Hans Van Bokhoven

List of Publications by Year
in descending order

307	25,786	4370 86	9311 143
papers	citations	h-index	g-index

325	325	325	29064
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Common genetic variants influence human subcortical brain structures. Nature, 2015, 520, 224-229.	13.7	772
2	Mutations in the gene encoding the 3â€²-5â€² DNA exonuclease TREX1 cause Aicardi-GoutiÃ¨res syndrome at the AGS1 locus. Nature Genetics, 2006, 38, 917-920.	9.4	752
3	Heterozygous Germline Mutations in the p53 Homolog p63 Are the Cause of EEC Syndrome. Cell, 1999, 99, 143-153.	13.5	638
4	Mutations in the O-Mannosyltransferase Gene POMT1 Give Rise to the Severe Neuronal Migration Disorder Walker-Warburg Syndrome. American Journal of Human Genetics, 2002, 71, 1033-1043.	2.6	636
5	A systematic, large-scale resequencing screen of X-chromosome coding exons in mental retardation. Nature Genetics, 2009, 41, 535-543.	9.4	528
6	POMT2 mutations cause Â-dystroglycan hypoglycosylation and Walker-Warburg syndrome. Journal of Medical Genetics, 2005, 42, 907-912.	1.5	374
7	Loss-of-Function Mutations in Euchromatin Histone Methyl Transferase 1 (EHMT1) Cause the 9q34 Subtelomeric Deletion Syndrome. American Journal of Human Genetics, 2006, 79, 370-377.	2.6	343
8	p63 Gene Mutations in EEC Syndrome, Limb-Mammary Syndrome, and Isolated Split Handâ€“Split Foot Malformation Suggest a Genotype-Phenotype Correlation. American Journal of Human Genetics, 2001, 69, 481-492.	2.6	331
9	Impaired glycosylation and cutis laxa caused by mutations in the vesicular H+-ATPase subunit ATP6V0A2. Nature Genetics, 2008, 40, 32-34.	9.4	330
10	Mutations in ARHGEF6, encoding a guanine nucleotide exchange factor for Rho GTPases, in patients with X-linked mental retardation. Nature Genetics, 2000, 26, 247-250.	9.4	329
11	Genetic and Epigenetic Networks in Intellectual Disabilities. Annual Review of Genetics, 2011, 45, 81-104.	3.2	329
12	Hay-Wells syndrome is caused by heterozygous missense mutations in the SAM domain of p63. Human Molecular Genetics, 2001, 10, 221-229.	1.4	319
13	Homozygous Mutation in SPATA16 Is Associated with Male Infertility in Human Globozoospermia. American Journal of Human Genetics, 2007, 81, 813-820.	2.6	273
14	p63-Associated Disorders. Cell Cycle, 2007, 6, 262-268.	1.3	267
15	Transcription Factor SOX3 Is Involved in X-Linked Mental Retardation with Growth Hormone Deficiency. American Journal of Human Genetics, 2002, 71, 1450-1455.	2.6	265
16	Deciphering the Glycosylome of Dystroglycanopathies Using Haploid Screens for Lassa Virus Entry. Science, 2013, 340, 479-483.	6.0	262
17	SRD5A3 Is Required for Converting Polyprenol to Dolichol and Is Mutated in a Congenital Glycosylation Disorder. Cell, 2010, 142, 203-217.	13.5	253
18	Novel genetic loci associated with hippocampal volume. Nature Communications, 2017, 8, 13624.	5.8	250

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19	ARX, a novel Prd-class-homeobox gene highly expressed in the telencephalon, is mutated in X-linked mental retardation. Human Molecular Genetics, 2002, 11, 981-991.	1.4	248
20	Mutations in the FKRP gene can cause muscle-eye-brain disease and Walker-Warburg syndrome. Journal of Medical Genetics, 2004, 41, e61-e61.	1.5	243
21	X-exome sequencing of 405 unresolved families identifies seven novel intellectual disability genes. Molecular Psychiatry, 2016, 21, 133-148.	4.1	243
22	Mutation of the gene encoding the ROR2 tyrosine kinase causes autosomal recessive Robinow syndrome. Nature Genetics, 2000, 25, 423-426.	9.4	242
23	Dominant isolated renal magnesium loss is caused by misrouting of the Na+,K+-ATPase Î³-subunit. Nature Genetics, 2000, 26, 265-266.	9.4	234
