Dagmar Wieczorek

List of Publications by Year
in descending order

226	17,066	15503 65	19747 117
papers	citations	h-index	g-index

239	239	239	27034
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Severe COVID-19 Is Marked by a Dysregulated Myeloid Cell Compartment. Cell, 2020, 182, 1419-1440.e23.	28.9	1,162
2	Range of genetic mutations associated with severe non-syndromic sporadic intellectual disability: an exome sequencing study. Lancet, The, 2012, 380, 1674-1682.	13.7	940
3	Deep sequencing reveals 50 novel genes for recessive cognitive disorders. Nature, 2011, 478, 57-63.	27.8	805
4	Germline KRAS and BRAF mutations in cardio-facio-cutaneous syndrome. Nature Genetics, 2006, 38, 294-296.	21.4	517
5	Mutations in GRIN2A and GRIN2B encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes. Nature Genetics, 2010, 42, 1021-1026.	21.4	431
6	<scp>SARS</scp> â€CoVâ€2 targets neurons of 3D human brain organoids. EMBO Journal, 2020, 39, e106230.	7.8	401
7	Swarm Learning for decentralized and confidential clinical machine learning. Nature, 2021, 594, 265-270.	27.8	375
8	Mutations in genes encoding subunits of RNA polymerases I and III cause Treacher Collins syndrome. Nature Genetics, 2011, 43, 20-22.	21.4	308
9	Longitudinal Multi-omics Analyses Identify Responses of Megakaryocytes, Erythroid Cells, and Plasmablasts as Hallmarks of Severe COVID-19. Immunity, 2020, 53, 1296-1314.e9.	14.3	278
10	Mutations in NSUN2 Cause Autosomal- Recessive Intellectual Disability. American Journal of Human Genetics, 2012, 90, 847-855.	6.2	243
11	De novo nonsense mutations in ASXL1 cause Bohring-Opitz syndrome. Nature Genetics, 2011, 43, 729-731.	21.4	236
12	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, 565-572.	6.2	225
13	Mutations in U4atac snRNA, a Component of the Minor Spliceosome, in the Developmental Disorder MOPD I. Science, 2011, 332, 238-240.	12.6	223
14	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.	3.2	220
15	Heterozygous missense mutations in SMARCA2 cause Nicolaides-Baraitser syndrome. Nature Genetics, 2012, 44, 445-449.	21.4	207
16	CEP152 is a genome maintenance protein disrupted in Seckel syndrome. Nature Genetics, 2011, 43, 23-26.	21.4	201
17	A comprehensive molecular study on Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes identifies a broad molecular and clinical spectrum converging on altered chromatin remodeling. Human Molecular Genetics, 2013, 22, 5121-5135.	2.9	190
18	Diagnostic Yield and Novel Candidate Genes by Exome Sequencing in 152 Consanguineous Families With Neurodevelopmental Disorders. JAMA Psychiatry, 2017, 74, 293.	11.0	186

