Dagmar Wieczorek

List of Publications by Year in Descending Order

225 papers	12,049 citations	59 h-index	102 g-index
239 ext. papers	15,031 ext. citations	7.9 avg, IF	5.5 L-index

#	Paper	IF	Citations
225	Progenitor cells derived from gene-engineered human induced pluripotent stem cells as synthetic cancer cell alternatives for in vitro pharmacology.. *Biotechnology Journal*, **2022**, e2100693	5.6	1
224	NFB-03. Neurological manifestations in children and adolescents with Neurofibromatosis type 1 - Implications for management and surveillance. *Neuro-Oncology*, **2022**, 24, i128-i128	1	
223	Acute myeloid leukemia-induced functional inhibition of healthy CD34+ hematopoietic stem and progenitor cells. *Stem Cells*, **2021**, 39, 1270-1284	5.8	0
222	Profound inhibition of CD73-dependent formation of anti-inflammatory adenosine in B cells of SLE patients. *EBioMedicine*, **2021**, 73, 103616	8.8	3
221	A DNA repair disorder caused by de novo monoallelic DDB1 variants is associated with a neurodevelopmental syndrome. *American Journal of Human Genetics*, **2021**, 108, 749-756	11	1
220	ANKRD11 variants: KBG syndrome and beyond. *Clinical Genetics*, **2021**, 100, 187-200	4	4
219	Swarm Learning for decentralized and confidential clinical machine learning. *Nature*, **2021**, 594, 265-270	50.4	89
218	C2orf69 mutations disrupt mitochondrial function and cause a multisystem human disorder with recurring autoinflammation. *Journal of Clinical Investigation*, **2021**, 131,	15.9	2
217	Defining the phenotypical spectrum associated with variants in. *Journal of Medical Genetics*, **2021**, 58, 33-40	5.8	3
216	QRICH1 variants in Ververi-Brady syndrome-delineation of the genotypic and phenotypic spectrum. *Clinical Genetics*, **2021**, 99, 199-207	4	1
215	Defining the genotypic and phenotypic spectrum of X-linked MSL3-related disorder. *Genetics in Medicine*, **2021**, 23, 384-395	8.1	0
214	Pontocerebellar hypoplasia due to bi-allelic variants in MINPP1. *European Journal of Human Genetics*, **2021**, 29, 411-421	5.3	5
213	Academic application of Good Cell Culture Practice for induced pluripotent stem cells. *ALTEX: Alternatives To Animal Experimentation*, **2021**, 38, 595-614	4.3	4
212	Intellectual disability associated with craniofacial dysmorphism, cleft palate, and congenital heart defect due to a de novo MEIS2 mutation: A clinical longitudinal study. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1216-1221	2.5	1
211	Mutations in Are a Novel Cause of Galloway-Mowat Syndrome. *Journal of the American Society of Nephrology: JASN*, **2021**, 32, 580-596	12.7	3
210	Unique variants in CLCN3, encoding an endosomal anion/proton exchanger, underlie a spectrum of neurodevelopmental disorders. *American Journal of Human Genetics*, **2021**, 108, 1450-1465	11	0
209	Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. *Brain*, **2021**,	11.2	8

208	Early IFN-β signatures and persistent dysfunction are distinguishing features of NK cells in severe COVID-19. *Immunity*, **2021**, 54, 2650-2669.e14	32.3	31
207	The recurrent missense mutation p.(Arg367Trp) in YARS1 causes a distinct neurodevelopmental phenotype. *Journal of Molecular Medicine*, **2021**, 99, 1755-1768	5.5	0
206	Genetics of craniofacial malformations. *Seminars in Fetal and Neonatal Medicine*, **2021**, 26, 101290	3.7	
205	Biallelic variants in YRDC cause a developmental disorder with progeroid features. *Human Genetics*, **2021**, 140, 1679-1693	6.3	1
204	Case Report: Severe Neonatal Course in Paternally Derived Familial Hypocalciuric Hypercalcemia. *Frontiers in Endocrinology*, **2021**, 12, 700612	5.7	0
203	Nine newly identified individuals refine the phenotype associated with MYT1L mutations. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1021-1031	2.5	7
