Hans Van Bokhoven

List of Publications by Citations

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	Mutations in the gene encoding the 3P5PDNA exonuclease TREX1 cause Aicardi-Goutiﾭes syndrome at the AGS1 locus. *Nature Genetics*, **2006**, 38, 917-20	36.3	633
307	Common genetic variants influence human subcortical brain structures. *Nature*, **2015**, 520, 224-9	50.4	601
306	Heterozygous germline mutations in the p53 homolog p63 are the cause of EEC syndrome. *Cell*, **1999**, 99, 143-53	56.2	577
305	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, 1033-43	11	573
304	A systematic, large-scale resequencing screen of X-chromosome coding exons in mental retardation. *Nature Genetics*, **2009**, 41, 535-43	36.3	454
303	POMT2 mutations cause alpha-dystroglycan hypoglycosylation and Walker-Warburg syndrome. *Journal of Medical Genetics*, **2005**, 42, 907-12	5.8	317
302	Mutations in ARHGEF6, encoding a guanine nucleotide exchange factor for Rho GTPases, in patients with X-linked mental retardation. *Nature Genetics*, **2000**, 26, 247-50	36.3	306
301	Loss-of-function mutations in euchromatin histone methyl transferase 1 (EHMT1) cause the 9q34 subtelomeric deletion syndrome. *American Journal of Human Genetics*, **2006**, 79, 370-7	11	288
300	p63 Gene mutations in eec syndrome, limb-mammary syndrome, and isolated split hand-split foot malformation suggest a genotype-phenotype correlation. *American Journal of Human Genetics*, **2001**, 69, 481-92	11	288
299	Impaired glycosylation and cutis laxa caused by mutations in the vesicular H+-ATPase subunit ATP6V0A2. *Nature Genetics*, **2008**, 40, 32-4	36.3	279
298	Hay-Wells syndrome is caused by heterozygous missense mutations in the SAM domain of p63. *Human Molecular Genetics*, **2001**, 10, 221-9	5.6	268
297	Genetic and epigenetic networks in intellectual disabilities. *Annual Review of Genetics*, **2011**, 45, 81-104	14.5	267
296	Transcription factor SOX3 is involved in X-linked mental retardation with growth hormone deficiency. *American Journal of Human Genetics*, **2002**, 71, 1450-5	11	238
295	Deciphering the glycosylome of dystroglycanopathies using haploid screens for lassa virus entry. *Science*, **2013**, 340, 479-83	33.3	229
294	p63-associated disorders. *Cell Cycle*, **2007**, 6, 262-8	4.7	214
293	ARX, a novel Prd-class-homeobox gene highly expressed in the telencephalon, is mutated in X-linked mental retardation. *Human Molecular Genetics*, **2002**, 11, 981-91	5.6	209
292	Mutation of the gene encoding the ROR2 tyrosine kinase causes autosomal recessive Robinow syndrome. *Nature Genetics*, **2000**, 25, 423-6	36.3	208

291	SRD5A3 is required for converting polyprenol to dolichol and is mutated in a congenital glycosylation disorder. *Cell*, **2010**, 142, 203-17	56.2	207
290	Dominant isolated renal magnesium loss is caused by misrouting of the Na(+),K(+)-ATPase gamma-subunit. *Nature Genetics*, **2000**, 26, 265-6	36.3	205
289	Homozygous mutation in SPATA16 is associated with male infertility in human globozoospermia. *American Journal of Human Genetics*, **2007**, 81, 813-20	11	204
288	Mutations in the FKRP gene can cause muscle-eye-brain disease and Walker-Warburg syndrome. *Journal of Medical Genetics*, **2004**, 41, e61	5.8	197
287	A new gene involved in X-linked mental retardation identified by analysis of an X;2 balanced translocation. *Nature Genetics*, **2000**, 24, 167-70	36.3	197
286	Complex transcriptional effects of p63 isoforms: identification of novel activation and repression domains. *Molecular and Cellular Biology*, **2002**, 22, 8659-68	4.8	193
285	Mutations in the pre-replication complex cause Meier-Gorlin syndrome. *Nature Genetics*, **2011**, 43, 356-9	36.3	186
284	Novel genetic loci associated with hippocampal volume. *Nature Communications*, **2017**, 8, 13624	17.4	173
283	MicroRNA networks direct neuronal development and plasticity. *Cellular and Molecular Life Sciences* , **2012**, 69, 89-102	10.3	173
282	WNT5A mutations in patients with autosomal dominant Robinow syndrome. *Developmental Dynamics*, **2010**, 239, 327-37	2.9	171
281	Splitting p63. *American Journal of Human Genetics*, **2002**, 71, 1-13	11	169
280	Mutations in ISPD cause Walker-Warburg syndrome and defective glycosylation of □ystroglycan. *Nature Genetics*, **2012**, 44, 581-5	36.3	168
279	X-exome sequencing of 405 unresolved families identifies seven novel intellectual disability genes. *Molecular Psychiatry*, **2016**, 21, 133-48	15.1	167
278	Disruption of an EHMT1-associated chromatin-modification module causes intellectual disability. *American Journal of Human Genetics*, **2012**, 91, 73-82	11	165
277	Mutations in PHF8 are associated with X linked mental retardation and cleft lip/cleft palate. *Journal of Medical Genetics*, **2005**, 42, 780-6	5.8	165
276	A C-terminal inhibitory domain controls the activity of p63 by an intramolecular mechanism. *Molecular and Cellular Biology*, **2002**, 22, 8601-11	4.8	165
275	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, 432-43	11	164
274	Genetic influences on schizophrenia and subcortical brain volumes: large-scale proof of concept. *Nature Neuroscience*, **2016**, 19, 420-431	25.5	163