24	Complex Transcriptional Effects of p63 Isoforms: Identification of Novel Activation and Repression Domainsâ€. Molecular and Cellular Biology, 2002, 22, 8659-8668.	1.1	224
25	Mutations in the pre-replication complex cause Meier-Gorlin syndrome. Nature Genetics, 2011, 43, 356-359.	9.4	219
26	A new gene involved in X-linked mental retardation identified by analysis of an X;2 balanced translocation. Nature Genetics, 2000, 24, 167-170.	9.4	215
27	WNT5A mutations in patients with autosomal dominant Robinow syndrome. Developmental Dynamics, 2010, 239, 327-337.	0.8	214
28	Disruption of an EHMT1-Associated Chromatin-Modification Module Causes Intellectual Disability. American Journal of Human Genetics, 2012, 91, 73-82.	2.6	214
29	Novel genetic loci underlying human intracranial volume identified through genome-wide association. Nature Neuroscience, 2016, 19, 1569-1582.	7.1	213
30	Genetic influences on schizophrenia and subcortical brain volumes: large-scale proof of concept. Nature Neuroscience, 2016, 19, 420-431.	7.1	204
31	MicroRNA networks direct neuronal development and plasticity. Cellular and Molecular Life Sciences, 2012, 69, 89-102.	2.4	202
32	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
33	Mutations in PHF8 are associated with X linked mental retardation and cleft lip/cleft palate. Journal of Medical Genetics, 2005, 42, 780-786.	1.5	194
34	Further clinical and molecular delineation of the 9q subtelomeric deletion syndrome supports a major contribution of EHMT1 haploinsufficiency to the core phenotype. Journal of Medical Genetics, 2009, 46, 598-606.	1.5	194
35	Genetic architecture of subcortical brain structures in 38,851 individuals. Nature Genetics, 2019, 51, 1624-1636.	9.4	192
36	Mutations in ISPD cause Walker-Warburg syndrome and defective glycosylation of Î±-dystroglycan. Nature Genetics, 2012, 44, 581-585.	9.4	191

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37	Mutations in different components of FGF signaling in LADD syndrome. Nature Genetics, 2006, 38, 414-417.	9.4	190
38	Splitting p63. American Journal of Human Genetics, 2002, 71, 1-13.	2.6	188
39	Submicroscopic Duplications of the Hydroxysteroid Dehydrogenase HSD17B10 and the E3 Ubiquitin Ligase HUWE1 Are Associated with Mental Retardation. American Journal of Human Genetics, 2008, 82, 432-443.	2.6	187
40	Epigenetic Regulation of Learning and Memory by Drosophila EHMT/G9a. PLoS Biology, 2011, 9, e1000569.	2.6	185
41	A C-Terminal Inhibitory Domain Controls the Activity of p63 by an Intramolecular Mechanism. Molecular and Cellular Biology, 2002, 22, 8601-8611.	1.1	183
42	OFD1 Is Mutated in X-Linked Joubert Syndrome and Interacts with LCA5-Encoded Lebercilin. American Journal of Human Genetics, 2009, 85, 465-481.	2.6	180
43	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
44	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
45	Pathogenesis of split-hand/split-foot malformation. Human Molecular Genetics, 2003, 12, 51R-60.	1.4	167
46	Disruption of the gene Euchromatin Histone Methyl Transferase1 (Eu-HMTase1) is associated with the 9q34 subtelomeric deletion syndrome. Journal of Medical Genetics, 2005, 42, 299-306.	1.5	162
47	The p63 gene in EEC and other syndromes. Journal of Medical Genetics, 2002, 39, 377-381.	1.5	159
48	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
49	Mutations inDYNC1H1cause severe intellectual disability with neuronal migration defects. Journal of Medical Genetics, 2012, 49, 179-183.	1.5	151
50	Mutations in the Human TBX4 Gene Cause Small Patella Syndrome. American Journal of Human Genetics, 2004, 74, 1239-1248.	2.6	149
51	Inhibitory control of the excitatory/inhibitory balance in psychiatric disorders. F1000Research, 2018, 7, 23.	0.8	149