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19	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. Nature Genetics, 2017, 49, 223-237.	21.4	186
20	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.	1.3	184
21	Expanding the phenotypic spectrum of lupus erythematosus in Aicardiâ€GoutiÃ¨res syndrome. Arthritis and Rheumatism, 2010, 62, 1469-1477.	6.7	183
22	ZMYND10 Is Mutated in Primary Ciliary Dyskinesia and Interacts with LRRC6. American Journal of Human Genetics, 2013, 93, 336-345.	6.2	183
23	Haploinsufficiency of a Spliceosomal GTPase Encoded by EFTUD2 Causes Mandibulofacial Dysostosis with Microcephaly. American Journal of Human Genetics, 2012, 90, 369-377.	6.2	180
24	Diagnostic approach to microcephaly in childhood: a twoâ€center study and review of the literature. Developmental Medicine and Child Neurology, 2014, 56, 732-741.	2.1	176
25	Mutations in SRCAP, Encoding SNF2-Related CREBBP Activator Protein, Cause Floating-Harbor Syndrome. American Journal of Human Genetics, 2012, 90, 308-313.	6.2	157
26	Genotyping in 46 patients with tentative diagnosis of Treacher Collins syndrome revealed unexpected phenotypic variation. European Journal of Human Genetics, 2004, 12, 879-890.	2.8	149
27	Spectrum of mutations in PTPN11 and genotypeâ€“phenotype correlation in 96 patients with Noonan syndrome and five patients with cardio-facio-cutaneous syndrome. European Journal of Human Genetics, 2003, 11, 201-206.	2.8	148
28	Early IFN-Î± signatures and persistent dysfunction are distinguishing features of NK cells in severe COVID-19. Immunity, 2021, 54, 2650-2669.e14.	14.3	145
29	Mutation Update for Kabuki Syndrome GenesKMT2DandKDM6Aand Further Delineation of X-Linked Kabuki Syndrome Subtype 2. Human Mutation, 2016, 37, 847-864.	2.5	134
30	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, 81-89.	2.5	133
31	Genetics of intellectual disability in consanguineous families. Molecular Psychiatry, 2019, 24, 1027-1039.	7.9	131
32	RPA and Rad51 constitute a cell intrinsic mechanism to protect the cytosol from self DNA. Nature Communications, 2016, 7, 11752.	12.8	127
33	Acetylcholine Receptor Pathway Mutations Explain Various Fetal Akinesia Deformation Sequence Disorders. American Journal of Human Genetics, 2008, 82, 464-476.	6.2	124
34	De novo CCND2 mutations leading to stabilization of cyclin D2 cause megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome. Nature Genetics, 2014, 46, 510-515.	21.4	118
35	Genotypeâ€phenotype correlation of Coffinâ€Siris syndrome caused by mutations in SMARCB1, SMARCA4, SMARCE1, and ARID1A. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 262-275.	1.6	117
36	SOS1 is the second most common Noonan gene but plays no major role in cardio-facio-cutaneous syndrome. Journal of Medical Genetics, 2007, 44, 651-656.	3.2	114

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37	A paternal deletion of MKRN3, MAGEL2 and NDN does not result in Praderâ€“Willi syndrome. European Journal of Human Genetics, 2009, 17, 582-590.	2.8	112
38	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, 285-293.	6.2	110
39	The genetic basis of DOORS syndrome: an exome-sequencing study. Lancet Neurology, The, 2014, 13, 44-58.	10.2	108
40	A mutation screen in patients with Kabuki syndrome. Human Genetics, 2011, 130, 715-724.	3.8	106
41	Integrative analysis revealed the molecular mechanism underlying <scp>RBM</scp>10â€mediated splicing regulation. EMBO Molecular Medicine, 2013, 5, 1431-1442.	6.9	106
42	Clinical relevance of systematic phenotyping and exome sequencing in patients with short stature. Genetics in Medicine, 2018, 20, 630-638.	2.4	101
43	UBQLN4 Represses Homologous Recombination and Is Overexpressed in Aggressive Tumors. Cell, 2019, 176, 505-519.e22.	28.9	100
44	Molecular and clinical characterization of cardioâ€facioâ€cutaneous (CFC) syndrome: Overlapping clinical manifestations with Costello syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 799-807.	1.2	96
45	A novel microdeletion syndrome involving 5q14.3-q15: clinical and molecular cytogenetic characterization of three patients. European Journal of Human Genetics, 2009, 17, 1592-1599.	2.8	96
46	Molecular and clinical analysis ofRAF1in Noonan syndrome and related disorders: dephosphorylation of serine 259 as the essential mechanism for mutant activation. Human Mutation, 2010, 31, 284-294.	2.5	96
47	Mutation and phenotypic spectrum in patients with cardioâ€facioâ€cutaneous and Costello syndrome. Clinical Genetics, 2008, 73, 62-70.	2.0	94
48	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.	3.8	93
49	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, 3121-3126.	3.6	92
50	Deficiency of <scp>ECHS</scp>1 causes mitochondrial encephalopathy with cardiac involvement. Annals of Clinical and Translational Neurology, 2015, 2, 492-509.	3.7	90
51	Genetic determination of human facial morphology: links between cleft-lips and normal variation. European Journal of Human Genetics, 2011, 19, 1192-1197.	2.8	89
52	A Noncoding, Regulatory Mutation Implicates HCFC1 in Nonsyndromic Intellectual Disability. American Journal of Human Genetics, 2012, 91, 694-702.	6.2	89
53	A review of craniofacial disorders caused by spliceosomal defects. Clinical Genetics, 2015, 88, 405-415.	2.0	85
54	Further delineation of Kabuki syndrome in 48 wellâ€defined new individuals. American Journal of Medical Genetics, Part A, 2005, 132A, 265-272.	1.2	84