273	Mutations in GDP-mannose pyrophosphorylase B cause congenital and limb-girdle muscular dystrophies associated with hypoglycosylation of α-dystroglycan. *American Journal of Human Genetics*, **2013**, 93, 29-41	11	162
272	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	5.8	156
271	OFD1 is mutated in X-linked Joubert syndrome and interacts with LCA5-encoded lebercilin. *American Journal of Human Genetics*, **2009**, 85, 465-81	11	153
270	Epigenetic regulation of learning and memory by Drosophila EHMT/G9a. *PLoS Biology*, **2011**, 9, e1000569	9.7	153
269	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
268	Novel genetic loci underlying human intracranial volume identified through genome-wide association. *Nature Neuroscience*, **2016**, 19, 1569-1582	25.5	147
267	Mutations in different components of FGF signaling in LADD syndrome. *Nature Genetics*, **2006**, 38, 414-7	36.3	144
266	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
265	Pathogenesis of split-hand/split-foot malformation. *Human Molecular Genetics*, **2003**, 12 Spec No 1, R51-60	5.6	137
264	Mutations in DYNC1H1 cause severe intellectual disability with neuronal migration defects. *Journal of Medical Genetics*, **2012**, 49, 179-83	5.8	131
263	The p63 gene in EEC and other syndromes. *Journal of Medical Genetics*, **2002**, 39, 377-81	5.8	130
262	Chromosome 1p21.3 microdeletions comprising DPYD and MIR137 are associated with intellectual disability. *Journal of Medical Genetics*, **2011**, 48, 810-8	5.8	129
261	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
260	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	5.8	128
259	MYCN haploinsufficiency is associated with reduced brain size and intestinal atresias in Feingold syndrome. *Nature Genetics*, **2005**, 37, 465-7	36.3	126
258	Mutations in the polyglutamine binding protein 1 gene cause X-linked mental retardation. *Nature Genetics*, **2003**, 35, 313-5	36.3	125
257	A homozygous nonsense mutation in the fukutin gene causes a Walker-Warburg syndrome phenotype. *Journal of Medical Genetics*, **2003**, 40, 845-8	5.8	121
256	Missense mutations in β1,3-N-acetylglucosaminyltransferase 1 (B3GNT1) cause Walker-Warburg syndrome. *Human Molecular Genetics*, **2013**, 22, 1746-54	5.6	118

