Dagmar Wieczorek

List of Publications by Citations

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	Range of genetic mutations associated with severe non-syndromic sporadic intellectual disability: an exome sequencing study. *Lancet, The*, **2012**, 380, 1674-82	40	765
224	Deep sequencing reveals 50 novel genes for recessive cognitive disorders. *Nature*, **2011**, 478, 57-63	50.4	649
223	Severe COVID-19 Is Marked by a Dysregulated Myeloid Cell Compartment. *Cell*, **2020**, 182, 1419-1440.e23	36.2	558
222	Germline KRAS and BRAF mutations in cardio-facio-cutaneous syndrome. *Nature Genetics*, **2006**, 38, 294-6	36.3	437
221	Mutations in GRIN2A and GRIN2B encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes. *Nature Genetics*, **2010**, 42, 1021-6	36.3	347
220	Mutations in genes encoding subunits of RNA polymerases I and III cause Treacher Collins syndrome. *Nature Genetics*, **2011**, 43, 20-2	36.3	239
219	SARS-CoV-2 targets neurons of 3D human brain organoids. *EMBO Journal*, **2020**, 39, e106230	13	206
218	De novo nonsense mutations in ASXL1 cause Bohring-Opitz syndrome. *Nature Genetics*, **2011**, 43, 729-31	36.3	198
217	CEP152 is a genome maintenance protein disrupted in Seckel syndrome. *Nature Genetics*, **2011**, 43, 23-6	36.3	185
216	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
215	Mutations in NSUN2 cause autosomal-recessive intellectual disability. *American Journal of Human Genetics*, **2012**, 90, 847-55	11	179
214	The core FOXG1 syndrome phenotype consists of postnatal microcephaly, severe mental retardation, absent language, dyskinesia, and corpus callosum hypogenesis. *Journal of Medical Genetics*, **2011**, 48, 396-406	5.8	179
213	Mutations in U4atac snRNA, a component of the minor spliceosome, in the developmental disorder MOPD I. *Science*, **2011**, 332, 238-40	33.3	179
212	Heterozygous missense mutations in SMARCA2 cause Nicolaides-Baraitser syndrome. *Nature Genetics*, **2012**, 44, 445-9, S1	36.3	170
211	Oculo-auriculo-vertebral spectrum (OAVS): clinical evaluation and severity scoring of 53 patients and proposal for a new classification. *European Journal of Medical Genetics*, **2005**, 48, 397-411	2.6	150
210	Expanding the phenotypic spectrum of lupus erythematosus in Aicardi-Goutières syndrome. *Arthritis and Rheumatism*, **2010**, 62, 1469-77		145
209	ZMYND10 is mutated in primary ciliary dyskinesia and interacts with LRRC6. *American Journal of Human Genetics*, **2013**, 93, 336-45	11	144

208	Haploinsufficiency of a spliceosomal GTPase encoded by EFTUD2 causes mandibulofacial dysostosis with microcephaly. *American Journal of Human Genetics*, **2012**, 90, 369-77	11	143
207	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
206	Mutations in SRCAP, encoding SNF2-related CREBBP activator protein, cause Floating-Harbor syndrome. *American Journal of Human Genetics*, **2012**, 90, 308-13	11	130
205	Spectrum of mutations in PTPN11 and genotype-phenotype correlation in 96 patients with Noonan syndrome and five patients with cardio-facio-cutaneous syndrome. *European Journal of Human Genetics*, **2003**, 11, 201-6	5.3	128
204	Genotyping in 46 patients with tentative diagnosis of Treacher Collins syndrome revealed unexpected phenotypic variation. *European Journal of Human Genetics*, **2004**, 12, 879-90	5.3	124
203	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
202	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
201	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. *Nature Genetics*, **2017**, 49, 223-237	36.3	116
200	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
199	A specific mutation in the distant sonic hedgehog (SHH) cis-regulator (ZRS) causes Werner mesomelic syndrome (WMS) while complete ZRS duplications underlie Haas type polysyndactyly and preaxial polydactyly (PPD) with or without triphalangeal thumb. *Human Mutation*, **2010**, 31, 81-9	4.7	104
198	Acetylcholine receptor pathway mutations explain various fetal akinesia deformation sequence disorders. *American Journal of Human Genetics*, **2008**, 82, 464-76	11	104
197	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
196	The genetic basis of DOORS syndrome: an exome-sequencing study. *Lancet Neurology, The*, **2014**, 13, 44-58	24.1	96
195	Swarm Learning for decentralized and confidential clinical machine learning. *Nature*, **2021**, 594, 265-270	50.4	89
194	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
193	RPA and Rad51 constitute a cell intrinsic mechanism to protect the cytosol from self DNA. *Nature Communications*, **2016**, 7, 11752	17.4	88
192	A paternal deletion of MKRN3, MAGEL2 and NDN does not result in Prader-Willi syndrome. *European Journal of Human Genetics*, **2009**, 17, 582-90	5.3	88
191	A mutation screen in patients with Kabuki syndrome. *Human Genetics*, **2011**, 130, 715-24	6.3	87