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55	A family with autosomal dominant oculo-auriculo-vertebral spectrum. Clinical Dysmorphology, 2007, 16, 1-7.	0.3	81
56	Loss of the BMP Antagonist, SMOC-1, Causes Ophthalmo-Acromelic (Waardenburg Anophthalmia) Syndrome in Humans and Mice. PLoS Genetics, 2011, 7, e1002114.	3.5	81
57	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.	6.2	81
58	BCL11B mutations in patients affected by a neurodevelopmental disorder with reduced type 2 innate lymphoid cells. Brain, 2018, 141, 2299-2311.	7.6	81
59	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	2.4	80
60	Syndrome identification based on 2D analysis software. European Journal of Human Genetics, 2006, 14, 1082-1089.	2.8	77
61	Clinical and mutation data in 12 patients with the clinical diagnosis of Nager syndrome. Human Genetics, 2013, 132, 885-898.	3.8	77
62	The ARID1B phenotype: What we have learned so far. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 276-289.	1.6	77
63	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, 513-524.	3.2	75
64	Extreme Growth Failure is a Common Presentation of Ligase IV Deficiency. Human Mutation, 2014, 35, 76-85.	2.5	74
65	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, 753-760.	2.8	73
66	De Novo Mutations in CHAMP1 Cause Intellectual Disability with Severe Speech Impairment. American Journal of Human Genetics, 2015, 97, 493-500.	6.2	71
67	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, 3969-3983.	2.9	70
68	Human facial dysostoses. Clinical Genetics, 2013, 83, 499-510.	2.0	69
69	Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. Brain, 2022, 145, 2991-3009.	7.6	69
70	RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome. Journal of Clinical Investigation, 2015, 125, 3585-3599.	8.2	69
71	Computer-based recognition of dysmorphic faces. European Journal of Human Genetics, 2003, 11, 555-560.	2.8	68
72	Histone acetylation dependent allelic expression imbalance of BAPX1 in patients with the oculo-auriculo-vertebral spectrum. Human Molecular Genetics, 2006, 15, 581-587.	2.9	68

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73	Acrofacial Dysostosis, Cincinnati Type, a Mandibulofacial Dysostosis Syndrome with Limb Anomalies, Is Caused by POLR1A Dysfunction. American Journal of Human Genetics, 2015, 96, 765-774.	6.2	67
74	Phenotype and genotype in Nicolaidesâ€“Baraitser syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 302-314.	1.6	66
75	Diagnostic algorithms in Charcotâ€“Marieâ€“Tooth neuropathies: experiences from a German genetic laboratory on the basis of 1206 index patients. Clinical Genetics, 2016, 89, 34-43.	2.0	66
76	Heterozygous HNRNPU variants cause early onset epilepsy and severe intellectual disability. Human Genetics, 2017, 136, 821-834.	3.8	66
77	Microcephaly with or without chorioretinopathy, lymphoedema, or mental retardation (MCLMR): review of phenotype associated with KIF11 mutations. European Journal of Human Genetics, 2014, 22, 881-887.	2.8	65
78	Altered Development of NKT Cells, Î³Î´ T Cells, CD8 T Cells and NK Cells in a PLZF Deficient Patient. PLoS ONE, 2011, 6, e24441.	2.5	65
79	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, 140A, 2039-2049.	1.2	64
80	Autosomal recessive POLR1D mutation with decrease of TCOF1 mRNA is responsible for Treacher Collins syndrome. Genetics in Medicine, 2014, 16, 720-724.	2.4	63
81	Mutations in the BAF-Complex Subunit DPF2 Are Associated with Coffin-Siris Syndrome. American Journal of Human Genetics, 2018, 102, 468-479.	6.2	63
82	POLR1B and neural crest cell anomalies in Treacher Collins syndrome type 4. Genetics in Medicine, 2020, 22, 547-556.	2.4	63
83	Mutations in chromatin regulators functionally link Cornelia de Lange syndrome and clinically overlapping phenotypes. Human Genetics, 2017, 136, 307-320.	3.8	61
84	Two patients with EP300 mutations and facial dysmorphism different from the classic Rubinsteinâ€“Taybi syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 181-184.	1.2	60
85	Rare Copy Number Variants Are a Common Cause of Short Stature. PLoS Genetics, 2013, 9, e1003365.	3.5	60
86	The face of Noonan syndrome: Does phenotype predict genotype. American Journal of Medical Genetics, Part A, 2010, 152A, 1960-1966.	1.2	59
87	Exome sequencing unravels unexpected differential diagnoses in individuals with the tentative diagnosis of Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes. Human Genetics, 2015, 134, 553-568.	3.8	59
88	Bi-allelic Variants in METTL5 Cause Autosomal-Recessive Intellectual Disability and Microcephaly. American Journal of Human Genetics, 2019, 105, 869-878.	6.2	58
89	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, 731-737.	3.2	56
90	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.	2.7	56