52	Chromosome 1p21.3 microdeletions comprising DPYD and MIR137 are associated with intellectual disability. Journal of Medical Genetics, 2011, 48, 810-818.	1.5	146
53	Mutations in the Embryonal Subunit of the Acetylcholine Receptor (CHRNG) Cause Lethal and Escobar Variants of Multiple Pterygium Syndrome. American Journal of Human Genetics, 2006, 79, 390-395.	2.6	145
54	A homozygous nonsense mutation in the Fukutin gene causes a Walker-Warburg syndrome phenotype. Journal of Medical Genetics, 2003, 40, 845-848.	1.5	141

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55	Mutations in the polyglutamine binding protein 1 gene cause X-linked mental retardation. Nature Genetics, 2003, 35, 313-315.	9.4	139
56	MYCN haploinsufficiency is associated with reduced brain size and intestinal atresias in Feingold syndrome. Nature Genetics, 2005, 37, 465-467.	9.4	138
57	Pattern ofp63 mutations and their phenotypesâ€"update. American Journal of Medical Genetics, Part A, 2006, 140A, 1396-1406.	0.7	137
58	Homozygous and heterozygous disruptions of ANK3: at the crossroads of neurodevelopmental and psychiatric disorders. Human Molecular Genetics, 2013, 22, 1960-1970.	1.4	137
59	Transcription factor p63 bookmarks and regulates dynamic enhancers during epidermal differentiation. EMBO Reports, 2015, 16, 863-878.	2.0	134
60	Missense mutations in Î²-1,3-N-acetylglucosaminyltransferase 1 (B3GNT1) cause Walkerâ€"Warburg syndrome. Human Molecular Genetics, 2013, 22, 1746-1754.	1.4	133
61	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. American Journal of Human Genetics, 2016, 98, 541-552.	2.6	132
62	Cloning and characterization of the human choroideremia gene. Human Molecular Genetics, 1994, 3, 1041-1046.	1.4	131
63	Structural variation in Xq28: MECP2 duplications in 1% of patients with unexplained XLMR and in 2% of male patients with severe encephalopathy. European Journal of Human Genetics, 2009, 17, 444-453.	1.4	130
64	Autosomal Recessive Dilated Cardiomyopathy due to DOLK Mutations Results from Abnormal Dystroglycan O-Mannosylation. PLoS Genetics, 2011, 7, e1002427.	1.5	130
65	Genotypeâ€"phenotype studies in nail-patella syndrome show that LMX1B mutation location is involved in the risk of developing nephropathy. European Journal of Human Genetics, 2005, 13, 935-946.	1.4	129
66	Intragenic deletion in the LARGE gene causes Walker-Warburg syndrome. Human Genetics, 2007, 121, 685-690.	1.8	126
67	Reduced exploration, increased anxiety, and altered social behavior: Autistic-like features of euchromatin histone methyltransferase 1 heterozygous knockout mice. Behavioural Brain Research, 2010, 208, 47-55.	1.2	126
68	Cooperation between the transcription factors p63 and IRF6 is essential to prevent cleft palate in mice. Journal of Clinical Investigation, 2010, 120, 1561-1569.	3.9	123
69	Missense mutation in PAK3, R67C, causes X-linked nonspecific mental retardation. American Journal of Medical Genetics Part A, 2000, 93, 294-298.	2.4	122
70	Exome sequencing of Pakistani consanguineous families identifies 30 novel candidate genes for recessive intellectual disability. Molecular Psychiatry, 2017, 22, 1604-1614.	4.1	118
71	Mutations in the FTSJ1 Gene Coding for a Novel S-Adenosylmethionineâ€"Binding Protein Cause Nonsyndromic X-Linked Mental Retardation. American Journal of Human Genetics, 2004, 75, 305-309.	2.6	117
72	The Opitz syndrome gene product, MID1, associates with microtubules. Proceedings of the National Academy of Sciences of the United States of America, 1999, 96, 2794-2799.	3.3	116

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73	Mutations in the p53 homolog p63: allele-specific developmental syndromes in humans. Trends in Molecular Medicine, 2002, 8, 133-139.	3.5	116