202	Characterization and application of electrically active neuronal networks established from human induced pluripotent stem cell-derived neural progenitor cells for neurotoxicity evaluation. *Stem Cell Research*, **2020**, 45, 101761	1.6	14
201	Seltene Tumorerkrankungen. *Onkologe*, **2020**, 26, 202-204	0.1	
200	SARS-CoV-2 targets neurons of 3D human brain organoids. *EMBO Journal*, **2020**, 39, e106230	13	206
199	Bi-allelic Variants in RALGAPA1 Cause Profound Neurodevelopmental Disability, Muscular Hypotonia, Infantile Spasms, and Feeding Abnormalities. *American Journal of Human Genetics*, **2020**, 106, 246-255	11	6
198	Choline transporter-like 1 deficiency causes a new type of childhood-onset neurodegeneration. *Brain*, **2020**, 143, 94-111	11.2	7
197	Longitudinal Multi-omics Analyses Identify Responses of Megakaryocytes, Erythroid Cells, and Plasmablasts as Hallmarks of Severe COVID-19. *Immunity*, **2020**, 53, 1296-1314.e9	32.3	109
196	Severe COVID-19 Is Marked by a Dysregulated Myeloid Cell Compartment. *Cell*, **2020**, 182, 1419-1440.e23	36.2	558
195	The () Promoter Polymorphisms (rs3063368, rs755622) Predict Acute Kidney Injury and Death after Cardiac Surgery. *Journal of Clinical Medicine*, **2020**, 9,	5.1	5
194	Novel EXOSC3 pathogenic variant results in a mild course of neurologic disease with cerebellum involvement. *European Journal of Medical Genetics*, **2020**, 63, 103649	2.6	5
193	POLR1B and neural crest cell anomalies in Treacher Collins syndrome type 4. *Genetics in Medicine*, **2020**, 22, 547-556	8.1	26
192	Fatal metabolic decompensation in carbonic anhydrase VA deficiency despite early treatment and control of hyperammonemia. *Genetics in Medicine*, **2020**, 22, 654-655	8.1	5
191	Fragile X mental retardation protein protects against tumour necrosis factor-mediated cell death and liver injury. *Gut*, **2020**, 69, 133-145	19.2	6

190	De novo variants in PAK1 lead to intellectual disability with macrocephaly and seizures. *Brain*, **2019**, 142, 3351-3359	11.2	9
189	Bi-allelic Variants in METTL5 Cause Autosomal-Recessive Intellectual Disability and Microcephaly. *American Journal of Human Genetics*, **2019**, 105, 869-878	11	33
188	Next-generation sequencing of 32 genes associated with hereditary aortopathies and related disorders of connective tissue in a cohort of 199 patients. *Genetics in Medicine*, **2019**, 21, 1832-1841	8.1	16
187	Moyamoya angiopathy in PHACE syndrome not associated with RNF213 variants. *Childs Nervous System*, **2019**, 35, 1231-1237	1.7	2
186	SON haploinsufficiency causes impaired pre-mRNA splicing of CAKUT genes and heterogeneous renal phenotypes. *Kidney International*, **2019**, 95, 1494-1504	9.9	9
185	Mutations in SMARCB1 and in other Coffin-Siris syndrome genes lead to various brain midline defects. *Nature Communications*, **2019**, 10, 2966	17.4	9
184	How I approach hereditary cancer predisposition in a child with cancer. *Pediatric Blood and Cancer*, **2019**, 66, e27916	3	3
183	Genome-Wide Analysis of the Nucleosome Landscape in Individuals with Coffin-Siris Syndrome. *Cytogenetic and Genome Research*, **2019**, 159, 1-11	1.9	3
182	Evolutionary conserved networks of human height identify multiple Mendelian causes of short stature. *European Journal of Human Genetics*, **2019**, 27, 1061-1071	5.3	7
181	De Novo Mutations Affecting the Catalytic C Subunit of PP2A, PPP2CA, Cause Syndromic Intellectual Disability Resembling Other PP2A-Related Neurodevelopmental Disorders. *American Journal of Human Genetics*, **2019**, 104, 139-156	11	18
180	UBQLN4 Represses Homologous Recombination and Is Overexpressed in Aggressive Tumors. *Cell*, **2019**, 176, 505-519.e22	56.2	68
179	Gene expression profiling in aggressive digital papillary adenocarcinoma sheds light on the architecture of a rare sweat gland carcinoma. *British Journal of Dermatology*, **2019**, 180, 1150-1160	4	13
178	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffin-Siris syndrome. *Genetics in Medicine*, **2019**, 21, 1295-1307	8.1	36