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91	Further delineation of the KAT6B molecular and phenotypic spectrum. European Journal of Human Genetics, 2015, 23, 1165-1170.	2.8	56
92	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.	6.2	55
93	Genetic Analysis of â€˜PAX6-Negativeâ€™ Individuals with Aniridia or Gillespie Syndrome. PLoS ONE, 2016, 11, e0153757.	2.5	54
94	Heterozygosity for ARID2 loss-of-function mutations in individuals with a Coffinâ€“Siris syndrome-like phenotype. Human Genetics, 2017, 136, 297-305.	3.8	53
95	Genetic screening confirms heterozygous mutations in ACAN as a major cause of idiopathic short stature. Scientific Reports, 2017, 7, 12225.	3.3	53
96	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, 563-569.	2.8	52
97	Mutations in MAP3K7 that Alter the Activity of the TAK1 Signaling Complex Cause Frontometaphyseal Dysplasia. American Journal of Human Genetics, 2016, 99, 392-406.	6.2	52
98	A new face of Borjesonâ€“Forssmanâ€“Lehmann syndrome? De novo mutations inPHF6in seven females with a distinct phenotype. Journal of Medical Genetics, 2013, 50, 838-847.	3.2	50
99	Behavioral phenotype in five individuals with de novo mutations within the GRIN2B gene. Behavioral and Brain Functions, 2013, 9, 20.	3.3	47
100	Reproduction abnormalities and twin pregnancies in parents of sporadic patients with oculo-auriculo-vertebral spectrum/Goldenhar syndrome. Human Genetics, 2007, 121, 369-376.	3.8	46
101	Autosomal recessive mental retardation: homozygosity mapping identifies 27 single linkage intervals, at least 14 novel loci and several mutation hotspots. Human Genetics, 2011, 129, 141-148.	3.8	45
102	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.	6.2	45
103	Cardioâ€facioâ€cutaneous (CFC) syndrome â€” a distinct entity? Report of three patients demonstrating the diagnostic difficulties in delineation of CFC syndrome. Clinical Genetics, 1997, 52, 37-46.	2.0	43
104	Bohringâ€“Opitz (Oberklaidâ€“Danks) syndrome: clinical study, review of the literature, and discussion of possible pathogenesis. European Journal of Human Genetics, 2011, 19, 513-519.	2.8	43
105	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.	3.8	43
106	Wilms' Tumor 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.	2.0	43
107	Identification of 34 novel and 56 knownFOXL2mutations in patients with blepharophimosis syndrome. Human Mutation, 2008, 29, E205-E219.	2.5	42
108	Congenital diaphragmatic hernia interval on chromosome 8p23.1 characterized by genetics and protein interaction networks. American Journal of Medical Genetics, Part A, 2012, 158A, 3148-3158.	1.2	42