255	Pattern of p63 mutations and their phenotypes--update. *American Journal of Medical Genetics, Part A*, **2006**, 140, 1396-406	2.5	117
254	Mutations in the human TBX4 gene cause small patella syndrome. *American Journal of Human Genetics*, **2004**, 74, 1239-48	11	115
253	Cloning and characterization of the human choroideremia gene. *Human Molecular Genetics*, **1994**, 3, 1041-6	5.6	115
252	Intragenic deletion in the LARGE gene causes Walker-Warburg syndrome. *Human Genetics*, **2007**, 121, 685-90	6.3	113
251	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, 390-5	11	113
250	Missense mutation in PAK3, R67C, causes X-linked nonspecific mental retardation. *American Journal of Medical Genetics Part A*, **2000**, 93, 294-8		112
249	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
248	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, 444-53	5.3	111
247	Autosomal recessive dilated cardiomyopathy due to DOLK mutations results from abnormal dystroglycan O-mannosylation. *PLoS Genetics*, **2011**, 7, e1002427	6	111
246	Homozygous and heterozygous disruptions of ANK3: at the crossroads of neurodevelopmental and psychiatric disorders. *Human Molecular Genetics*, **2013**, 22, 1960-70	5.6	108
245	Mutations in the p53 homolog p63: allele-specific developmental syndromes in humans. *Trends in Molecular Medicine*, **2002**, 8, 133-9	11.5	105
244	The Opitz syndrome gene product, MID1, associates with microtubules. *Proceedings of the National Academy of Sciences of the United States of America*, **1999**, 96, 2794-9	11.5	105
243	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
242	Genotype-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, 935-46	5.3	104
241	P63 gene mutations and human developmental syndromes. *American Journal of Medical Genetics Part A*, **2002**, 112, 284-90		101
240	Mutations in the FTSJ1 gene coding for a novel S-adenosylmethionine-binding protein cause nonsyndromic X-linked mental retardation. *American Journal of Human Genetics*, **2004**, 75, 305-9	11	99
239	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
238	The expanding phenotype of POMT1 mutations: from Walker-Warburg syndrome to congenital muscular dystrophy, microcephaly, and mental retardation. *Human Mutation*, **2006**, 27, 453-9	4.7	91

237	A novel ribosomal S6-kinase (RSK4; RPS6KA6) is commonly deleted in patients with complex X-linked mental retardation. *Genomics*, **1999**, 62, 332-43	4.3	91
236	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. *American Journal of Human Genetics*, **2016**, 98, 541-552	11	89
235	Transcription factor p63 bookmarks and regulates dynamic enhancers during epidermal differentiation. *EMBO Reports*, **2015**, 16, 863-78	6.5	89
234	Mutation frequencies of X-linked mental retardation genes in families from the EuroMRX consortium. *Human Mutation*, **2007**, 28, 207-8	4.7	89
233	Inhibitory control of the excitatory/inhibitory balance in psychiatric disorders. *F1000Research*, **2018**, 7, 23	3.6	86
232	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
231	Glyc-O-genetics of Walker-Warburg syndrome. *Clinical Genetics*, **2005**, 67, 281-9	4	84
230	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
229	Gain-of-function mutation in ADULT syndrome reveals the presence of a second transactivation domain in p63. *Human Molecular Genetics*, **2002**, 11, 799-804	5.6	83
228	Genetic architecture of subcortical brain structures in 38,851 individuals. *Nature Genetics*, **2019**, 51, 1624-1636	36.3	81
227	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, 2257-65	5.6	80
226	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
225	Chromosomal copy number changes in patients with non-syndromic X linked mental retardation detected by array CGH. *Journal of Medical Genetics*, **2006**, 43, 362-70	5.8	78
224	A novel cerebello-ocular syndrome with abnormal glycosylation due to abnormalities in dolichol metabolism. *Brain*, **2010**, 133, 3210-20	11.2	76
223	Analysis of the p63 gene in classical EEC syndrome, related syndromes, and non-syndromic orofacial clefts. *Journal of Medical Genetics*, **2002**, 39, 559-66	5.8	76
222	PLEXIN-D1, a novel plexin family member, is expressed in vascular endothelium and the central nervous system during mouse embryogenesis. *Developmental Dynamics*, **2002**, 225, 336-43	2.9	75
221	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, 1341-54	11	75
220	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, 538-46	11	75

219	Genetic and epigenetic defects in mental retardation. *International Journal of Biochemistry and Cell Biology*, **2009**, 41, 96-107	5.6	74
218	PRPS1 mutations: four distinct syndromes and potential treatment. *American Journal of Human Genetics*, **2010**, 86, 506-18	11	74
217	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
216	Aicardi-Goutières syndrome displays genetic heterogeneity with one locus (AGS1) on chromosome 3p21. *American Journal of Human Genetics*, **2000**, 67, 213-21	11	70
215	Exome sequencing of Pakistani consanguineous families identifies 30 novel candidate genes for recessive intellectual disability. *Molecular Psychiatry*, **2017**, 22, 1604-1614	15.1	69
214	Functional convergence of histone methyltransferases EHMT1 and KMT2C involved in intellectual disability and autism spectrum disorder. *PLoS Genetics*, **2017**, 13, e1006864	6	67
213	Mutations in MED12 cause X-linked Ohdo syndrome. *American Journal of Human Genetics*, **2013**, 92, 401-6	11	67
212	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
211	Loss of the BMP antagonist, SMOC-1, causes Ophthalmo-acromelic (Waardenburg Anophthalmia) syndrome in humans and mice. *PLoS Genetics*, **2011**, 7, e1002114	6	67
210	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, 254-61	11	67
209	ZNF674: a new kruppel-associated box-containing zinc-finger gene involved in nonsyndromic X-linked mental retardation. *American Journal of Human Genetics*, **2006**, 78, 265-78	11	67
208	Mutation spectrum in the CHM gene of Danish and Swedish choroideremia patients. *Human Molecular Genetics*, **1994**, 3, 1047-51	5.6	66
207	Arts syndrome is caused by loss-of-function mutations in PRPS1. *American Journal of Human Genetics*, **2007**, 81, 507-18	11	65
206	Genotype-phenotype correlations in MYCN-related Feingold syndrome. *Human Mutation*, **2008**, 29, 1125-32	4.7	64
205	MicroRNA-137 Controls AMPA-Receptor-Mediated Transmission and mGluR-Dependent LTD. *Cell Reports*, **2015**, 11, 1876-84	10.6	62
204	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
203	Mutations in CSPP1 lead to classical Joubert syndrome. *American Journal of Human Genetics*, **2014**, 94, 80-6	11	62
202	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