177	Family-based germline sequencing in children with cancer. *Oncogene*, **2019**, 38, 1367-1380	9.2	17
176	Genetics of intellectual disability in consanguineous families. *Molecular Psychiatry*, **2019**, 24, 1027-1039	15.1	79
175	The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant. *Annals of Neurology*, **2018**, 83, 926-934	9.4	11
174	Mutations in the BAF-Complex Subunit DPF2 Are Associated with Coffin-Siris Syndrome. *American Journal of Human Genetics*, **2018**, 102, 468-479	11	37
173	Genetic predisposition in children with cancer - affected families acceptance of Trio-WES. *European Journal of Pediatrics*, **2018**, 177, 53-60	4.1	20

172	Clinical relevance of systematic phenotyping and exome sequencing in patients with short stature. *Genetics in Medicine*, **2018**, 20, 630-638	8.1	68
171	Further evidence for complex inheritance of holoprosencephaly: Lessons learned from pre- and postnatal diagnostic testing in Germany. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2018**, 178, 198-205	3.1	2
170	BCL11B mutations in patients affected by a neurodevelopmental disorder with reduced type 2 innate lymphoid cells. *Brain*, **2018**, 141, 2299-2311	11.2	36
169	Angelman Syndrome-Affected Individual with a Numerically Normal Karyotype and Isodisomic Paternal Uniparental Disomy of Chromosome 15 due to Maternal Robertsonian Translocation (14;15) by Monosomy Rescue. *Cytogenetic and Genome Research*, **2018**,	1.9	1
168	De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. *American Journal of Human Genetics*, **2018**, 102, 1195-1203	11	24
167	Severe neurocognitive and growth disorders due to variation in THOC2, an essential component of nuclear mRNA export machinery. *Human Mutation*, **2018**, 39, 1126-1138	4.7	8
166	Distinctive facial features in idiopathic Moyamoya disease in Caucasians: a first systematic analysis. *PeerJ*, **2018**, 6, e4740	3.1	7
165	Genetik von Intelligenz und kognitiven St□ungen □ ein komplexes, aber relevantes Thema nicht nur f□ die Humangenetik. *Medizinische Genetik*, **2018**, 30, 305-305	0.5	
164	Autosomal dominant intellectual disability. *Medizinische Genetik*, **2018**, 30, 318-322	0.5	18
163	Penetrance and Expressivity in Inherited Cancer Predisposing Syndromes. *Trends in Cancer*, **2018**, 4, 718-728	12.5	21
162	Genetic variants in components of the NALCN-UNC80-UNC79 ion channel complex cause a broad clinical phenotype (NALCN channelopathies). *Human Genetics*, **2018**, 137, 753-768	6.3	19
161	WilmsRTumor 1 Gene Expression Using a Standardized European LeukemiaNet-Certified Assay Compared to Other Methods for Detection of Minimal Residual Disease in Myelodysplastic Syndrome and Acute Myelogenous Leukemia after Allogeneic Blood Stem Cell Transplantation. *Biology of Blood and Marrow Transplantation*, **2018**, 24, 2337-2343	4.7	36
160	De novo FBXO11 mutations are associated with intellectual disability and behavioural anomalies. *Human Genetics*, **2018**, 137, 401-411	6.3	20
159	Isolated PREPL deficiency associated with congenital myasthenic syndrome-22. *Klinische Padiatrie*, **2018**, 230, 281-283	0.9	6
158	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. *Human Mutation*, **2018**, 39, 1246-1261	4.7	21
157	Heterozygosity for ARID2 loss-of-function mutations in individuals with a Coffin-Siris syndrome-like phenotype. *Human Genetics*, **2017**, 136, 297-305	6.3	33
156	Diagnostic Yield and Novel Candidate Genes by Exome Sequencing in 152 Consanguineous Families With Neurodevelopmental Disorders. *JAMA Psychiatry*, **2017**, 74, 293-299	14.5	116
155	Mutations in chromatin regulators functionally link Cornelia de Lange syndrome and clinically overlapping phenotypes. *Human Genetics*, **2017**, 136, 307-320	6.3	40

154	Heterozygous HNRNPU variants cause early onset epilepsy and severe intellectual disability. *Human Genetics*, **2017**, 136, 821-834	6.3	39