190	SOS1 is the second most common Noonan gene but plays no major role in cardio-facio-cutaneous syndrome. *Journal of Medical Genetics*, **2007**, 44, 651-6	5.8	85
189	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
188	A novel microdeletion syndrome involving 5q14.3-q15: clinical and molecular cytogenetic characterization of three patients. *European Journal of Human Genetics*, **2009**, 17, 1592-9	5.3	81
187	Mutation and phenotypic spectrum in patients with cardio-facio-cutaneous and Costello syndrome. *Clinical Genetics*, **2008**, 73, 62-70	4	81
186	Molecular and clinical characterization of cardio-facio-cutaneous (CFC) syndrome: overlapping clinical manifestations with Costello syndrome. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 799-807	2.5	79
185	Genetics of intellectual disability in consanguineous families. *Molecular Psychiatry*, **2019**, 24, 1027-1039	15.1	79
184	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
183	Molecular and clinical analysis of RAF1 in Noonan syndrome and related disorders: dephosphorylation of serine 259 as the essential mechanism for mutant activation. *Human Mutation*, **2010**, 31, 284-94	4.7	77
182	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
181	Deficiency of ECHS1 causes mitochondrial encephalopathy with cardiac involvement. *Annals of Clinical and Translational Neurology*, **2015**, 2, 492-509	5.3	69
180	A noncoding, regulatory mutation implicates HCFC1 in nonsyndromic intellectual disability. *American Journal of Human Genetics*, **2012**, 91, 694-702	11	69
179	Clinical relevance of systematic phenotyping and exome sequencing in patients with short stature. *Genetics in Medicine*, **2018**, 20, 630-638	8.1	68
178	A family with autosomal dominant oculo-auriculo-vertebral spectrum. *Clinical Dysmorphology*, **2007**, 16, 1-7	0.9	68
177	Further delineation of Kabuki syndrome in 48 well-defined new individuals **2005**, 132A, 265-72		68
176	UBQLN4 Represses Homologous Recombination and Is Overexpressed in Aggressive Tumors. *Cell*, **2019**, 176, 505-519.e22	56.2	68
175	Integrative analysis revealed the molecular mechanism underlying RBM10-mediated splicing regulation. *EMBO Molecular Medicine*, **2013**, 5, 1431-42	12	67
174	Genetic determination of human facial morphology: links between cleft-lips and normal variation. *European Journal of Human Genetics*, **2011**, 19, 1192-7	5.3	67
173	Loss of the BMP antagonist, SMOC-1, causes Ophthalmo-acromelic (Waardenburg Anophthalmia) syndrome in humans and mice. *PLoS Genetics*, **2011**, 7, e1002114	6	67

172	Extreme growth failure is a common presentation of ligase IV deficiency. *Human Mutation*, **2014**, 35, 76-85	4.7	63
171	Mutations in ZIC2 in human holoprosencephaly: description of a novel ZIC2 specific phenotype and comprehensive analysis of 157 individuals. *Journal of Medical Genetics*, **2010**, 47, 513-24	5.8	63
170	Syndrome identification based on 2D analysis software. *European Journal of Human Genetics*, **2006**, 14, 1082-9	5.3	63
169	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
168	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
167	A review of craniofacial disorders caused by spliceosomal defects. *Clinical Genetics*, **2015**, 88, 405-15	4	60
166	Is there a higher incidence of maternal uniparental disomy 14 [upd(14)mat]? Detection of 10 new patients by methylation-specific PCR. *American Journal of Medical Genetics, Part A*, **2006**, 140, 2039-49	2.5	57
165	Computer-based recognition of dysmorphic faces. *European Journal of Human Genetics*, **2003**, 11, 555-60	5.3	56
164	De Novo Mutations in CHAMP1 Cause Intellectual Disability with Severe Speech Impairment. *American Journal of Human Genetics*, **2015**, 97, 493-500	11	55
163	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
162	Histone acetylation dependent allelic expression imbalance of BAPX1 in patients with the oculo-auriculo-vertebral spectrum. *Human Molecular Genetics*, **2006**, 15, 581-7	5.6	55
161	Clinical and mutation data in 12 patients with the clinical diagnosis of Nager syndrome. *Human Genetics*, **2013**, 132, 885-98	6.3	54
160	Two patients with EP300 mutations and facial dysmorphism different from the classic Rubinstein-Taybi syndrome. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 181-4	2.5	52
159	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
158	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
157	Human facial dysostoses. *Clinical Genetics*, **2013**, 83, 499-510	4	51
156	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
155	Rare copy number variants are a common cause of short stature. *PLoS Genetics*, **2013**, 9, e1003365	6	50