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109	WDR73Mutations Cause Infantile Neurodegeneration and Variable Glomerular Kidney Disease. Human Mutation, 2015, 36, 1021-1028.	2.5	42
110	De novo MECP2 duplication in two females with random X-inactivation and moderate mental retardation. European Journal of Human Genetics, 2011, 19, 507-512.	2.8	41
111	Novel Mutations Including Deletions of the EntireOFD1Gene in 30 Families with Type 1 Orofaciodigital Syndrome: A Study of the Extensive Clinical Variability. Human Mutation, 2013, 34, 237-247.	2.5	41
112	DOORS syndrome: Phenotype, genotype and comparison with Coffinâ€Siris syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 327-332.	1.6	40
113	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, 837-843.	1.2	39
114	Delineation of PIGV mutation spectrum and associated phenotypes in hyperphosphatasia with mental retardation syndrome. European Journal of Human Genetics, 2014, 22, 762-767.	2.8	39
115	Expanding the phenotype of IQSEC2 mutations: truncating mutations in severe intellectual disability. European Journal of Human Genetics, 2014, 22, 289-292.	2.8	39
116	De Novo Mutations Affecting the Catalytic CÎ± Subunit of PP2A, PPP2CA, Cause Syndromic Intellectual Disability Resembling Other PP2A-Related Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 104, 139-156.	6.2	39
117	Automated syndrome detection in a set of clinical facial photographs. American Journal of Medical Genetics, Part A, 2011, 155, 2161-2169.	1.2	38
118	Expanding the clinical spectrum of the â€˜<scp>HDAC8</scp>â€phenotypeâ€™ â€“ implications for molecular diagnostics, counseling and risk prediction. Clinical Genetics, 2016, 89, 564-573.	2.0	38
119	Genetic variants in components of the NALCNâ€“UNC80â€“UNC79 ion channel complex cause a broad clinical phenotype (NALCN channelopathies). Human Genetics, 2018, 137, 753-768.	3.8	38
120	Homozygous and Compound-Heterozygous Mutations in TGDS Cause Catel-Manzke Syndrome. American Journal of Human Genetics, 2014, 95, 763-770.	6.2	37
121	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.	6.2	37
122	Loss-of-function variants in HIVEP2 are a cause of intellectual disability. European Journal of Human Genetics, 2016, 24, 556-561.	2.8	36
123	Penetrance and Expressivity in Inherited Cancer Predisposing Syndromes. Trends in Cancer, 2018, 4, 718-728.	7.4	36
124	Goltzâ€“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, 1207-1215.	2.8	35
125	Infectious and Immunologic Phenotype of MECP2 Duplication Syndrome. Journal of Clinical Immunology, 2015, 35, 168-181.	3.8	35
126	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.	2.8	35

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127	Bainbridgeâ€“Ropers syndrome caused by loss-of-function variants in ASXL3: a recognizable condition. European Journal of Human Genetics, 2017, 25, 183-191.	2.8	35
128	Family-based germline sequencing in children with cancer. Oncogene, 2019, 38, 1367-1380.	5.9	33
129	Microcephalic osteodysplastic primordial dwarfism type I with biallelic mutations in the RNU4ATAC gene. Clinical Genetics, 2012, 82, 140-146.	2.0	31
130	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. Human Mutation, 2018, 39, 1246-1261.	2.5	31
131	Variants in CPLX1 in two families with autosomal-recessive severe infantile myoclonic epilepsy and ID. European Journal of Human Genetics, 2017, 25, 889-893.	2.8	30
132	Genetic predisposition in children with cancer â€“ affected families' acceptance of Trio-WES. European Journal of Pediatrics, 2018, 177, 53-60.	2.7	30
133	Esophageal atresia, hypoplasia of zygomatic complex, microcephaly, cup-shaped ears, congenital heart defect, and mental retardationâ€”New MCA/MR syndrome in two affected sibs and a mildly affected mother?. American Journal of Medical Genetics, Part A, 2007, 143A, 1135-1142.	1.2	29
134	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.	1.3	29
135	Genotypeâ€“phenotype correlation in eight new patients with a deletion encompassing 2q31.1. American Journal of Medical Genetics, Part A, 2010, 152A, 1213-1224.	1.2	29
136	Cohen syndrome diagnosis using whole genome arrays. Journal of Medical Genetics, 2011, 48, 136-140.	3.2	29
137	De novo FBXO11 mutations are associated with intellectual disability and behavioural anomalies. Human Genetics, 2018, 137, 401-411.	3.8	29
138	De novo variants in PAK1 lead to intellectual disability with macrocephaly and seizures. Brain, 2019, 142, 3351-3359.	7.6	29
139	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.	2.8	28
140	Severe neurocognitive and growth disorders due to variation in THOC2 , an essential component of nuclear mRNA export machinery. Human Mutation, 2018, 39, 1126-1138.	2.5	28
141	Females with de novo aberrations in PHF6: Clinical overlap of Borjesonâ€“Forssmanâ€“Lehmann with Coffinâ€“Siris syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 290-301.	1.6	27
142	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.	2.8	27
143	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.	1.9	26
144	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.	2.4	26