153	Variants in CPLX1 in two families with autosomal-recessive severe infantile myoclonic epilepsy and ID. *European Journal of Human Genetics*, **2017**, 25, 889-893	5.3	19
152	Autosomal dominant frontometaphyseal dysplasia: Delineation of the clinical phenotype. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1739-1746	2.5	16
151	New insights into the imprinted MEG8-DMR in 14q32 and clinical and molecular description of novel patients with Temple syndrome. *European Journal of Human Genetics*, **2017**, 25, 935-945	5.3	21
150	Central nervous system anomalies in two females with Borjeson-Forssman-Lehmann syndrome. *Epilepsy and Behavior*, **2017**, 69, 104-109	3.2	4
149	Bainbridge-Ropers syndrome caused by loss-of-function variants in ASXL3: a recognizable condition. *European Journal of Human Genetics*, **2017**, 25, 183-191	5.3	28
148	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. *Nature Genetics*, **2017**, 49, 223-237	36.3	116
147	Autism spectrum disorder and Li-Fraumeni syndrome: purely coincidental or mechanistically associated?. *Molecular and Cellular Pediatrics*, **2017**, 4, 8	3.3	
146	Genetic screening confirms heterozygous mutations in ACAN as a major cause of idiopathic short stature. *Scientific Reports*, **2017**, 7, 12225	4.9	37
145	WDR26 Haploinsufficiency Causes a Recognizable Syndrome of Intellectual Disability, Seizures, Abnormal Gait, and Distinctive Facial Features. *American Journal of Human Genetics*, **2017**, 101, 139-148	11	31
144	Identification of new TRIP12 variants and detailed clinical evaluation of individuals with non-syndromic intellectual disability with or without autism. *Human Genetics*, **2017**, 136, 179-192	6.3	29
143	De novo microdeletions and point mutations affecting SOX2 in three individuals with intellectual disability but without major eye malformations. *American Journal of Medical Genetics, Part A*, **2017**, 173, 435-443	2.5	14
142	Hematopoietic Stem Cell Transplantation in an Infant with Immunodeficiency, Centromeric Instability, and Facial Anomaly Syndrome. *Frontiers in Immunology*, **2017**, 8, 773	8.4	10
141	Identification of causative variants in TXNL4A in Burn-McKeown syndrome and isolated choanal atresia. *European Journal of Human Genetics*, **2017**, 25, 1126-1133	5.3	7
140	Loss-of-function variants in HIVEP2 are a cause of intellectual disability. *European Journal of Human Genetics*, **2016**, 24, 556-61	5.3	28
139	Diagnostic algorithms in Charcot-Marie-Tooth neuropathies: experiences from a German genetic laboratory on the basis of 1206 index patients. *Clinical Genetics*, **2016**, 89, 34-43	4	48
138	De Novo Mutations in SON Disrupt RNA Splicing of Genes Essential for Brain Development and Metabolism, Causing an Intellectual-Disability Syndrome. *American Journal of Human Genetics*, **2016**, 99, 711-719	11	44
137	De novo nonsense and frameshift variants of TCF20 in individuals with intellectual disability and postnatal overgrowth. *European Journal of Human Genetics*, **2016**, 24, 1739-1745	5.3	20

136	ASPP2 deficiency causes features of 1q41q42 microdeletion syndrome. *Cell Death and Differentiation*, **2016**, 23, 1973-1984	12.7	5
135	Mutations in MAP3K7 that Alter the Activity of the TAK1 Signaling Complex Cause Frontometaphyseal Dysplasia. *American Journal of Human Genetics*, **2016**, 99, 392-406	11	34
134	Novel microdeletions on chromosome 14q32.2 suggest a potential role for non-coding RNAs in Kagami-Ogata syndrome. *European Journal of Human Genetics*, **2016**, 24, 1724-1729	5.3	19
133	RPA and Rad51 constitute a cell intrinsic mechanism to protect the cytosol from self DNA. *Nature Communications*, **2016**, 7, 11752	17.4	88
132	Genome-wide methylation analysis of retrocopy-associated CpG islands and their genomic environment. *Epigenetics*, **2016**, 11, 216-26	5.7	6