74	Functional convergence of histone methyltransferases EHMT1 and KMT2C involved in intellectual disability and autism spectrum disorder. PLoS Genetics, 2017, 13, e1006864.	1.5	116
75	P63 gene mutations and human developmental syndromes. American Journal of Medical Genetics Part A, 2002, 112, 284-290.	2.4	115
76	Asymmetry within and around the human planum temporale is sexually dimorphic and influenced by genes involved in steroid hormone receptor activity. Cortex, 2015, 62, 41-55.	1.1	114
77	A Novel Ribosomal S6-Kinase (RSK4; RPS6KA6) Is Commonly Deleted in Patients with Complex X-Linked Mental Retardation. Genomics, 1999, 62, 332-343.	1.3	113
78	CLPB Mutations Cause 3-Methylglutaconic Aciduria, Progressive Brain Atrophy, Intellectual Disability, Congenital Neutropenia, Cataracts, Movement Disorder. American Journal of Human Genetics, 2015, 96, 245-257.	2.6	111
79	The expanding phenotype ofPOMT1mutations: from Walker-Warburg syndrome to congenital muscular dystrophy, microcephaly, and mental retardation. Human Mutation, 2006, 27, 453-459.	1.1	106
80	Gain-of-function mutation in ADULT syndrome reveals the presence of a second transactivation domain in p63. Human Molecular Genetics, 2002, 11, 799-804.	1.4	104
81	Mutation frequencies of X-linked mental retardation genes in families from the EuroMRX consortium. Human Mutation, 2007, 28, 207-208.	1.1	103
82	SLC29A3 gene is mutated in pigmented hypertrichosis with insulin-dependent diabetes mellitus syndrome and interacts with the insulin signaling pathway. Human Molecular Genetics, 2009, 18, 2257-2265.	1.4	100
83	PRPS1 Mutations: Four Distinct Syndromes and Potential Treatment. American Journal of Human Genetics, 2010, 86, 506-518.	2.6	99
84	Identification of ANKRD11 and ZNF778 as candidate genes for autism and variable cognitive impairment in the novel 16q24.3 microdeletion syndrome. European Journal of Human Genetics, 2010, 18, 429-435.	1.4	99
85	Glyc-O-genetics of Walker-Warburg syndrome. Clinical Genetics, 2004, 67, 281-289.	1.0	98
86	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
87	Mutations in the Epithelial Cadherin-p120-Catenin Complex Cause Mendelian Non-Syndromic Cleft Lip with or without Cleft Palate. American Journal of Human Genetics, 2018, 102, 1143-1157.	2.6	94
88	Analysis of the p63 gene in classical EEC syndrome, related syndromes, and non-syndromic orofacial clefts. Journal of Medical Genetics, 2002, 39, 559-566.	1.5	93
89	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
90	De novo mutations in beta-catenin (CTNNB1) appear to be a frequent cause of intellectual disability: expanding the mutational and clinical spectrum. Human Genetics, 2015, 134, 97-109.	1.8	93

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91	Neuronal network dysfunction in a model for Kleefstra syndrome mediated by enhanced NMDAR signaling. Nature Communications, 2019, 10, 4928.	5.8	92
92	Involvement of the kinesin family membersKIF4AandKIF5Cin intellectual disability and synaptic function. Journal of Medical Genetics, 2014, 51, 487-494.	1.5	90
93	Variants in PUS7 Cause Intellectual Disability with Speech Delay, Microcephaly, Short Stature, and Aggressive Behavior. American Journal of Human Genetics, 2018, 103, 1045-1052.	2.6	89
94	Limb Mammary Syndrome: A New Genetic Disorder with Mammary Hypoplasia, Ectrodactyly, and Other Hand/Foot Anomalies Maps to Human Chromosome 3q27. American Journal of Human Genetics, 1999, 64, 538-546.	2.6	88
95	PLEXIN-D1, a novel plexin family member, is expressed in vascular endothelium and the central nervous system during mouse embryogenesis. Developmental Dynamics, 2002, 225, 336-343.	0.8	88