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145	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, 685-688.	1.3	25
146	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.	0.7	25
147	Wide clinical variability in conditions with coarse facial features and hypertrichosis caused by mutations in ABCC9. American Journal of Medical Genetics, Part A, 2013, 161, 295-300.	1.2	24
148	â€˜Splitting versus lumpingâ€™: Templeâ€“Baraitser and Zimmermannâ€“Laband Syndromes. Human Genetics, 2015, 134, 1089-1097.	3.8	24
149	Autosomal dominant frontometaphyseal dysplasia: Delineation of the clinical phenotype. American Journal of Medical Genetics, Part A, 2017, 173, 1739-1746.	1.2	24
150	Autosomal dominant intellectual disability. Medizinische Genetik, 2018, 30, 318-322.	0.2	24
151	Five patients with novel overlapping interstitial deletions in 8q22.2q22.3. American Journal of Medical Genetics, Part A, 2011, 155, 1857-1864.	1.2	23
152	Treacher Collins syndrome: clinical implications for the paediatricianâ€”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, 1611-1618.	2.7	23
153	X-linked intellectual disability type Nascimento is a clinically distinct, probably underdiagnosed entity. Orphanet Journal of Rare Diseases, 2013, 8, 146.	2.7	22
154	Mutations in SMARCB1 and in other Coffinâ€“Siris syndrome genes lead to various brain midline defects. Nature Communications, 2019, 10, 2966.	12.8	22
155	ANKRD11 variants: <scp>KBG</scp> syndrome and beyond. Clinical Genetics, 2021, 100, 187-200.	2.0	21
156	FOXL2copy number changes in the molecular pathogenesis of BPES: unique cohort of 17 deletions. Human Mutation, 2010, 31, n/a-n/a.	2.5	20
157	Autosomal dominant spinal muscular atrophy with lower extremity predominance: A recognizable phenotype of BICD2 mutations. Muscle and Nerve, 2016, 54, 496-500.	2.2	20
158	The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant. Annals of Neurology, 2018, 83, 926-934.	5.3	20
159	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.	1.2	19
160	Nine newly identified individuals refine the phenotype associated with MYT1L mutations. American Journal of Medical Genetics, Part A, 2020, 182, 1021-1031.	1.2	19
161	First Report of a Single Exon Deletion in TCOF1 Causing Treacher Collins Syndrome. Molecular Syndromology, 2011, 2, 53-59.	0.8	18
162	Hematopoietic Stem Cell Transplantation in an Infant with Immunodeficiency, Centromeric Instability, and Facial Anomaly Syndrome. Frontiers in Immunology, 2017, 8, 773.	4.8	18

#	Article	IF	Citations
163	Choline transporter-like 1 deficiency causes a new type of childhood-onset neurodegeneration. Brain, 2020, 143, 94-111.	7.6	18
164	160Âkb deletion in ISPD unmasking a recessive mutation in a patient with Walkerâ€“Warburg syndrome. European Journal of Medical Genetics, 2013, 56, 689-694.	1.3	17
165	SON haploinsufficiency causes impaired pre-mRNA splicing of CAKUT genes and heterogeneous renalÂphenotypes. Kidney International, 2019, 95, 1494-1504.	5.2	17
166	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.	1.5	17
167	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.	6.2	17
168	Two adults with Rubinsteinâ€“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, 2849-2854.	1.2	16
169	Severe nemaline myopathy associated with consecutive mutations E74D and H75Y on a single ACTA1 allele. Neuromuscular Disorders, 2009, 19, 481-484.	0.6	16
170	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.	6.2	16
171	Mutations in PRDM15 Are a Novel Cause of Galloway-Mowat Syndrome. Journal of the American Society of Nephrology: JASN, 2021, 32, 580-596.	6.1	15
172	Two brothers with Burn-McKeown syndrome. Clinical Dysmorphology, 2003, 12, 171-174.	0.3	14
173	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, 1223-1230.	1.2	14
174	Fragile X mental retardation protein protects against tumour necrosis factor-mediated cell death and liver injury. Gut, 2020, 69, 133-145.	12.1	14
175	Profound inhibition of CD73-dependent formation of anti-inflammatory adenosine in B cells of SLE patients. EBioMedicine, 2021, 73, 103616.	6.1	14
176	Hallermann-Streiff Syndrome: No Evidence for a Link to Laminopathies. Molecular Syndromology, 2011, 2, 27-34.	0.8	13
177	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, 1349-1355.	2.8	13
178	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, 728-733.	1.2	13
179	Pontocerebellar hypoplasia due to bi-allelic variants in MINPP1. European Journal of Human Genetics, 2021, 29, 411-421.	2.8	13
180	C2orf69 mutations disrupt mitochondrial function and cause a multisystem human disorder with recurring autoinflammation. Journal of Clinical Investigation, 2021, 131, .	8.2	13