131	Identification and Functional Characterization of Two Intronic NIPBL Mutations in Two Patients with Cornelia de Lange Syndrome. *BioMed Research International*, **2016**, 2016, 8742939	3	11
130	Genetic Analysis of PAX6-Negative Individuals with Aniridia or Gillespie Syndrome. *PLoS ONE*, **2016**, 11, e0153757	3.7	34
129	Mutation Update for Kabuki Syndrome Genes KMT2D and KDM6A and Further Delineation of X-Linked Kabuki Syndrome Subtype 2. *Human Mutation*, **2016**, 37, 847-64	4.7	89
128	Expanding the clinical spectrum of the HDAC8-phenotype: implications for molecular diagnostics, counseling and risk prediction. *Clinical Genetics*, **2016**, 89, 564-73	4	29
127	X-linked recessive VACTERL-H due to a mutation in FANCB in a preterm boy. *Clinical Dysmorphology*, **2016**, 25, 73-6	0.9	8
126	Autosomal dominant spinal muscular atrophy with lower extremity predominance: A recognizable phenotype of BICD2 mutations. *Muscle and Nerve*, **2016**, 54, 496-500	3.4	18
125	A syndrome of microcephaly, short stature, polysyndactyly, and dental anomalies caused by a homozygous KATNB1 mutation. *American Journal of Medical Genetics, Part A*, **2016**, 170, 728-33	2.5	10
124	Tentative clinical diagnosis of Lujan-Fryns syndrome--A conglomeration of different genetic entities?. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 94-102	2.5	10
123	Exome sequencing unravels unexpected differential diagnoses in individuals with the tentative diagnosis of Coffin-Siris and Nicolaides-Baraitser syndromes. *Human Genetics*, **2015**, 134, 553-68	6.3	47
122	Infectious and immunologic phenotype of MECP2 duplication syndrome. *Journal of Clinical Immunology*, **2015**, 35, 168-81	5.7	27
121	Acrofacial Dysostosis, Cincinnati Type, a Mandibulofacial Dysostosis Syndrome with Limb Anomalies, Is Caused by POLR1A Dysfunction. *American Journal of Human Genetics*, **2015**, 96, 765-74	11	46
120	A review of craniofacial disorders caused by spliceosomal defects. *Clinical Genetics*, **2015**, 88, 405-15	4	60
119	Reconstruction of images from Gabor graphs with applications in facial image processing. *International Journal of Wavelets, Multiresolution and Information Processing*, **2015**, 13, 1550019	0.9	5

118	Splitting versus lumping: Temple-Baraitser and Zimmermann-Laband Syndromes. *Human Genetics*, **2015**, 134, 1089-97	6.3	17
117	De Novo Mutations in CHAMP1 Cause Intellectual Disability with Severe Speech Impairment. *American Journal of Human Genetics*, **2015**, 97, 493-500	11	55
116	Further delineation of the KAT6B molecular and phenotypic spectrum. *European Journal of Human Genetics*, **2015**, 23, 1165-70	5.3	45
115	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
114	Deficiency of ECHS1 causes mitochondrial encephalopathy with cardiac involvement. *Annals of Clinical and Translational Neurology*, **2015**, 2, 492-509	5.3	69
113	Neue syndromale Krankheitsbilder mit Mikrozephalie. *Medizinische Genetik*, **2015**, 27, 369-376	0.5	
112	Spinocerebellar ataxia 28: a novel AFG3L2 mutation in a German family with young onset, slow progression and saccadic slowing. *Cerebellum and Ataxias*, **2015**, 2, 19	1.7	21
111	WDR73 Mutations Cause Infantile Neurodegeneration and Variable Glomerular Kidney Disease. *Human Mutation*, **2015**, 36, 1021-8	4.7	31
110	3p14 deletion is a rare contiguous gene syndrome: report of 2 new patients and an overview of 14 patients. *American Journal of Medical Genetics, Part A*, **2015**, 167, 1223-30	2.5	12
109	Loss-of-function variants of SETD5 cause intellectual disability and the core phenotype of microdeletion 3p25.3 syndrome. *European Journal of Human Genetics*, **2015**, 23, 753-60	5.3	55
108	RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome. *Journal of Clinical Investigation*, **2015**, 125, 3585-99	15.9	48
107	Diagnostic approach to microcephaly in childhood: a two-center study and review of the literature. *Developmental Medicine and Child Neurology*, **2014**, 56, 732-41	3.3	123