201	Feingold syndrome: clinical review and genetic mapping. *American Journal of Medical Genetics Part A*, **2003**, 122A, 294-300		62
200	The genetics of cognitive epigenetics. *Neuropharmacology*, **2014**, 80, 83-94	5.5	61
199	Mouse choroideremia gene mutation causes photoreceptor cell degeneration and is not transmitted through the female germline. *Human Molecular Genetics*, **1997**, 6, 851-8	5.6	61
198	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
197	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
196	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
195	Meier-Gorlin syndrome: report of eight additional cases and review. *American Journal of Medical Genetics Part A*, **2001**, 102, 115-24		58
194	p63 control of desmosome gene expression and adhesion is compromised in AEC syndrome. *Human Molecular Genetics*, **2013**, 22, 531-43	5.6	56
193	Nail-patella syndrome: identification of mutations in the LMX1B gene in Dutch families. *Journal of the American Society of Nephrology: JASN*, **2000**, 11, 1762-1766	12.7	56
192	Plexin D1 expression is induced on tumor vasculature and tumor cells: a novel target for diagnosis and therapy?. *Cancer Research*, **2005**, 65, 8317-23	10.1	55
191	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
190	De novo MECP2 frameshift mutation in a boy with moderate mental retardation, obesity and gynaecomastia. *Clinical Genetics*, **2002**, 61, 359-62	4	54
189	Human ISPD Is a Cytidyltransferase Required for Dystroglycan O-Mannosylation. *Chemistry and Biology*, **2015**, 22, 1643-52		53
188	Feedback regulators of hypoxia-inducible factors and their role in cancer biology. *Cell Cycle*, **2010**, 9, 2749-63	4.7	52
187	Trismus-pseudocamptodactyly syndrome is caused by recurrent mutation of MYH8. *American Journal of Medical Genetics, Part A*, **2006**, 140, 2387-93	2.5	52
186	Zinc finger 81 (ZNF81) mutations associated with X-linked mental retardation. *Journal of Medical Genetics*, **2004**, 41, 394-9	5.8	52
185	In-frame deletion in MECP2 causes mild nonspecific mental retardation. *American Journal of Medical Genetics Part A*, **2002**, 107, 81-3		52
184	Human syndromes with congenital patellar anomalies and the underlying gene defects. *Clinical Genetics*, **2005**, 68, 302-19	4	52

183	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
182	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
181	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
180	Genetic players in esophageal atresia and tracheoesophageal fistula. *Current Opinion in Genetics and Development*, **2005**, 15, 341-7	4.9	51
179	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
178	De novo mutations in PLXND1 and REV3L cause Mbius syndrome. *Nature Communications*, **2015**, 6, 7199	17.4	50
177	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
176	MCT8 mutation analysis and identification of the first female with Allan-Herndon-Dudley syndrome due to loss of MCT8 expression. *European Journal of Human Genetics*, **2008**, 16, 1029-37	5.3	49
175	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	4.5	48
174	Non-specific X-linked semidominant mental retardation by mutations in a Rab GDP-dissociation inhibitor. *Human Molecular Genetics*, **1998**, 7, 1311-5	5.6	48
173	Mutation screening in 86 known X-linked mental retardation genes by droplet-based multiplex PCR and massive parallel sequencing. *The HUGO Journal*, **2009**, 3, 41-9		47
172	CDK19 is disrupted in a female patient with bilateral congenital retinal folds, microcephaly and mild mental retardation. *Human Genetics*, **2010**, 128, 281-91	6.3	47
171	Histone Methylation by the Kleefstra Syndrome Protein EHMT1 Mediates Homeostatic Synaptic Scaling. *Neuron*, **2016**, 91, 341-55	13.9	47
170	Differential altered stability and transcriptional activity of Np63 mutants in distinct ectodermal dysplasias. *Journal of Cell Science*, **2011**, 124, 2200-7	5.3	46
169	Molecular evidence that AEC syndrome and Rapp-Hodgkin syndrome are variable expression of a single genetic disorder. *Clinical Genetics*, **2004**, 66, 79-80	4	46
168	Novel mutations in LRP6 highlight the role of WNT signaling in tooth agenesis. *Genetics in Medicine*, **2016**, 18, 1158-1162	8.1	45
167	Disruption of the epigenetic code: an emerging mechanism in mental retardation. *Neurobiology of Disease*, **2010**, 39, 3-12	7.5	45
166	Spectrum of p63 mutations in a selected patient cohort affected with ankyloblepharon-ectodermal defects-cleft lip/palate syndrome (AEC). *American Journal of Medical Genetics, Part A*, **2009**, 149A, 1948-51	2.5	44

