Anita Rauch

List of Publications by Year
in descending order

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255	17,020	17776 65	22488 117
papers	citations	h-index	g-index

270	270	270	25224
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Human COQ4 deficiency: delineating the clinical, metabolic and neuroimaging phenotypes. Journal of Medical Genetics, 2022, 59, 878-887.	1.5	9
2	Response to Cueto-GonzÃ¡lez etÂal. Genetics in Medicine, 2022, 24, 757.	1.1	0
3	The MAP3K7 gene: Further delineation of clinical characteristics and genotype/phenotype correlations. Human Mutation, 2022, 43, 1377-1395.	1.1	5
4	Characterization of SETD1A haploinsufficiency in humans and Drosophila defines a novel neurodevelopmental syndrome. Molecular Psychiatry, 2021, 26, 2013-2024.	4.1	43
5	New insights into the clinical and molecular spectrum of the novel CYFIP2-related neurodevelopmental disorder and impairment of the WRC-mediated actin dynamics. Genetics in Medicine, 2021, 23, 543-554.	1.1	32
6	The broad phenotypic spectrum of PPP2R1A-related neurodevelopmental disorders correlates with the degree of biochemical dysfunction. Genetics in Medicine, 2021, 23, 352-362.	1.1	23
7	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	1.1	16
8	<scp>MED27</scp> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. Annals of Neurology, 2021, 89, 828-833.	2.8	14
9	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	2.6	48
10	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. Genetics in Medicine, 2021, 23, 1028-1040.	1.1	34
11	Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. Human Genetics, 2021, 140, 1109-1120.	1.8	18
12	Loss-of-function and missense variants in NSD2 cause decreased methylation activity and are associated with a distinct developmental phenotype. Genetics in Medicine, 2021, 23, 1474-1483.	1.1	24
13	CSNK2B: A broad spectrum of neurodevelopmental disability and epilepsy severity. Epilepsia, 2021, 62, e103-e109.	2.6	13
14	Rare variants in KDR, encoding VEGF Receptor 2, are associated with tetralogy of Fallot. Genetics in Medicine, 2021, 23, 1952-1960.	1.1	7
15	Confirmation of Ogden syndrome as an Xâ€linked recessive fatal disorder due to a recurrent <scp>NAA10</scp> variant and review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 2546-2560.	0.7	12
16	Expanding the phenotype: Four new cases and hope for treatment in <scp>Bachmannâ€Bupp</scp> syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 3485-3493.	0.7	10
17	Generation and characterization of an endogenously tagged SPG11-human iPSC line by CRISPR/Cas9 mediated knock-in. Stem Cell Research, 2021, 56, 102520.	0.3	1
18	Novel Biallelic Variants in KIF21A Cause a Novel Phenotype of Fetal Akinesia with Neurodevelopmental Defects. , 2021, 52, .		0

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19	Fetal tuberous sclerosis and diagnosis of paternal gonadal mosaicism. Ultrasound in Obstetrics and Gynecology, 2020, 55, 691-692.	0.9	2
20	A clinical scoring system for congenital contractural arachnodactyly. Genetics in Medicine, 2020, 22, 124-131.	1.1	17
21	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	0.7	42
22	High-resolution chromosomal microarrayÂanalysis for copy-number variations in high-functioning autism reveals large aberration typical for intellectual disability. Journal of Neural Transmission, 2020, 127, 81-94.	1.4	5
23	Rare copy number variants in individuals at clinical high risk for psychosis: Enrichment of synaptic/brainâ€related functional pathways. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2020, 183, 140-151.	1.1	0
