Hans Van Bokhoven

List of Publications by Year in Descending Order

308 papers	20,787 citations	81 h-index	132 g-index
325 ext. papers	23,684 ext. citations	8.4 avg, IF	6.12 L-index

#	Paper	IF	Citations
308	Generation of induced pluripotent stem cell lines carrying monoallelic (UCSFi001-A-60) or biallelic (UCSFi001-A-61; UCSFi001-A-62) frameshift variants in CACNA1A using CRISPR/Cas9.. *Stem Cell Research*, **2022**, 61, 102730	1.6	
307	Loss-of-function variants in the schizophrenia risk gene SETD1A alter neuronal network activity in human neurons through the cAMP/PKA pathway.. *Cell Reports*, **2022**, 39, 110790	10.6	1
306	The complexities of CACNA1A in clinical neurogenetics. *Journal of Neurology*, **2021**, 1	5.5	1
305	Biallelic variants in TMEM222 cause a new autosomal recessive neurodevelopmental disorder. *Genetics in Medicine*, **2021**, 23, 1246-1254	8.1	0
304	Human Induced Pluripotent Stem Cell-Based Modelling of Spinocerebellar Ataxias. *Stem Cell Reviews and Reports*, **2021**, 1	7.3	0
303	Cadherin-13 is a critical regulator of GABAergic modulation in human stem-cell-derived neuronal networks. *Molecular Psychiatry*, **2021**,	15.1	16
302	De novo and bi-allelic variants in AP1G1 cause neurodevelopmental disorder with developmental delay, intellectual disability, and epilepsy. *American Journal of Human Genetics*, **2021**, 108, 1330-1341	11	3
301	Systemic cell therapy for muscular dystrophies : The ultimate transplantable muscle progenitor cell and current challenges for clinical efficacy. *Stem Cell Reviews and Reports*, **2021**, 17, 878-899	7.3	3
300	Surveillance and prevalence of fragile X syndrome in Indonesia. *Intractable and Rare Diseases Research*, **2021**, 10, 11-16	1.4	1
299	Bi-allelic variants in the ER quality-control mannosidase gene EDEM3 cause a congenital disorder of glycosylation. *American Journal of Human Genetics*, **2021**, 108, 1342-1349	11	1
298	Analysis of a Set of KDM5C Regulatory Genes Mutated in Neurodevelopmental Disorders Identifies Temporal Coexpression Brain Signatures. *Genes*, **2021**, 12,	4.2	2
297	Imbalanced autophagy causes synaptic deficits in a human model for neurodevelopmental disorders. *Autophagy*, **2021**, 1-20	10.2	8
296	Human neuronal networks on micro-electrode arrays are a highly robust tool to study disease-specific genotype-phenotype correlations in vitro. *Stem Cell Reports*, **2021**, 16, 2182-2196	8	13
295	De Novo Variants in CNOT1, a Central Component of the CCR4-NOT Complex Involved in Gene Expression and RNA and Protein Stability, Cause Neurodevelopmental Delay. *American Journal of Human Genetics*, **2020**, 107, 164-172	11	13
294	Distinct Pathogenic Genes Causing Intellectual Disability and Autism Exhibit a Common Neuronal Network Hyperactivity Phenotype. *Cell Reports*, **2020**, 30, 173-186.e6	10.6	21
293	The phenomenal epigenome in neurodevelopmental disorders. *Human Molecular Genetics*, **2020**, 29, R42-R50	5.6	9
292	Pathogenic variant in NFIX gene affecting three sisters due to paternal mosaicism. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2731-2736	2.5	1