#	Article	IF	Citations
181	A Novel Homozygous <i>WDR72</i> Mutation in Two Siblings with Amelogenesis Imperfecta and Mild Short Stature. Molecular Syndromology, 2012, 3, 223-229.	0.8	12
182	Identification and Functional Characterization of Two IntronicNIPBLMutations in Two Patients with Cornelia de Lange Syndrome. BioMed Research International, 2016, 2016, 1-8.	1.9	12
183	X-linked recessive VACTERL-H due to a mutation in FANCB in a preterm boy. Clinical Dysmorphology, 2016, 25, 73-76.	0.3	12
184	Tentative clinical diagnosis of Lujanâ€Fryns syndromeâ€"A conglomeration of different genetic entities?. American Journal of Medical Genetics, Part A, 2016, 170, 94-102.	1.2	11
185	Evolutionary conserved networks of human height identify multiple Mendelian causes of short stature. European Journal of Human Genetics, 2019, 27, 1061-1071.	2.8	11
186	Defining the phenotypical spectrum associated with variants in TUBB2A. Journal of Medical Genetics, 2021, 58, 33-40.	3.2	11
187	Homozygous truncating PTPRF mutation causes athelia. Human Genetics, 2014, 133, 1041-1047.	3.8	10
188	Academic application of Good Cell Culture Practice for induced pluripotent stem cells. ALTEX: Alternatives To Animal Experimentation, 2021, 38, 595-614.	1.5	10
189	Identification of causative variants in TXNL4A in Burn-McKeown syndrome and isolated choanal atresia. European Journal of Human Genetics, 2017, 25, 1126-1133.	2.8	10
190	Classification and Visualization Based on Derived Image Features: Application to Genetic Syndromes. PLoS ONE, 2014, 9, e109033.	2.5	9
191	Isolated PREPL deficiency associated with congenital myasthenic syndrome-22. Klinische Padiatrie, 2018, 230, 281-283.	0.6	9
192	Fatal metabolic decompensation in carbonic anhydrase VA deficiency despite early treatment and control of hyperammonemia. Genetics in Medicine, 2020, 22, 654-655.	2.4	9
193	The Macrophage Migration Inhibitory Factor (MIF) Promoter Polymorphisms (rs3063368, rs755622) Predict Acute Kidney Injury and Death after Cardiac Surgery. Journal of Clinical Medicine, 2020, 9, 2936.	2.4	9
194	Intellectual disability associated with craniofacial dysmorphism, cleft palate, and congenital heart defect due to a de novo <scp>MEIS2</scp> mutation: A clinical longitudinal study. American Journal of Medical Genetics, Part A, 2021, 185, 1216-1221.	1.2	9
195	Parental origin and functional relevance of a de novo UBE3A variant. European Journal of Medical Genetics, 2011, 54, 19-24.	1.3	8
196	Distinctive facial features in idiopathic Moyamoya disease in Caucasians: a first systematic analysis. PeerJ, 2018, 6, e4740.	2.0	8
197	Genome-wide methylation analysis of retrocopy-associated CpG islands and their genomic environment. Epigenetics, 2016, 11, 216-226.	2.7	7
198	Central nervous system anomalies in two females with Borjeson-Forssman-Lehmann syndrome. Epilepsy and Behavior, 2017, 69, 104-109.	1.7	7