154	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
153	RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome. *Journal of Clinical Investigation*, **2015**, 125, 3585-99	15.9	48
152	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
151	The face of Noonan syndrome: Does phenotype predict genotype. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1960-6	2.5	47
150	Novel mutations in BCOR in three patients with oculo-facio-cardio-dental syndrome, but none in Lenz microphthalmia syndrome. *European Journal of Human Genetics*, **2005**, 13, 563-9	5.3	47
149	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
148	Further delineation of the KAT6B molecular and phenotypic spectrum. *European Journal of Human Genetics*, **2015**, 23, 1165-70	5.3	45
147	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
146	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
145	Biallelic loss of function of the promyelocytic leukaemia zinc finger (PLZF) gene causes severe skeletal defects and genital hypoplasia. *Journal of Medical Genetics*, **2008**, 45, 731-7	5.8	43
144	Reproduction abnormalities and twin pregnancies in parents of sporadic patients with oculo-auriculo-vertebral spectrum/Goldenhar syndrome. *Human Genetics*, **2007**, 121, 369-76	6.3	41
143	Identification of 34 novel and 56 known FOXL2 mutations in patients with Blepharophimosis syndrome. *Human Mutation*, **2008**, 29, E205-19	4.7	41
142	Altered development of NKT cells, T cells, CD8 T cells and NK cells in a PLZF deficient patient. *PLoS ONE*, **2011**, 6, e24441	3.7	41
141	Mutations in chromatin regulators functionally link Cornelia de Lange syndrome and clinically overlapping phenotypes. *Human Genetics*, **2017**, 136, 307-320	6.3	40
140	Autosomal recessive mental retardation: homozygosity mapping identifies 27 single linkage intervals, at least 14 novel loci and several mutation hotspots. *Human Genetics*, **2011**, 129, 141-8	6.3	40
139	Heterozygous HNRNPU variants cause early onset epilepsy and severe intellectual disability. *Human Genetics*, **2017**, 136, 821-834	6.3	39
138	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
137	Bohring-Opitz (Oberklaid-Danks) syndrome: clinical study, review of the literature, and discussion of possible pathogenesis. *European Journal of Human Genetics*, **2011**, 19, 513-9	5.3	38

136	Mutations in the BAF-Complex Subunit DPF2 Are Associated with Coffin-Siris Syndrome. *American Journal of Human Genetics*, **2018**, 102, 468-479	11	37
135	Genetic screening confirms heterozygous mutations in ACAN as a major cause of idiopathic short stature. *Scientific Reports*, **2017**, 7, 12225	4.9	37
134	De novo MECP2 duplication in two females with random X-inactivation and moderate mental retardation. *European Journal of Human Genetics*, **2011**, 19, 507-12	5.3	37
133	BCL11B mutations in patients affected by a neurodevelopmental disorder with reduced type 2 innate lymphoid cells. *Brain*, **2018**, 141, 2299-2311	11.2	36
132	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
131	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
130	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
129	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffin-Siris syndrome. *Genetics in Medicine*, **2019**, 21, 1295-1307	8.1	36
128	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
127	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
126	Cardio-facio-cutaneous (CFC) syndrome--a distinct entity? Report of three patients demonstrating the diagnostic difficulties in delineation of CFC syndrome. *Clinical Genetics*, **1997**, 52, 37-46	4	35
125	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
124	Genetic Analysis of PAX6-NegativeRIndividuals with Aniridia or Gillespie Syndrome. *PLoS ONE*, **2016**, 11, e0153757	3.7	34
123	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
122	Bi-allelic Variants in METTL5 Cause Autosomal-Recessive Intellectual Disability and Microcephaly. *American Journal of Human Genetics*, **2019**, 105, 869-878	11	33
121	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
120	Expanding the phenotype of IQSEC2 mutations: truncating mutations in severe intellectual disability. *European Journal of Human Genetics*, **2014**, 22, 289-92	5.3	33
119	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