291	Neuronal network dysfunction in a model for Kleefstra syndrome mediated by enhanced NMDAR signaling. *Nature Communications*, **2019**, 10, 4928	17.4	42
290	Bi-allelic Variants in METTL5 Cause Autosomal-Recessive Intellectual Disability and Microcephaly. *American Journal of Human Genetics*, **2019**, 105, 869-878	11	33
289	Mutations in GDF11 and the extracellular antagonist, Follistatin, as a likely cause of Mendelian forms of orofacial clefting in humans. *Human Mutation*, **2019**, 40, 1813-1825	4.7	14
288	Phenotypic spectrum associated with a CRADD founder variant underlying frontotemporal predominant pachygyria in the Finnish population. *European Journal of Human Genetics*, **2019**, 27, 1235-1243	5.3	8
287	Deletions and loss-of-function variants in TP63 associated with orofacial clefting. *European Journal of Human Genetics*, **2019**, 27, 1101-1112	5.3	7
286	A de novo variant in the X-linked gene CNKSR2 is associated with seizures and mild intellectual disability in a female patient. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e00861	2.3	9
285	Cytidine Diphosphate-Ribitol Analysis for Diagnostics and Treatment Monitoring of Cytidine Diphosphate-l-Ribitol Pyrophosphorylase A Muscular Dystrophy. *Clinical Chemistry*, **2019**, 65, 1295-1306	5.5	8
284	Mutations in PIGU Impair the Function of the GPI Transamidase Complex, Causing Severe Intellectual Disability, Epilepsy, and Brain Anomalies. *American Journal of Human Genetics*, **2019**, 105, 395-402	11	27
283	Intrinsic Myogenic Potential of Skeletal Muscle-Derived Pericytes from Patients with Myotonic Dystrophy Type 1. *Molecular Therapy - Methods and Clinical Development*, **2019**, 15, 120-132	6.4	2
282	Homozygous Missense Variants in NTNG2, Encoding a Presynaptic Netrin-G2 Adhesion Protein, Lead to a Distinct Neurodevelopmental Disorder. *American Journal of Human Genetics*, **2019**, 105, 1048-1056	11	13
281	Histone demethylase KDM5C is a SAHA-sensitive central hub at the crossroads of transcriptional axes involved in multiple neurodevelopmental disorders. *Human Molecular Genetics*, **2019**, 28, 4089-4102	5.6	8
280	Duplication of 1q31.3q41 in two affected siblings due to paternal insertional translocation. *BMJ Case Reports*, **2019**, 12,	0.9	
279	Genetic architecture of subcortical brain structures in 38,851 individuals. *Nature Genetics*, **2019**, 51, 1624-1636	30.9	81
278	Loss of function of SVBP leads to autosomal recessive intellectual disability, microcephaly, ataxia, and hypotonia. *Genetics in Medicine*, **2019**, 21, 1790-1796	8.1	13
277	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	8.1	10
276	Increased H3K9 methylation and impaired expression of Protocadherins are associated with the cognitive dysfunctions of the Kleefstra syndrome. *Nucleic Acids Research*, **2018**, 46, 4950-4965	20.1	22
275	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	15.1	20
274	Transcriptome Analysis Identifies Multifaceted Regulatory Mechanisms Dictating a Genetic Switch from Neuronal Network Establishment to Maintenance During Postnatal Prefrontal Cortex Development. *Cerebral Cortex*, **2018**, 28, 833-851	5.1	11