24	Severe reaction to radiotherapy provoked by hypomorphic germline mutations in ATM (ataxiaâ€“telangiectasia mutated gene). Molecular Genetics & Genomic Medicine, 2020, 8, e1409.	0.6	8
25	Novel morphological and genetic features of fumarate hydratase deficient renal cell carcinoma in <scp>HLRCC</scp> syndrome patients with a tailored therapeutic approach. Genes Chromosomes and Cancer, 2020, 59, 611-619.	1.5	19
26	Genome-wide non-invasive prenatal testing in single- and multiple-pregnancies at any risk: Identification of maternal polymorphisms to reduce the number of unnecessary invasive confirmation testing. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2020, 252, 19-29.	0.5	7
27	CDK5RAP2 primary microcephaly is associated with hypothalamic, retinal and cochlear developmental defects. Journal of Medical Genetics, 2020, 57, 389-399.	1.5	17
28	Insight Into the Ontogeny of GnRH Neurons From Patients Born Without a Nose. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 1538-1551.	1.8	7
29	Bi-allelic Pathogenic Variants in HS2ST1 Cause a Syndrome Characterized by Developmental Delay and Corpus Callosum, Skeletal, and Renal Abnormalities. American Journal of Human Genetics, 2020, 107, 1044-1061.	2.6	11
30	Swiss newborn screening for severe T and B cell deficiency with a combined TREC/KREC assay â€“ management recommendations. Swiss Medical Weekly, 2020, 150, w20254.	0.8	17
31	Prenatal diagnosis of HNF1Bâ€associated renal cysts: Is there a need to differentiate intragenic variants from 17q12 microdeletion syndrome?. Prenatal Diagnosis, 2019, 39, 1136-1147.	1.1	16
32	De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Callosum, Axon, Cardiac, Ocular, and Genital Defects. American Journal of Human Genetics, 2019, 105, 854-868.	2.6	29
33	Spatially clustering de novo variants in CYFIP2, encoding the cytoplasmic FMRP interacting protein 2, cause intellectual disability and seizures. European Journal of Human Genetics, 2019, 27, 747-759.	1.4	47
34	CUGC for Simpson-Golabi-Behmel syndrome (SGBS). European Journal of Human Genetics, 2019, 27, 663-668.	1.4	9
35	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	2.6	56
36	Elucidation of the phenotypic spectrum and genetic landscape in primary and secondary microcephaly. Genetics in Medicine, 2019, 21, 2043-2058.	1.1	57

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37	Further corroboration of distinct functional features in SCN2A variants causing intellectual disability or epileptic phenotypes. Molecular Medicine, 2019, 25, 6.	1.9	42
38	Deleterious Variation in BRSK2 Associates with a Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 701-708.	2.6	19
39	Evolutionary conserved networks of human height identify multiple Mendelian causes of short stature. European Journal of Human Genetics, 2019, 27, 1061-1071.	1.4	11
40	De novo truncating variants in the intronless IRF2BPL are responsible for developmental epileptic encephalopathy. Genetics in Medicine, 2019, 21, 1008-1014.	1.1	34
41	The role of recessive inheritance in early-onset epileptic encephalopathies: a combined whole-exome sequencing and copy number study. European Journal of Human Genetics, 2019, 27, 408-421.	1.4	52
42	Prevalence of genetic susceptibility for breast and ovarian cancer in a non-cancer related study population: secondary germline findings from a Swiss single centre cohort. Swiss Medical Weekly, 2019, 149, w20092.	0.8	1
43	Surprisingly good outcome in antenatal diagnosis of severe hydrocephalus related to CCDC88C deficiency. European Journal of Medical Genetics, 2018, 61, 189-196.	0.7	9
44	Phenotype and genotype of 87 patients with Mowatâ€“Wilson syndrome and recommendations for care. Genetics in Medicine, 2018, 20, 965-975.	1.1	67