#	Article	IF	Citations
199	How I approach hereditary cancer predisposition in a child with cancer. Pediatric Blood and Cancer, 2019, 66, e27916.	1.5	7
200	Novel EXOSC3 pathogenic variant results in a mild course of neurologic disease with cerebellum involvement. European Journal of Medical Genetics, 2020, 63, 103649.	1.3	7
201	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.	6.2	6
202	Acute myeloid leukemia-induced functional inhibition of healthy CD34+ hematopoietic stem and progenitor cells. Stem Cells, 2021, 39, 1270-1284.	3.2	6
203	Reconstruction of images from Gabor graphs with applications in facial image processing. International Journal of Wavelets, Multiresolution and Information Processing, 2015, 13, 1550019.	1.3	5
204	ASPP2 deficiency causes features of 1q41q42 microdeletion syndrome. Cell Death and Differentiation, 2016, 23, 1973-1984.	11.2	5
205	Genome-Wide Analysis of the Nucleosome Landscape in Individuals with Coffin-Siris Syndrome. Cytogenetic and Genome Research, 2019, 159, 1-11.	1.1	5
206	<scp>QRICH1</scp> variants in <scp>Ververiâ€Brady</scp> syndromeâ€”delineation of the genotypic and phenotypic spectrum. Clinical Genetics, 2021, 99, 199-207.	2.0	5
207	Genetics of craniofacial malformations. Seminars in Fetal and Neonatal Medicine, 2021, 26, 101290.	2.3	5
208	Moyamoya angiopathy in PHACE syndrome not associated with RNF213 variants. Child's Nervous System, 2019, 35, 1231-1237.	1.1	4
209	Defining the genotypic and phenotypic spectrum of X-linked MSL3-related disorder. Genetics in Medicine, 2021, 23, 384-395.	2.4	4
210	Homozygous myotonic dystrophy: Clinical findings in two patients and review of the literature. American Journal of Medical Genetics, Part A, 2007, 143A, 2058-2061.	1.2	3
211	A patient with a de-novo deletion 3p25.3 and features overlapping with Rubinsteinâ€“Taybi syndrome. Clinical Dysmorphology, 2014, 23, 67-70.	0.3	3
212	Further evidence for complex inheritance of holoprosencephaly: Lessons learned from preâ€•and postnatal diagnostic testing in Germany. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 198-205.	1.6	3
213	Maternal transmission of a mild Coffinâ€“Siris syndrome phenotype caused by a SOX11 missense variant. European Journal of Human Genetics, 2022, 30, 126-132.	2.8	3
214	The recurrent missense mutation p.(Arg367Trp) in YARS1 causes a distinct neurodevelopmental phenotype. Journal of Molecular Medicine, 2021, 99, 1755-1768.	3.9	3
215	Biallelic variants in YRDC cause a developmental disorder with progeroid features. Human Genetics, 2021, 140, 1679-1693.	3.8	3
216	Case Report: Severe Neonatal Course in Paternally Derived Familial Hypocalciuric Hypercalcemia. Frontiers in Endocrinology, 2021, 12, 700612.	3.5	3

#	Article	IF	Citations
217	Endocannabinoid dysfunction in neurological disease: neuro-ocular DAGLA-related syndrome. Brain, 2022, 145, 3383-3390.	7.6	3
218	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, 156, 9-13.	1.1	2
219	Progenitor cells derived from geneâ€engineered human induced pluripotent stem cells as synthetic cancer cell alternatives for in vitro pharmacology. Biotechnology Journal, 2022, , 2100693.	3.5	2
220	Oculo-oto-facial dysplasia (OOFD) versus Burnâ€“McKeown syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 2381-2382.	1.2	1
221	Autism spectrum disorder and Li-Fraumeni syndrome: purely coincidental or mechanistically associated?. Molecular and Cellular Pediatrics, 2017, 4, 8.	1.8	1
222	Mikrozephaliesyndrome und geistige Behinderung. Medizinische Genetik, 2009, 21, 224-230.	0.2	0
223	Liquid Biopsies. Medizinische Genetik, 2016, 28, 233-233.	0.2	0
224	P 308. Autosomal Recessive Mutations in the NALCN Gene: A Rare Cause of a Severe Developmental Disorder with Facial Dysmorphia, Epilepsy and Cheyneâ€“Stokes/Biotâ€™s Respiration with Central Apneas. , 2018, , .		0
225	RANBP2 mutation mimicking viral encephalitis. Neuropediatrics, 2022, 0, .	0.6	0
226	NFB-03. Neurological manifestations in children and adolescents with Neurofibromatosis type 1 - Implications for management and surveillance. Neuro-Oncology, 2022, 24, i128-i128.	1.2	0