273	Biallelic variants in LINGO1 are associated with autosomal recessive intellectual disability, microcephaly, speech and motor delay. *Genetics in Medicine*, **2018**, 20, 778-784	8.1	16
272	mosaicism in apparently unaffected parents is associated with autism spectrum disorder and neurocognitive dysfunction. *Molecular Autism*, **2018**, 9, 5	6.5	9
271	Inhibitory control of the excitatory/inhibitory balance in psychiatric disorders. *F1000Research*, **2018**, 7, 23	3.6	86
270	Mutant p63 Affects Epidermal Cell Identity through Rewiring the Enhancer Landscape. *Cell Reports*, **2018**, 25, 3490-3503.e4	10.6	25
269	Variants in PUS7 Cause Intellectual Disability with Speech Delay, Microcephaly, Short Stature, and Aggressive Behavior. *American Journal of Human Genetics*, **2018**, 103, 1045-1052	11	51
268	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	11	52
267	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. *American Journal of Human Genetics*, **2018**, 103, 144-153	11	18
266	MicroRNA-338 Attenuates Cortical Neuronal Outgrowth by Modulating the Expression of Axon Guidance Genes. *Molecular Neurobiology*, **2017**, 54, 3439-3452	6.2	19
265	The schizophrenia risk gene MIR137 acts as a hippocampal gene network node orchestrating the expression of genes relevant to nervous system development and function. *Progress in Neuro-Psychopharmacology and Biological Psychiatry*, **2017**, 73, 109-118	5.5	21
264	Novel genetic loci associated with hippocampal volume. *Nature Communications*, **2017**, 8, 13624	17.4	173
263	Haploinsufficiency of EHMT1 improves pattern separation and increases hippocampal cell proliferation. *Scientific Reports*, **2017**, 7, 40284	4.9	14
262	Altered expression of circadian rhythm and extracellular matrix genes in the medial prefrontal cortex of a valproic acid rat model of autism. *Progress in Neuro-Psychopharmacology and Biological Psychiatry*, **2017**, 77, 128-132	5.5	9
261	MicroRNA-338 modulates cortical neuronal placement and polarity. *RNA Biology*, **2017**, 14, 905-913	4.8	7
260	Adaptive and maladaptive functioning in Kleefstra syndrome compared to other rare genetic disorders with intellectual disabilities. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1821-1830	2.5	19
259	Duplicated Enhancer Region Increases Expression of CTSB and Segregates with Keratolytic Winter Erythema in South African and Norwegian Families. *American Journal of Human Genetics*, **2017**, 100, 737-750	11	19
258	Sleep Disturbance as a Precursor of Severe Regression in Kleefstra Syndrome Suggests a Need for Firm and Rapid Pharmacological Treatment. *Clinical Neuropharmacology*, **2017**, 40, 185-188	1.4	8
257	Identification of a de novo variant in CHUK in a patient with an EEC/AEC syndrome-like phenotype and hypogammaglobulinemia. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1813-1820	2.5	5
256	The Context of Symptom Measures: Interpretation and Clinical Diagnosis of Autism Spectrum Disorders in Intellectual Disabilities. *Journal of the American Academy of Child and Adolescent Psychiatry*, **2017**, 56, 618-619	7.2	8

255	Autosomal recessive transmission of a rare HOXC13 variant causes pure hair and nail ectodermal dysplasia. *Clinical and Experimental Dermatology*, **2017**, 42, 585-589	1.8	3
254	220th ENMC workshop: Dystroglycan and the dystroglycanopathies Naarden, The Netherlands, 27-29 May 2016. *Neuromuscular Disorders*, **2017**, 27, 387-395	2.9	6
253	Mutations in two large pedigrees highlight the role of ZNF711 in X-linked intellectual disability. *Gene*, **2017**, 605, 92-98	3.8	17
252	Functional convergence of histone methyltransferases EHMT1 and KMT2C involved in intellectual disability and autism spectrum disorder. *PLoS Genetics*, **2017**, 13, e1006864	6	67
251	B3GALNT2 mutations associated with non-syndromic autosomal recessive intellectual disability reveal a lack of genotype-phenotype associations in the muscular dystrophy-dystroglycanopathies. *Genome Medicine*, **2017**, 9, 118	14.4	9
250	Drosophila Courtship Conditioning As a Measure of Learning and Memory. *Journal of Visualized Experiments*, **2017**,	1.6	20
249	Homozygous Truncating Variants in TBC1D23 Cause Pontocerebellar Hypoplasia and Alter Cortical Development. *American Journal of Human Genetics*, **2017**, 101, 428-440	11	31
248	Novel IRF6 Mutations Detected in Orofacial Cleft Patients by Targeted Massively Parallel Sequencing. *Journal of Dental Research*, **2017**, 96, 179-185	8.1	10
247	Exome sequencing of Pakistani consanguineous families identifies 30 novel candidate genes for recessive intellectual disability. *Molecular Psychiatry*, **2017**, 22, 1604-1614	15.1	69
246	Integrated transcriptional analysis unveils the dynamics of cellular differentiation in the developing mouse hippocampus. *Scientific Reports*, **2017**, 7, 18073	4.9	5
245	Missense variants in AIMP1 gene are implicated in autosomal recessive intellectual disability without neurodegeneration. *European Journal of Human Genetics*, **2016**, 24, 392-9	5.3	14
244	Euchromatin histone methyltransferase 1 regulates cortical neuronal network development. *Scientific Reports*, **2016**, 6, 35756	4.9	24
243	ARHGAP12 Functions as a Developmental Brake on Excitatory Synapse Function. *Cell Reports*, **2016**, 14, 1355-1368	10.6	15
242	Perinatal reduction of functional serotonin transporters results in developmental delay. *Neuropharmacology*, **2016**, 109, 96-111	5.5	17
241	X-exome sequencing of 405 unresolved families identifies seven novel intellectual disability genes. *Molecular Psychiatry*, **2016**, 21, 133-48	15.1	167
240	Novel mutations in LRP6 highlight the role of WNT signaling in tooth agenesis. *Genetics in Medicine*, **2016**, 18, 1158-1162	8.1	45
239	MicroRNA-181 promotes synaptogenesis and attenuates axonal outgrowth in cortical neurons. *Cellular and Molecular Life Sciences*, **2016**, 73, 3555-67	10.3	31
238	Genetic influences on schizophrenia and subcortical brain volumes: large-scale proof of concept. *Nature Neuroscience*, **2016**, 19, 420-431	25.5	163