45	Clinical and experimental evidence suggest a link between KIF7 and C5orf42-related ciliopathies through Sonic Hedgehog signaling. European Journal of Human Genetics, 2018, 26, 197-209.	1.4	23
46	Clinical relevance of systematic phenotyping and exome sequencing in patients with short stature. Genetics in Medicine, 2018, 20, 630-638.	1.1	101
47	Novel STRA6 null mutations in the original family described with Matthewâ€“Wood syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 134-138.	0.7	10
48	Autosomal recessive primary microcephaly due to ASPM mutations: An update. Human Mutation, 2018, 39, 319-332.	1.1	53
49	Need for high-resolution Genetic Analysis in iPSC: Results and Lessons from the ForIPS Consortium. Scientific Reports, 2018, 8, 17201.	1.6	70
50	Bi-allelic TMEM94 Truncating Variants Are Associated with Neurodevelopmental Delay, Congenital Heart Defects, and Distinct Facial Dysmorphism. American Journal of Human Genetics, 2018, 103, 948-967.	2.6	18
51	De Novo Variants in the F-Box Protein FBXO11 in 20 Individuals with a Variable Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 103, 305-316.	2.6	48
52	Clinical and functional characterization of two novel ZBTB20 mutations causing Primrose syndrome. Human Mutation, 2018, 39, 959-964.	1.1	11
53	Biallelic UFM1 and UFC1 mutations expand the essential role of ufmylation in brain development. Brain, 2018, 141, 1934-1945.	3.7	70
54	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. Nature Genetics, 2017, 49, 238-248.	9.4	131

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55	Microarrays in prenatal diagnosis. Best Practice and Research in Clinical Obstetrics and Gynaecology, 2017, 42, 53-63.	1.4	43
56	FOXP2variants in 14 individuals with developmental speech and language disorders broaden the mutational and clinical spectrum. Journal of Medical Genetics, 2017, 54, 64-72.	1.5	67
57	STAG1mutations cause a novel cohesinopathy characterised by unspecific syndromic intellectual disability. Journal of Medical Genetics, 2017, 54, 479-488.	1.5	35
58	Confirmation of mutations inPROSCas a novel cause of vitamin B_₆-dependent epilepsy. Journal of Medical Genetics, 2017, 54, 809-814.	1.5	66
59	YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction. American Journal of Human Genetics, 2017, 100, 907-925.	2.6	125
60	The HHID syndrome of hypertrichosis, hyperkeratosis, abnormal corpus callosum, intellectual disability, and minor anomalies is caused by mutations in ARID1B. American Journal of Medical Genetics, Part A, 2017, 173, 1440-1443.	0.7	11
61	Low-Level Chromosomal Mosaicism in Neurodevelopmental Disorders. Molecular Syndromology, 2017, 8, 266-271.	0.3	5
62	Monoallelic BMP2 Variants Predicted to Result in Haploinsufficiency Cause Craniofacial, Skeletal, and Cardiac Features Overlapping Those of 20p12 Deletions. American Journal of Human Genetics, 2017, 101, 985-994.	2.6	44
63	Genotype-phenotype evaluation of MED13L defects in the light of a novel truncating and a recurrent missense mutation. European Journal of Medical Genetics, 2017, 60, 451-464.	0.7	34
64	High resolution chromosomal microarray analysis in paediatric obsessive-compulsive disorder. BMC Medical Genomics, 2017, 10, 68.	0.7	21
65	MCAD-Deficiency with Severe Neonatal Onset, Fatal Outcome and Normal Acylcarnitine Profile. International Journal of Neonatal Screening, 2017, 3, 21.	1.2	0
66	Plasma metabolomics reveals a diagnostic metabolic fingerprint for mitochondrial aconitase (ACO2) deficiency. PLoS ONE, 2017, 12, e0176363.	1.1	40