118	WDR73 Mutations Cause Infantile Neurodegeneration and Variable Glomerular Kidney Disease. *Human Mutation*, **2015**, 36, 1021-8	4.7	31
117	Early IFN-Ⅰ signatures and persistent dysfunction are distinguishing features of NK cells in severe COVID-19. *Immunity*, **2021**, 54, 2650-2669.e14	32.3	31
116	Behavioral phenotype in five individuals with de novo mutations within the GRIN2B gene. *Behavioral and Brain Functions*, **2013**, 9, 20	4.1	30
115	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
114	Homozygous and compound-heterozygous mutations in TGDS cause Catel-Manzke syndrome. *American Journal of Human Genetics*, **2014**, 95, 763-70	11	30
113	Automated syndrome detection in a set of clinical facial photographs. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 2161-9	2.5	30
112	Microcephalic osteodysplastic primordial dwarfism type I with biallelic mutations in the RNU4ATAC gene. *Clinical Genetics*, **2012**, 82, 140-6	4	29
111	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
110	Goltz-Gorlin (focal dermal hypoplasia) and the microphthalmia with linear skin defects (MLS) syndrome: no evidence of genetic overlap. *European Journal of Human Genetics*, **2009**, 17, 1207-15	5.3	29
109	Expanding the clinical spectrum of the ℞HDAC8-phenotype℞ implications for molecular diagnostics, counseling and risk prediction. *Clinical Genetics*, **2016**, 89, 564-73	4	29
108	Loss-of-function variants in HIVEP2 are a cause of intellectual disability. *European Journal of Human Genetics*, **2016**, 24, 556-61	5.3	28
107	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
106	Cohen syndrome diagnosis using whole genome arrays. *Journal of Medical Genetics*, **2011**, 48, 136-40	5.8	28
105	Infectious and immunologic phenotype of MECP2 duplication syndrome. *Journal of Clinical Immunology*, **2015**, 35, 168-81	5.7	27
104	POLR1B and neural crest cell anomalies in Treacher Collins syndrome type 4. *Genetics in Medicine*, **2020**, 22, 547-556	8.1	26
103	Microcephaly, microtia, preauricular tags, choanal atresia and developmental delay in three unrelated patients: a mandibulofacial dysostosis distinct from Treacher Collins syndrome. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 837-43	2.5	25
102	Genotype-phenotype correlation in eight new patients with a deletion encompassing 2q31.1. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1213-24	2.5	25
101	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

100	Impact of geometry and viewing angle on classification accuracy of 2D based analysis of dysmorphic faces. *European Journal of Medical Genetics*, **2008**, 51, 44-53	2.6	24
99	Left-ventricular non-compaction (LVNC): a clinical feature more often observed in terminal deletion 1p36 than previously expected. *European Journal of Medical Genetics*, **2008**, 51, 685-8	2.6	23
98	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
97	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
96	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
95	Penetrance and Expressivity in Inherited Cancer Predisposing Syndromes. *Trends in Cancer*, **2018**, 4, 718-728	12.9	21
94	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. *Human Mutation*, **2018**, 39, 1246-1261	4.7	21
93	Genetic predisposition in children with cancer - affected families' acceptance of Trio-WES. *European Journal of Pediatrics*, **2018**, 177, 53-60	4.1	20
92	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
91	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
90	Five patients with novel overlapping interstitial deletions in 8q22.2q22.3. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1857-64	2.5	20
89	De novo FBXO11 mutations are associated with intellectual disability and behavioural anomalies. *Human Genetics*, **2018**, 137, 401-411	6.3	20
88	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
87	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
86	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
85	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
84	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
83	De Novo Mutations Affecting the Catalytic Cβ 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

82	Autosomal dominant intellectual disability. *Medizinische Genetik*, **2018**, 30, 318-322	0.5	18
81	Splitting versus lumping: Temple-Baraitser and Zimmermann-Laband Syndromes. *Human Genetics*, **2015**, 134, 1089-97	6.3	17
80	Family-based germline sequencing in children with cancer. *Oncogene*, **2019**, 38, 1367-1380	9.2	17
79	Autosomal dominant frontometaphyseal dysplasia: Delineation of the clinical phenotype. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1739-1746	2.5	16
78	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
77	X-linked intellectual disability type Nascimento is a clinically distinct, probably underdiagnosed entity. *Orphanet Journal of Rare Diseases*, **2013**, 8, 146	4.2	16
76	FOXL2 copy number changes in the molecular pathogenesis of BPES: unique cohort of 17 deletions. *Human Mutation*, **2010**, 31, E1332-47	4.7	16
75	Esophageal atresia, hypoplasia of zygomatic complex, microcephaly, cup-shaped ears, congenital heart defect, and mental retardation--new MCA/MR syndrome in two affected sibs and a mildly affected mother?. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 1135-42	2.5	16
74	Two adults with Rubinstein-Taybi syndrome with mild mental retardation, glaucoma, normal growth and skull circumference, and camptodactyly of third fingers. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 2849-54	2.5	15
73	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
72	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
71	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
70	First Report of a Single Exon Deletion in TCOF1 Causing Treacher Collins Syndrome. *Molecular Syndromology*, **2012**, 2, 53-59	1.5	13
69	Severe nemaline myopathy associated with consecutive mutations E74D and H75Y on a single ACTA1 allele. *Neuromuscular Disorders*, **2009**, 19, 481-4	2.9	13
68	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
67	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
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