237	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. *American Journal of Human Genetics*, **2016**, 98, 541-552	11	89
236	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, 183-91	9.7	37
235	Histone Methylation by the Kleefstra Syndrome Protein EHMT1 Mediates Homeostatic Synaptic Scaling. *Neuron*, **2016**, 91, 341-55	13.9	47
234	Novel genetic loci underlying human intracranial volume identified through genome-wide association. *Nature Neuroscience*, **2016**, 19, 1569-1582	25.5	147
233	Tooth agenesis and orofacial clefting: genetic brothers in arms?. *Human Genetics*, **2016**, 135, 1299-1327	6.3	31
232	Common genetic variants influence human subcortical brain structures. *Nature*, **2015**, 520, 224-9	50.4	601
231	Variants in CUL4B are associated with cerebral malformations. *Human Mutation*, **2015**, 36, 106-17	4.7	28
230	Homozygous SLC6A17 mutations cause autosomal-recessive intellectual disability with progressive tremor, speech impairment, and behavioral problems. *American Journal of Human Genetics*, **2015**, 96, 386-96	11	16
229	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	3.1	35
228	MicroRNA-137 Controls AMPA-Receptor-Mediated Transmission and mGluR-Dependent LTD. *Cell Reports*, **2015**, 11, 1876-84	10.6	62
227	Gene regulatory mechanisms orchestrated by p63 in epithelial development and related disorders. *Biochimica Et Biophysica Acta - Gene Regulatory Mechanisms*, **2015**, 1849, 590-600	6	17
226	Absence of □and □dystroglycan is associated with Walker-Warburg syndrome. *Neurology*, **2015**, 84, 2177-82	6.5	29
225	Clinical Variability in a Family with an Ectodermal Dysplasia Syndrome and a Nonsense Mutation in the TP63 Gene. *Fetal and Pediatric Pathology*, **2015**, 34, 400-6	1.7	4
224	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	6.3	62
223	A de novo microdeletion in NRXN1 in a Dutch patient with mild intellectual disability, microcephaly and gonadal dysgenesis. *Genetical Research*, **2015**, 97, e19	1.1	
222	Transcription factor p63 bookmarks and regulates dynamic enhancers during epidermal differentiation. *EMBO Reports*, **2015**, 16, 863-78	6.5	89
221	Long-term consequences of chronic fluoxetine exposure on the expression of myelination-related genes in the rat hippocampus. *Translational Psychiatry*, **2015**, 5, e642	8.6	15
220	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	7.5	35