67	Genetic and neurodevelopmental spectrum ofSYNGAP1-associated intellectual disability and epilepsy. Journal of Medical Genetics, 2016, 53, 511-522.	1.5	135
68	Mutations in CRADD Result in Reduced Caspase-2-Mediated Neuronal Apoptosis and Cause Megalencephaly with a Rare Lissencephaly Variant. American Journal of Human Genetics, 2016, 99, 1117-1129.	2.6	50
69	A recurrent germline mutation in the PIGA gene causes Simpsonâ€Golabiâ€Behmel syndrome type 2. American Journal of Medical Genetics, Part A, 2016, 170, 392-402.	0.7	34
70	The value of plasma vitamin B₆ profiles in early onset epileptic encephalopathies. Journal of Inherited Metabolic Disease, 2016, 39, 733-741.	1.7	19
71	Gene discovery for Mendelian conditions via social networking: de novo variants in KDM1A cause developmental delay and distinctive facial features. Genetics in Medicine, 2016, 18, 788-795.	1.1	88
72	Noninvasive prenatal testing: more caution in counseling is needed in high risk pregnancies with ultrasound abnormalities. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2016, 200, 72-75.	0.5	15

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73	Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. JAMA Psychiatry, 2016, 73, 20.	6.0	195
74	N⁸â€acetylspermidine as a potential plasma biomarker for Snyderâ€Robinson syndrome identified by clinical metabolomics. Journal of Inherited Metabolic Disease, 2016, 39, 131-137.	1.7	33
75	Mutations in <scp>CDK</scp> 5 <scp>RAP</scp> 2 cause Seckel syndrome. Molecular Genetics & Genomic Medicine, 2015, 3, 467-480.	0.6	55
76	A severe congenital myasthenic syndrome with â€œdropped headâ€•caused by novel MUSK mutations. Muscle and Nerve, 2015, 52, 668-673.	1.0	21
77	IntragenicKANSL1mutations and chromosome 17q21.31 deletions: broadening the clinical spectrum and genotypeâ€“phenotype correlations in a large cohort of patients. Journal of Medical Genetics, 2015, 52, 804-814.	1.5	47
78	Variants inCUL4Bare Associated with Cerebral Malformations. Human Mutation, 2015, 36, 106-117.	1.1	37
79	De novo missense mutations in the NAA10 gene cause severe non-syndromic developmental delay in males and females. European Journal of Human Genetics, 2015, 23, 602-609.	1.4	72
80	PDE3A mutations cause autosomal dominant hypertension with brachydactyly. Nature Genetics, 2015, 47, 647-653.	9.4	146
81	Molecular Diversity and Associated Phenotypic Spectrum of GermlineCBLMutations. Human Mutation, 2015, 36, 787-796.	1.1	36
82	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. American Journal of Human Genetics, 2015, 96, 784-796.	2.6	53
83	Mutations in XRCC4 cause primary microcephaly, short stature and increased genomic instability. Human Molecular Genetics, 2015, 24, 3708-17.	1.4	26
84	Hydrops, fetal pleural effusions and chylothorax in three patients with CBL mutations. American Journal of Medical Genetics, Part A, 2015, 167, 394-399.	0.7	24
85	Infantile Epileptic Encephalopathy, Transient Choreoathetotic Movements, and Hypersomnia due to a De Novo Missense Mutation in the SCN2A Gene. Neuropediatrics, 2014, 45, 261-264.	0.3	30
86	The clinical significance of small copy number variants in neurodevelopmental disorders. Journal of Medical Genetics, 2014, 51, 677-688.	1.5	72
87	Dysmorphology at a distance: results of a web-based diagnostic service. European Journal of Human Genetics, 2014, 22, 327-332.	1.4	14
88	Exome sequencing in unspecific intellectual disability and rare disorders. Molecular Cytogenetics, 2014, 7, I26.	0.4	1
89	A newly recognized 13q12.3 microdeletion syndrome characterized by intellectual disability, microcephaly, and eczema/atopic dermatitis encompassing the HMGB1 and KATNAL1 genes. American Journal of Medical Genetics, Part A, 2014, 164, 1277-1283.	0.7	25