96	A novel cerebello-ocular syndrome with abnormal glycosylation due to abnormalities in dolichol metabolism. Brain, 2010, 133, 3210-3220.	3.7	87
97	Chromosomal copy number changes in patients with non-syndromic X linked mental retardation detected by array CGH. Journal of Medical Genetics, 2005, 43, 362-370.	1.5	85
98	Mutations in the ZNF41 Gene Are Associated with Cognitive Deficits: Identification of a New Candidate for X-Linked Mental Retardation. American Journal of Human Genetics, 2003, 73, 1341-1354.	2.6	83
99	Genetic and epigenetic defects in mental retardation. International Journal of Biochemistry and Cell Biology, 2009, 41, 96-107.	1.2	83
100	Disruption of the Podosome Adaptor Protein TKS4 (SH3PXD2B) Causes the Skeletal Dysplasia, Eye, and Cardiac Abnormalities of Frank-Ter Haar Syndrome. American Journal of Human Genetics, 2010, 86, 254-261.	2.6	83
101	MicroRNA-137 Controls AMPA-Receptor-Mediated Transmission and mGluR-Dependent LTD. Cell Reports, 2015, 11, 1876-1884.	2.9	82
102	Loss of the BMP Antagonist, SMOC-1, Causes Ophthalmo-Acromelic (Waardenburg Anophthalmia) Syndrome in Humans and Mice. PLoS Genetics, 2011, 7, e1002114.	1.5	81
103	Arts Syndrome Is Caused by Loss-of-Function Mutations in PRPS1. American Journal of Human Genetics, 2007, 81, 507-518.	2.6	80
104	Heterozygous Mutations of FREM1 Are Associated with an Increased Risk of Isolated Metopic Craniosynostosis in Humans and Mice. PLoS Genetics, 2011, 7, e1002278.	1.5	80
105	Mutations in MED12 Cause X-Linked Ohdo Syndrome. American Journal of Human Genetics, 2013, 92, 401-406.	2.6	78
106	The genetics of cognitive epigenetics. Neuropharmacology, 2014, 80, 83-94.	2.0	78
107	Aicardi-GoutiÃ¨res Syndrome Displays Genetic Heterogeneity with One Locus (AGS1) on Chromosome 3p21. American Journal of Human Genetics, 2000, 67, 213-221.	2.6	77
108	Cadherin-13 is a critical regulator of GABAergic modulation in human stem-cell-derived neuronal networks. Molecular Psychiatry, 2022, 27, 1-18.	4.1	77

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109	Feingold syndrome: Clinical review and genetic mapping. American Journal of Medical Genetics Part A, 2003, 122A, 294-300.	2.4	76
110	De novo mutations in PLXND1 and REV3L cause MÃ¶bius syndrome. Nature Communications, 2015, 6, 7199.	5.8	76
111	ZNF674: A New KrÃ¼ppel-Associated Boxâ€“Containing Zinc-Finger Gene Involved in Nonsyndromic X-Linked Mental Retardation. American Journal of Human Genetics, 2006, 78, 265-278.	2.6	75
112	Mutations in CSPP1 Lead to Classical Joubert Syndrome. American Journal of Human Genetics, 2014, 94, 80-86.	2.6	75
113	Histone Methylation by the Kleefstra Syndrome Protein EHMT1 Mediates Homeostatic Synaptic Scaling. Neuron, 2016, 91, 341-355.	3.8	74
114	Genotype-phenotype correlations in MYCN-related Feingold syndrome. Human Mutation, 2008, 29, 1125-1132.	1.1	72
115	Trismus-pseudocamptodactyly syndrome is caused by recurrent mutation ofMYH8. American Journal of Medical Genetics, Part A, 2006, 140A, 2387-2393.	0.7	71
116	Mutation spectrum in the CHM gene of Danish and Swedish choroideremia patients. Human Molecular Genetics, 1994, 3, 1047-1051.	1.4	69
117	Impaired epithelial differentiation of induced pluripotent stem cells from ectodermal dysplasia-related patients is rescued by the small compound APR-246/PRIMA-1 ^{MET}. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 2152-2156.	3.3	69
118	Meier-Gorlin syndrome: Report of eight additional cases and review. American Journal of Medical Genetics Part A, 2001, 102, 115-124.	2.4	68
119	Mutant p63 causes defective expansion of ectodermal progenitor cells and impaired FGF signalling in AEC syndrome. EMBO Molecular Medicine, 2012, 4, 192-205.	3.3	68