219	De novo mutations in PLXND1 and REV3L cause Möbius syndrome. *Nature Communications*, **2015**, 6, 7199	17.4	50
218	Human ISPD Is a Cytidyltransferase Required for Dystroglycan O-Mannosylation. *Chemistry and Biology*, **2015**, 22, 1643-52		53
217	Disease mutations in CMP-sialic acid transporter SLC35A1 result in abnormal α-dystroglycan O-mannosylation, independent from sialic acid. *Human Molecular Genetics*, **2015**, 24, 2241-6	5.6	26
216	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	3.8	95
215	Mutations Affecting the SAND Domain of DEAF1 Cause Intellectual Disability with Severe Speech Impairment and Behavioral Problems. *American Journal of Human Genetics*, **2015**, 96, 178	11	78
214	CLPB mutations cause 3-methylglutaconic aciduria, progressive brain atrophy, intellectual disability, congenital neutropenia, cataracts, movement disorder. *American Journal of Human Genetics*, **2015**, 96, 245-57	11	84
213	An etiologic regulatory mutation in IRF6 with loss- and gain-of-function effects. *Human Molecular Genetics*, **2014**, 23, 2711-20	5.6	39
212	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	3.1	49
211	The genetics of cognitive epigenetics. *Neuropharmacology*, **2014**, 80, 83-94	5.5	61
210	A complex microcephaly syndrome in a Pakistani family associated with a novel missense mutation in RBBP8 and a heterozygous deletion in NRXN1. *Gene*, **2014**, 538, 30-5	3.8	8
209	A genome-wide search for quantitative trait loci affecting the cortical surface area and thickness of Heschl's gyrus. *Genes, Brain and Behavior*, **2014**, 13, 675-85	3.6	26
208	Involvement of the kinesin family members KIF4A and KIF5C in intellectual disability and synaptic function. *Journal of Medical Genetics*, **2014**, 51, 487-94	5.8	60
207	Development and Developmental Disorders of the Cerebral Cortex **2014**, 523-642		1
206	Clinical assessment of five patients with BRWD3 mutation at Xq21.1 gives further evidence for mild to moderate intellectual disability and macrocephaly. *European Journal of Medical Genetics*, **2014**, 57, 200-6	2.6	12
205	Mutations affecting the SAND domain of DEAF1 cause intellectual disability with severe speech impairment and behavioral problems. *American Journal of Human Genetics*, **2014**, 94, 649-61	11	51
204	Exome sequencing identifies three novel candidate genes implicated in intellectual disability. *PLoS ONE*, **2014**, 9, e112687	3.7	16
203	A mutation in TP63 causing a mild ectodermal dysplasia phenotype. *Journal of Investigative Dermatology*, **2014**, 134, 2277-2280	4.3	2
202	Identifying genes responsible for intellectual disability in consanguineous families. *Human Heredity*, **2014**, 77, 150-60	1.1	20

201	Mutations in CSPP1 lead to classical Joubert syndrome. *American Journal of Human Genetics*, **2014**, 94, 80-6	11	62
200	A 3-base pair deletion, c.9711_9713del, in DMD results in intellectual disability without muscular dystrophy. *European Journal of Human Genetics*, **2014**, 22, 480-5	5.3	26
199	Causes of Congenital Malformations **2014**, 105-164		1
198	Identification of pathogenic gene variants in small families with intellectually disabled siblings by exome sequencing. *Journal of Medical Genetics*, **2013**, 50, 802-11	5.8	70
197	Homozygous and heterozygous disruptions of ANK3: at the crossroads of neurodevelopmental and psychiatric disorders. *Human Molecular Genetics*, **2013**, 22, 1960-70	5.6	108
196	Genomic approaches for studying craniofacial disorders. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2013**, 163C, 218-31	3.1	24
195	Hippocampal dysfunction in the Euchromatin histone methyltransferase 1 heterozygous knockout mouse model for Kleefstra syndrome. *Human Molecular Genetics*, **2013**, 22, 852-66	5.6	59
194	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, 844-9	5.3	18
193	Mutations in MED12 cause X-linked Ohdo syndrome. *American Journal of Human Genetics*, **2013**, 92, 401-6	11	67
192	Mutations in GDP-mannose pyrophosphorylase B cause congenital and limb-girdle muscular dystrophies associated with hypoglycosylation of □dystroglycan. *American Journal of Human Genetics*, **2013**, 93, 29-41	11	162
191	A novel homozygous 10 nucleotide deletion in BBS10 causes Bardet-Biedl syndrome in a Pakistani family. *Gene*, **2013**, 519, 177-81	3.8	10
190	Exome sequencing overrides formal genetics: ASPM mutations in a case study of apparent X-linked microcephalic intellectual deficit. *Clinical Genetics*, **2013**, 83, 288-90	4	8
189	Deciphering the glycosylome of dystroglycanopathies using haploid screens for lassa virus entry. *Science*, **2013**, 340, 479-83	33.3	229
188	Missense mutations in □1,3-N-acetylglucosaminyltransferase 1 (B3GNT1) cause Walker-Warburg syndrome. *Human Molecular Genetics*, **2013**, 22, 1746-54	5.6	118
187	Human intellectual disability genes form conserved functional modules in Drosophila. *PLoS Genetics*, **2013**, 9, e1003911	6	32
186	p63 control of desmosome gene expression and adhesion is compromised in AEC syndrome. *Human Molecular Genetics*, **2013**, 22, 531-43	5.6	56
185	APR-246/PRIMA-1(MET) 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, 2157-62	11.5	31
184	Role of p63 and the Notch pathway in cochlea development and sensorineural deafness. *Proceedings of the National Academy of Sciences of the United States of America*, **2013**, 110, 7300-5	11.5	31