165	Disruptions of the novel KIAA1202 gene are associated with X-linked mental retardation. *Human Genetics*, **2006**, 118, 578-90	6.3	44
164	Low frequency of MECP2 mutations in mentally retarded males. *European Journal of Human Genetics*, **2002**, 10, 487-90	5.3	44
163	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, 1968-77	5.6	43
162	Neuronal network dysfunction in a model for Kleefstra syndrome mediated by enhanced NMDAR signaling. *Nature Communications*, **2019**, 10, 4928	17.4	42
161	High resolution profiling of X chromosomal aberrations by array comparative genomic hybridisation. *Journal of Medical Genetics*, **2004**, 41, 425-32	5.8	42
160	X-linked mental retardation associated with cleft lip/palate maps to Xp11.3-q21.3 **1999**, 85, 216-220		42
159	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, 539-47	6.3	40
158	An etiologic regulatory mutation in IRF6 with loss- and gain-of-function effects. *Human Molecular Genetics*, **2014**, 23, 2711-20	5.6	39
157	MECP2 analysis in mentally retarded patients: implications for routine DNA diagnostics. *European Journal of Human Genetics*, **2004**, 12, 24-8	5.3	39
156	TAp63 is important for cardiac differentiation of embryonic stem cells and heart development. *Stem Cells*, **2011**, 29, 1672-83	5.8	38
155	Neurologic aspects of MECP2 gene duplication in male patients. *Pediatric Neurology*, **2009**, 41, 187-91	2.9	38
154	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
153	Expanding phenotype of XNP mutations: mild to moderate mental retardation. *American Journal of Medical Genetics Part A*, **2002**, 110, 243-7		37
152	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
151	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
150	Genotype-phenotype studies in three families with mutations in the polyglutamine-binding protein 1 gene (PQBP1). *Clinical Genetics*, **2004**, 66, 318-26	4	34
149	Bi-allelic Variants in METTL5 Cause Autosomal-Recessive Intellectual Disability and Microcephaly. *American Journal of Human Genetics*, **2019**, 105, 869-878	11	33
148	Expression of Rab small GTPases in epithelial Caco-2 cells: Rab21 is an apically located GTP-binding protein in polarised intestinal epithelial cells. *European Journal of Cell Biology*, **2000**, 79, 308-16	6.1	33

147	Human intellectual disability genes form conserved functional modules in Drosophila. *PLoS Genetics*, **2013**, 9, e1003911	6	32
146	MicroRNA-181 promotes synaptogenesis and attenuates axonal outgrowth in cortical neurons. *Cellular and Molecular Life Sciences*, **2016**, 73, 3555-67	10.3	31
145	Homozygous Truncating Variants in TBC1D23 Cause Pontocerebellar Hypoplasia and Alter Cortical Development. *American Journal of Human Genetics*, **2017**, 101, 428-440	11	31
144	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
143	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
142	The small patella syndrome: description of five cases from three families and examination of possible allelism with familial patella aplasia-hypoplasia and nail-patella syndrome. *Journal of Medical Genetics*, **2001**, 38, 209-14	5.8	31
141	Tooth agenesis and orofacial clefting: genetic brothers in arms?. *Human Genetics*, **2016**, 135, 1299-1327	6.3	31
140	Progressive intestinal, neurological and psychiatric problems in two adult males with cerebral creatine deficiency caused by an SLC6A8 mutation. *Clinical Genetics*, **2005**, 68, 379-81	4	30
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