120	Hippocampal dysfunction in the Euchromatin histone methyltransferase 1 heterozygous knockout mouse model for Kleefstra syndrome. Human Molecular Genetics, 2013, 22, 852-866.	1.4	68
121	Mouse Choroideremia Gene Mutation Causes Photoreceptor Cell Degeneration and is not Transmitted through the Female Germline. Human Molecular Genetics, 1997, 6, 851-858.	1.4	67
122	Human ISPD Is a Cytidyltransferase Required for Dystroglycan O-Mannosylation. Chemistry and Biology, 2015, 22, 1643-1652.	6.2	67
123	Non-specific X-linked semidominant mental retardation by mutations in a Rab GDP-dissociation inhibitor. Human Molecular Genetics, 1998, 7, 1311-1315.	1.4	65
124	p63 control of desmosome gene expression and adhesion is compromised in AEC syndrome. Human Molecular Genetics, 2013, 22, 531-543.	1.4	65
125	Reduced Euchromatin histone methyltransferase 1 causes developmental delay, hypotonia, and cranial abnormalities associated with increased bone gene expression in Kleefstra syndrome mice. Developmental Biology, 2014, 386, 395-407.	0.9	65
126	Zinc finger 81 (ZNF81) mutations associated with X-linked mental retardation. Journal of Medical Genetics, 2004, 41, 394-399.	1.5	63

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127	Plexin D1 Expression Is Induced on Tumor Vasculature and Tumor Cells: A Novel Target for Diagnosis and Therapy?. Cancer Research, 2005, 65, 8317-8323.	0.4	63
128	GATAD2Bloss-of-function mutations cause a recognisable syndrome with intellectual disability and are associated with learning deficits and synaptic undergrowth inDrosophila. Journal of Medical Genetics, 2013, 50, 507-514.	1.5	63
129	Human neuronal networks on micro-electrode arrays are a highly robust tool to study disease-specific genotype-phenotype correlations inÂvitro. Stem Cell Reports, 2021, 16, 2182-2196.	2.3	63
130	Nail-Patella Syndrome. Journal of the American Society of Nephrology: JASN, 2000, 11, 1762-1766.	3.0	62
131	Human syndromes with congenital patellar anomalies and the underlying gene defects. Clinical Genetics, 2005, 68, 302-319.	1.0	61
132	Deâ€ƒnovo MECP2 frameshift mutation in a boy with moderate mental retardation, obesity and gynaecomastia. Clinical Genetics, 2002, 61, 359-362.	1.0	59
133	Genetic players in esophageal atresia and tracheoesophageal fistula. Current Opinion in Genetics and Development, 2005, 15, 341-347.	1.5	59
134	Regulation of vitamin metabolism by p53 and p63 in development and cancer. Cell Cycle, 2010, 9, 2749-2757.	1.3	59
135	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
136	Novel mutations in LRP6 highlight the role of WNT signaling in tooth agenesis. Genetics in Medicine, 2016, 18, 1158-1162.	1.1	58
137	Bi-allelic Variants in METTL5 Cause Autosomal-Recessive Intellectual Disability and Microcephaly. American Journal of Human Genetics, 2019, 105, 869-878.	2.6	58
138	In-frame deletion inMECP2 causes mild nonspecific mental retardation. American Journal of Medical Genetics Part A, 2002, 107, 81-83.	2.4	56
139	MCT8 mutation analysis and identification of the first female with Allanâ€“Herndonâ€“Dudley syndrome due to loss of MCT8 expression. European Journal of Human Genetics, 2008, 16, 1029-1037.	1.4	56
140	Differential altered stability and transcriptional activity of Î”Np63 mutants in distinct ectodermal dysplasias. Journal of Cell Science, 2011, 124, 2200-2207.	1.2	56
141	Disruptions of the novel KIAA1202 gene are associated with X-linked mental retardation. Human Genetics, 2006, 118, 578-590.	1.8	55
142	An etiologic regulatory mutation in IRF6 with loss- and gain-of-function effects. Human Molecular Genetics, 2014, 23, 2711-2720.	1.4	55
143	X-linked mental retardation associated with cleft lip/palate maps to Xp11.3-q21.3. , 1999, 85, 216-220.		53