183	GATAD2B loss-of-function mutations cause a recognisable syndrome with intellectual disability and are associated with learning deficits and synaptic undergrowth in Drosophila. *Journal of Medical Genetics*, **2013**, 50, 507-14	5.8	51
182	Impaired epithelial differentiation of induced pluripotent stem cells from ectodermal dysplasia-related patients is rescued by the small compound APR-246/PRIMA-1MET. *Proceedings of the National Academy of Sciences of the United States of America*, **2013**, 110, 2152-6	11.5	54
181	Phosphoribosylpyrophosphate synthetase superactivity and recurrent infections is caused by a p.Val142Leu mutation in PRS-I. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 455-60	2.5	22
180	MicroRNA networks direct neuronal development and plasticity. *Cellular and Molecular Life Sciences* , **2012**, 69, 89-102	10.3	173
179	Mutations in DDHD2, encoding an intracellular phospholipase A(1), cause a recessive form of complex hereditary spastic paraplegia. *American Journal of Human Genetics*, **2012**, 91, 1073-81	11	128
178	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, 312-6	6.6	7
177	Disruption of an EHMT1-associated chromatin-modification module causes intellectual disability. *American Journal of Human Genetics*, **2012**, 91, 73-82	11	165
176	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, 1122-7	11	60
175	Interpretation of clinical relevance of X-chromosome copy number variations identified in a large cohort of individuals with cognitive disorders and/or congenital anomalies. *European Journal of Medical Genetics*, **2012**, 55, 586-98	2.6	12
174	Mutant p63 causes defective expansion of ectodermal progenitor cells and impaired FGF signalling in AEC syndrome. *EMBO Molecular Medicine*, **2012**, 4, 192-205	12	62
173	Mutations in DYNC1H1 cause severe intellectual disability with neuronal migration defects. *Journal of Medical Genetics*, **2012**, 49, 179-83	5.8	131
172	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	36.3	147
171	Mutations in ISPD cause Walker-Warburg syndrome and defective glycosylation of □dystroglycan. *Nature Genetics*, **2012**, 44, 581-5	36.3	168
170	Genetic and epigenetic networks in intellectual disabilities. *Annual Review of Genetics*, **2011**, 45, 81-104	14.5	267
169	Heterozygous mutations of FREM1 are associated with an increased risk of isolated metopic craniosynostosis in humans and mice. *PLoS Genetics*, **2011**, 7, e1002278	6	67
168	Mutations in the pre-replication complex cause Meier-Gorlin syndrome. *Nature Genetics*, **2011**, 43, 356-9	36.3	186
167	Homozygosity mapping in outbred families with mental retardation. *European Journal of Human Genetics*, **2011**, 19, 597-601	5.3	23
166	A novel deletion mutation in proteoglycan-4 underlies camptodactyly-arthropathy-coxa-vara-pericarditis syndrome in a consanguineous pakistani family. *Archives of Medical Research*, **2011**, 42, 110-4	6.6	13