90	Microcephalic osteodysplastic primordial dwarfism type II (MOPD II) with multiple vascular complications misdiagnosed as Dubowitz syndrome. European Journal of Pediatrics, 2014, 173, 1253-1256.	1.3	17

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91	Mutations Affecting the SAND Domain of DEAF1 Cause Intellectual Disability with Severe Speech Impairment and Behavioral Problems. American Journal of Human Genetics, 2014, 94, 649-661.	2.6	59
92	LETM1 haploinsufficiency causes mitochondrial defects in Wolf-Hirschhorn syndrome patient cells: implications for dissecting underlying pathomechanisms in this condition. DMM Disease Models and Mechanisms, 2014, 7, 535-45.	1.2	26
93	Highâ€resolution chromosomal microarrays in prenatal diagnosis significantly increase diagnostic power. Prenatal Diagnosis, 2014, 34, 525-533.	1.1	42
94	Nephrocalcinosis (Enamel Renal Syndrome) Caused by Autosomal Recessive FAM20A Mutations. Nephron Physiology, 2013, 122, 1-6.	1.5	84
95	Further delineation of genotypeâ€"phenotype correlation in homozygous 2p21 deletion syndromes: First description of patients without cystinuria. American Journal of Medical Genetics, Part A, 2013, 161, 1853-1859.	0.7	22
96	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.	1.4	13
97	Somatic mosaicism in a mother of two children with Pittâ€"Hopkins syndrome. Clinical Genetics, 2013, 83, 73-77.	1.0	31
98	Dosage changes of MED13L further delineate its role in congenital heart defects and intellectual disability. European Journal of Human Genetics, 2013, 21, 1100-1104.	1.4	57
99	De Novo Mutations in the Genome Organizer CTCF Cause Intellectual Disability. American Journal of Human Genetics, 2013, 93, 124-131.	2.6	151
100	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.	1.4	190
101	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.	1.5	50
102	Rare Copy Number Variants Are a Common Cause of Short Stature. PLoS Genetics, 2013, 9, e1003365.	1.5	60
103	Association Between C677T Polymorphism of Methylene Tetrahydrofolate Reductase and Congenital Heart Disease. Circulation: Cardiovascular Genetics, 2013, 6, 347-353.	5.1	31
104	Biallelic SEMA3A defects cause a novel type of syndromic short stature. American Journal of Medical Genetics, Part A, 2013, 161, 2880-2889.	0.7	9
105	Genome-wide association study identifies loci on 12q24 and 13q32 associated with Tetralogy of Fallot. Human Molecular Genetics, 2013, 22, 1473-1481.	1.4	82
106	Phenotype-specific effect of chromosome 1q21.1 rearrangements and GJA5 duplications in 2436 congenital heart disease patients and 6760 controls. Human Molecular Genetics, 2012, 21, 1513-1520.	1.4	101
107	Range of genetic mutations associated with severe non-syndromic sporadic intellectual disability: an exome sequencing study. Lancet, The, 2012, 380, 1674-1682.	6.3	940
108	Contribution of Global Rare Copy-Number Variants to the Risk of Sporadic Congenital Heart Disease. American Journal of Human Genetics, 2012, 91, 489-501.	2.6	272

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109	Macrocerebellum: Significance and Pathogenic Considerations. Cerebellum, 2012, 11, 1026-1036.	1.4	25
110	NovelKIF7mutations extend the phenotypic spectrum of acrocallosal syndrome. Journal of Medical Genetics, 2012, 49, 713-720.	1.5	28
111	WachstumsstÃ¶rungen als Leitsymptom. Medizinische Genetik, 2012, 24, 123-137.	0.1	0
112	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.	2.6	225
113	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. Nature, 2011, 478, 97-102.	13.7	394