144	A novel translation re-initiation mechanism for the p63 gene revealed by amino-terminal truncating mutations in Rapp-Hodgkin/Hay-Wells-like syndromes. Human Molecular Genetics, 2008, 17, 1968-1977.	1.4	53

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145	Molecular evidence that AEC syndrome and Rapp-Hodgkin syndrome are variable expression of a single genetic disorder. Clinical Genetics, 2004, 66, 79-80.	1.0	52
146	Exome sequencing and CRISPR/Cas genome editing identify mutations of ZAK as a cause of limb defects in humans and mice. Genome Research, 2016, 26, 183-191.	2.4	52
147	Comparison of 12 Reference Genes for Normalization of Gene Expression Levels in Epstein-Barr Virus-Transformed Lymphoblastoid Cell Lines and Fibroblasts. Molecular Diagnosis and Therapy, 2006, 10, 197-204.	1.6	51
148	High resolution profiling of X chromosomal aberrations by array comparative genomic hybridisation. Journal of Medical Genetics, 2004, 41, 425-432.	1.5	50
149	CDK19 is disrupted in a female patient with bilateral congenital retinal folds, microcephaly and mild mental retardation. Human Genetics, 2010, 128, 281-291.	1.8	50
150	Spectrum of p63 mutations in a selected patient cohort affected with ankyloblepharonâ€ectodermal defectsâ€cleft lip/palate syndrome (AEC). American Journal of Medical Genetics, Part A, 2009, 149A, 1948-1951.	0.7	49
151	Disruption of the epigenetic code: An emerging mechanism in mental retardation. Neurobiology of Disease, 2010, 39, 3-12.	2.1	49
152	TAp63 Is Important for Cardiac Differentiation of Embryonic Stem Cells and Heart Development. Stem Cells, 2011, 29, 1672-1683.	1.4	49
153	Mutation screening in 86 known X-linked mental retardation genes by droplet-based multiplex PCR and massive parallel sequencing. The HUGO Journal, 2009, 3, 41-49.	4.1	48
154	Low frequency of MECP2 mutations in mentally retarded males. European Journal of Human Genetics, 2002, 10, 487-490.	1.4	46
155	Loss of SLC38A5 and FTSJ1 at Xp11.23 in three brothers with non-syndromic mental retardation due to a microdeletion in an unstable genomic region. Human Genetics, 2007, 121, 539-547.	1.8	46
156	Tooth agenesis and orofacial clefting: genetic brothers in arms?. Human Genetics, 2016, 135, 1299-1327.	1.8	46
157	De novo and inherited mutations in the X-linked gene CLCN4 are associated with syndromic intellectual disability and behavior and seizure disorders in males and females. Molecular Psychiatry, 2018, 23, 222-230.	4.1	45
158	Distinct Pathogenic Genes Causing Intellectual Disability and Autism Exhibit a Common Neuronal Network Hyperactivity Phenotype. Cell Reports, 2020, 30, 173-186.e6.	2.9	44
159	Expanding phenotype ofXNP mutations: Mild to moderate mental retardation. American Journal of Medical Genetics Part A, 2002, 110, 243-247.	2.4	43
160	The role of chromatin repressive marks in cognition and disease: A focus on the repressive complex GLP/G9a. Neurobiology of Learning and Memory, 2015, 124, 88-96.	1.0	43
161	Genotype-phenotype studies in three families with mutations in the polyglutamine-binding protein 1 gene (PQBP1). Clinical Genetics, 2004, 66, 318-326.	1.0	42
162	Elevated microRNA-181c and microRNA-30d levels in the enlarged amygdala of the valproic acid rat model of autism. Neurobiology of Disease, 2015, 80, 42-53.	2.1	42

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163	Imbalanced autophagy causes synaptic deficits in a human model for neurodevelopmental disorders. Autophagy, 2022, 18, 423-442.	4.3	42
164	MECP2 analysis in mentally retarded patients: implications for routine DNA diagnostics. European Journal of Human Genetics, 2004, 12, 24-28.	1.4	41
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