106	Phenotype and genotype in Nicolaides-Baraitser syndrome. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2014**, 166C, 302-14	3.1	51
105	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, 285-93	11	82
104	Females with de novo aberrations in PHF6: clinical overlap of Borjeson-Forssman-Lehmann with Coffin-Siris syndrome. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2014**, 166C, 290-301	3.1	21
103	De novo CCND2 mutations leading to stabilization of cyclin D2 cause megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome. *Nature Genetics*, **2014**, 46, 510-515	36.3	100
102	Homozygous truncating PTPRF mutation causes athelia. *Human Genetics*, **2014**, 133, 1041-7	6.3	6
101	Genotype-phenotype correlation of Coffin-Siris syndrome caused by mutations in SMARCB1, SMARCA4, SMARCE1, and ARID1A. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2014**, 166C, 262-75	3.1	74

100	Classification and visualization based on derived image features: application to genetic syndromes. *PLoS ONE*, **2014**, 9, e109033	3.7	9
99	Microcephaly with or without chorioretinopathy, lymphoedema, or mental retardation (MCLMR): review of phenotype associated with KIF11 mutations. *European Journal of Human Genetics*, **2014**, 22, 881-7	5.3	51
98	DOORS syndrome: phenotype, genotype and comparison with Coffin-Siris syndrome. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2014**, 166C, 327-32	3.1	33
97	The ARID1B phenotype: what we have learned so far. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2014**, 166C, 276-89	3.1	62
96	Compound heterozygosity of low-frequency promoter deletions and rare loss-of-function mutations in TXNL4A causes Burn-McKeown syndrome. *American Journal of Human Genetics*, **2014**, 95, 698-707	11	45
95	Extreme growth failure is a common presentation of ligase IV deficiency. *Human Mutation*, **2014**, 35, 76-85	4.7	63
94	Delineation of PIGV mutation spectrum and associated phenotypes in hyperphosphatasia with mental retardation syndrome. *European Journal of Human Genetics*, **2014**, 22, 762-7	5.3	30
93	Homozygous and compound-heterozygous mutations in TGDS cause Catel-Manzke syndrome. *American Journal of Human Genetics*, **2014**, 95, 763-70	11	30
92	A patient with a de-novo deletion 3p25.3 and features overlapping with Rubinstein-Taybi syndrome. *Clinical Dysmorphology*, **2014**, 23, 67-70	0.9	1
91	The genetic basis of DOORS syndrome: an exome-sequencing study. *Lancet Neurology, The*, **2014**, 13, 44-58	24.1	96
90	Expanding the phenotype of IQSEC2 mutations: truncating mutations in severe intellectual disability. *European Journal of Human Genetics*, **2014**, 22, 289-92	5.3	33
89	Autosomal recessive POLR1D mutation with decrease of TCOF1 mRNA is responsible for Treacher Collins syndrome. *Genetics in Medicine*, **2014**, 16, 720-4	8.1	36
88	Behavioral phenotype in five individuals with de novo mutations within the GRIN2B gene. *Behavioral and Brain Functions*, **2013**, 9, 20	4.1	30
87	Oto-facial syndrome and esophageal atresia, intellectual disability and zygomatic anomalies - expanding the phenotypes associated with EFTUD2 mutations. *Orphanet Journal of Rare Diseases*, **2013**, 8, 110	4.2	39
86	Clinical and mutation data in 12 patients with the clinical diagnosis of Nager syndrome. *Human Genetics*, **2013**, 132, 885-98	6.3	54
85	Integrative analysis revealed the molecular mechanism underlying RBM10-mediated splicing regulation. *EMBO Molecular Medicine*, **2013**, 5, 1431-42	12	67
84	ZMYND10 is mutated in primary ciliary dyskinesia and interacts with LRRC6. *American Journal of Human Genetics*, **2013**, 93, 336-45	11	144
83	Platelet defects in congenital variant of Rett syndrome patients with FOXG1 mutations or reduced expression due to a position effect at 14q12. *European Journal of Human Genetics*, **2013**, 21, 1349-55	5.3	12