165	TAp63 is important for cardiac differentiation of embryonic stem cells and heart development. *Stem Cells*, **2011**, 29, 1672-83	5.8	38
164	Comprehensive genetic analysis of OEIS complex reveals no evidence for a recurrent microdeletion or duplication. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 38-49	2.5	13
163	A novel Xp22.11 deletion causing a syndrome of craniosynostosis and periventricular nodular heterotopia. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 3144-7	2.5	5
162	Characterization of a novel transcript of the EHMT1 gene reveals important diagnostic implications for Kleefstra syndrome. *Human Mutation*, **2011**, 32, 853-9	4.7	10
161	Chromosome 1p21.3 microdeletions comprising DPYD and MIR137 are associated with intellectual disability. *Journal of Medical Genetics*, **2011**, 48, 810-8	5.8	129
160	Epigenetic regulation of learning and memory by Drosophila EHMT/G9a. *PLoS Biology*, **2011**, 9, e1000569	9.7	153
159	Loss of the BMP antagonist, SMOC-1, causes Ophthalmo-acromelic (Waardenburg Anophthalmia) syndrome in humans and mice. *PLoS Genetics*, **2011**, 7, e1002114	6	67
158	Autosomal recessive dilated cardiomyopathy due to DOLK mutations results from abnormal dystroglycan O-mannosylation. *PLoS Genetics*, **2011**, 7, e1002427	6	111
157	Differential altered stability and transcriptional activity of ΔNp63 mutants in distinct ectodermal dysplasias. *Journal of Cell Science*, **2011**, 124, 2200-7	5.3	46
156	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, 429-35	5.3	83
155	Deletions encompassing 1q41q42.1 and clinical features of autosomal dominant Robinow syndrome. *Clinical Genetics*, **2010**, 77, 404-7	4	14
154	A homozygous FKRP start codon mutation is associated with Walker-Warburg syndrome, the severe end of the clinical spectrum. *Clinical Genetics*, **2010**, 78, 275-81	4	27
153	Cooperation between the transcription factors p63 and IRF6 is essential to prevent cleft palate in mice. *Journal of Clinical Investigation*, **2010**, 120, 1561-9	15.9	111
152	Correlation of enzyme activity and clinical phenotype in POMT1-associated dystroglycanopathies. *Neurology*, **2010**, 74, 157-64	6.5	29
151	Genome-wide profiling of p63 DNA-binding sites identifies an element that regulates gene expression during limb development in the 7q21 SHFM1 locus. *PLoS Genetics*, **2010**, 6, e1001065	6	142
150	A novel cerebello-ocular syndrome with abnormal glycosylation due to abnormalities in dolichol metabolism. *Brain*, **2010**, 133, 3210-20	11.2	76
149	Feedback regulators of hypoxia-inducible factors and their role in cancer biology. *Cell Cycle*, **2010**, 9, 2749-63	4.7	52
148	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	3.4	104

147	Embryonic stem cells as an ectodermal cellular model of human p63-related dysplasia syndromes. *Biochemical and Biophysical Research Communications*, **2010**, 395, 131-5	3.4	12
146	SRD5A3 is required for converting polyprenol to dolichol and is mutated in a congenital glycosylation disorder. *Cell*, **2010**, 142, 203-17	56.2	207
145	WNT5A mutations in patients with autosomal dominant Robinow syndrome. *Developmental Dynamics*, **2010**, 239, 327-37	2.9	171
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4	Cadherin-13 is a critical regulator of GABAergic modulation in human stem cell derived neuronal networks		1

3	Distinct pathogenic genes causing intellectual disability and autism exhibit overlapping effects on neuronal network development	1
2	Neuronal network dysfunction in a human model for Kleefstra syndrome mediated by enhanced NMDAR signaling	1
1	Loss-of-function variants in the schizophrenia risk gene SETD1A alter neuronal network activity in human neurons through cAMP/PKA pathway	1