Arthritis. PLoS ONE, 2012, 7, e33199.	1.1	61
187	Disease gene identification strategies for exome sequencing. European Journal of Human Genetics, 2012, 20, 490-497.	1.4	412
188	CantÃº Syndrome Is Caused by Mutations in ABCC9. American Journal of Human Genetics, 2012, 90, 1094-1101.	2.6	141
189	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
190	STAT1Mutations in Autosomal Dominant Chronic Mucocutaneous Candidiasis. New England Journal of Medicine, 2011, 365, 54-61.	13.9	614
191	De novo nonsense mutations in ASXL1 cause Bohring-Opitz syndrome. Nature Genetics, 2011, 43, 729-731.	9.4	236
192	Unlocking Mendelian disease using exome sequencing. Genome Biology, 2011, 12, 228.	13.9	228
193	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
194	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
195	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
196	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
197	Exome Sequencing of Late Recurrence T-Cell Acute Lymphoblastic Leukemia in Children Confirms Second Leukemia and Exposes Predisposition Candidate Genes. Blood, 2011, 118, 755-755.	0.6	0
198	BTG1 regulates glucocorticoid receptor autoinduction in acute lymphoblastic leukemia. Blood, 2010, 115, 4810-4819.	0.6	69

#	Article	IF	Citations
199	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
200	Exome Sequencing Identifies WDR35 Variants Involved in Sensenbrenner Syndrome. American Journal of Human Genetics, 2010, 87, 418-423.	2.6	260
201	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
202	Massively parallel sequencing of ataxia genes after array-based enrichment. Human Mutation, 2010, 31, 494-499.	1.1	86
203	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
204	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
205	De novo mutations of SETBP1 cause Schinzel-Giedion syndrome. Nature Genetics, 2010, 42, 483-485.	9.4	417
206	A de novo paradigm for mental retardation. Nature Genetics, 2010, 42, 1109-1112.	9.4	751
207	Dystrophin Gene Mutation Location and the Risk of Cognitive Impairment in Duchenne Muscular Dystrophy. PLoS ONE, 2010, 5, e8803.	1.1	147
208	Accurate Distinction of Pathogenic from Benign CNVs in Mental Retardation. PLoS Computational Biology, 2010, 6, e1000752.	1.5	46
209	Functional Differences Between Mesenchymal Stem Cell Populations Are Reflected by Their Transcriptome. Stem Cells and Development, 2010, 19, 481-490.	1.1	124
210	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
211	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
212	Nur77 and Nurr1, Two Novel Players In MSC Migration and Immune Function.. Blood, 2010, 116, 3854-3854.	0.6	1
213	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
214	489 MICROARRAY PATHWAY ANALYSIS OF INHIBITION OF CHONDROGENESIS OF HUMAN MESENCHYMAL STEM CELLS BY OSTEOARTHRITIC SYNOVIUM DERIVED CONDITIONED MEDIA. Osteoarthritis and Cartilage, 2009, 17, S262-S263.	0.6	0
215	Molecular Signature of Migratory Human Mesenchymal Stromal Cells; Influence of the Cell Cycle.. Blood, 2009, 114, 1450-1450.	0.6	1
216	Error probabilities for local extrema in gene expression data. Pattern Recognition Letters, 2007, 28, 2133-2142.	2.6	1