82	160 kb deletion in ISPD unmasking a recessive mutation in a patient with Walker-Warburg syndrome. *European Journal of Medical Genetics*, **2013**, 56, 689-94	2.6	14
81	Wide clinical variability in conditions with coarse facial features and hypertrichosis caused by mutations in ABCC9. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 295-300	2.5	20
80	Novel mutations including deletions of the entire OFD1 gene in 30 families with type 1 orofaciodigital syndrome: a study of the extensive clinical variability. *Human Mutation*, **2013**, 34, 237-47	4.7	35
79	Human facial dysostoses. *Clinical Genetics*, **2013**, 83, 499-510	4	51
78	A comprehensive molecular study on Coffin-Siris and Nicolaides-Baraitser syndromes identifies a broad molecular and clinical spectrum converging on altered chromatin remodeling. *Human Molecular Genetics*, **2013**, 22, 5121-35	5.6	138
77	A new face of Borjeson-Forssman-Lehmann syndrome? De novo mutations in PHF6 in seven females with a distinct phenotype. *Journal of Medical Genetics*, **2013**, 50, 838-47	5.8	36
76	Effects of RANK-ligand antibody (denosumab) treatment on bone turnover markers in a girl with juvenile Paget's disease. *Journal of Clinical Endocrinology and Metabolism*, **2013**, 98, 3121-6	5.6	78
75	Rare copy number variants are a common cause of short stature. *PLoS Genetics*, **2013**, 9, e1003365	6	50
74	X-linked intellectual disability type Nascimento is a clinically distinct, probably underdiagnosed entity. *Orphanet Journal of Rare Diseases*, **2013**, 8, 146	4.2	16
73	Microcephalic osteodysplastic primordial dwarfism type I with biallelic mutations in the RNU4ATAC gene. *Clinical Genetics*, **2012**, 82, 140-6	4	29
72	Range of genetic mutations associated with severe non-syndromic sporadic intellectual disability: an exome sequencing study. *Lancet, The*, **2012**, 380, 1674-82	40	765
71	Congenital diaphragmatic hernia interval on chromosome 8p23.1 characterized by genetics and protein interaction networks. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 3148-58	2.5	36
70	Treacher Collins syndrome: clinical implications for the paediatrician--a new mutation in a severely affected newborn and comparison with three further patients with the same mutation, and review of the literature. *European Journal of Pediatrics*, **2012**, 171, 1611-8	4.1	18
69	Heterozygous missense mutations in SMARCA2 cause Nicolaides-Baraitser syndrome. *Nature Genetics*, **2012**, 44, 445-9, S1	36.3	170
68	A noncoding, regulatory mutation implicates HCFC1 in nonsyndromic intellectual disability. *American Journal of Human Genetics*, **2012**, 91, 694-702	11	69
67	Mutations in SRCAP, encoding SNF2-related CREBBP activator protein, cause Floating-Harbor syndrome. *American Journal of Human Genetics*, **2012**, 90, 308-13	11	130
66	Haploinsufficiency of a spliceosomal GTPase encoded by EFTUD2 causes mandibulofacial dysostosis with microcephaly. *American Journal of Human Genetics*, **2012**, 90, 369-77	11	143
65	Haploinsufficiency of ARID1B, a member of the SWI/SNF-a chromatin-remodeling complex, is a frequent cause of intellectual disability. *American Journal of Human Genetics*, **2012**, 90, 565-72	11	182

64	Mutations in NSUN2 cause autosomal-recessive intellectual disability. *American Journal of Human Genetics*, **2012**, 90, 847-55	11	179
63	First Report of a Single Exon Deletion in TCOF1 Causing Treacher Collins Syndrome. *Molecular Syndromology*, **2012**, 2, 53-59	1.5	13
62	Miller (Genee-Wiedemann) syndrome represents a clinically and biochemically distinct subgroup of postaxial acrofacial dysostosis associated with partial deficiency of DHODH. *Human Molecular Genetics*, **2012**, 21, 3969-83	5.6	51
61	A Novel Homozygous WDR72 Mutation in Two Siblings with Amelogenesis Imperfecta and Mild Short Stature. *Molecular Syndromology*, **2012**, 3, 223-9	1.5	10
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