114	Microdeletions of chromosome 7p21, including TWIST1, associated with significant microcephaly, facial dysmorphism, and short stature. European Journal of Medical Genetics, 2011, 54, 256-261.	0.7	9
115	7ÂMb de novo deletion within 8q21 in a patient with distal arthrogryposis type 2B (DA2B). European Journal of Medical Genetics, 2011, 54, e495-e500.	0.7	10
116	Familial short stature due to a 5q22.1â€“q23.2 duplication refines the 5q duplication spectrum. European Journal of Medical Genetics, 2011, 54, e521-e524.	0.7	5
117	The molecular basis of the cartilage-hair hypoplasiaâ€“anauxetic dysplasia spectrum. Best Practice and Research in Clinical Endocrinology and Metabolism, 2011, 25, 131-142.	2.2	46
118	The shortest of the short: Pericentrin mutations and beyond. Best Practice and Research in Clinical Endocrinology and Metabolism, 2011, 25, 125-130.	2.2	40
119	De novo nonsense mutations in ASXL1 cause Bohring-Opitz syndrome. Nature Genetics, 2011, 43, 729-731.	9.4	236
120	Genome-wide copy number profiling using a 100K SNP array reveals novel disease-related genes BORIS and TSHZ1 in juvenile angiofibroma. International Journal of Oncology, 2011, 39, 1143-51.	1.4	12
121	CEP152 is a genome maintenance protein disrupted in Seckel syndrome. Nature Genetics, 2011, 43, 23-26.	9.4	201
122	Clinical utility gene card for: Mowatâ€“Wilson syndrome. European Journal of Human Genetics, 2011, 19, 4-4.	1.4	5
123	The MEF2C-Related and 5q14.3q15 Microdeletion Syndrome. Molecular Syndromology, 2011, 2, 164-170.	0.3	51
124	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.	1.5	220
125	NEK1 Mutations Cause Short-Rib Polydactyly Syndrome Type Majewski. American Journal of Human Genetics, 2011, 88, 106-114.	2.6	151
126	Expanding the clinical spectrum associated with defects in CNTNAP2 and NRXN1. BMC Medical Genetics, 2011, 12, 106.	2.1	109

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127	The smallest teeth in the world are caused by mutations in the PCNT gene. American Journal of Medical Genetics, Part A, 2011, 155, 1398-1403.	0.7	37
128	Identification of a novel loss-of-function calcium channel gene mutation in short QT syndrome (SQTS6). European Heart Journal, 2011, 32, 1077-1088.	1.0	178
129	In-Frame Deletion and Missense Mutations of the C-Terminal Helicase Domain of <i>SMARCA2</i> in Three Patients with Nicolaides-Baraitser Syndrome. Molecular Syndromology, 2011, 2, 237-244.	0.3	58
130	Disruption of the histone acetyltransferase MYST4 leads to a Noonan syndromeâ€“like phenotype and hyperactivated MAPK signaling in humans and mice. Journal of Clinical Investigation, 2011, 121, 3479-3491.	3.9	89
131	Disturbed Wnt Signalling due to a Mutation in CCDC88C Causes an Autosomal Recessive Non-Syndromic Hydrocephalus with Medial Diverticulum. Molecular Syndromology, 2010, 1, 99-112.	0.3	82
132	Mesomelia-Synostoses Syndrome Results from Deletion of SULF1 and SLCO5A1 Genes at 8q13. American Journal of Human Genetics, 2010, 87, 95-100.	2.6	42
133	Severe clinical course of Hirschsprung disease in a Mowat-Wilson syndrome patient. Journal of Applied Genetics, 2010, 51, 111-113.	1.0	14
134	Mutations in MEF2C from the 5q14.3q15 microdeletion syndrome region are a frequent cause of severe mental retardation and diminish MECP2 and CDKL5 expression. Human Mutation, 2010, 31, 722-733.	1.1	163
135	The face of Noonan syndrome: Does phenotype predict genotype. American Journal of Medical Genetics, Part A, 2010, 152A, 1960-1966.	0.7	59
136	Altered TGFÎ² signaling and cardiovascular manifestations in patients with autosomal recessive cutis laxa type I caused by fibulin-4 deficiency. European Journal of Human Genetics, 2010, 18, 895-901.	1.4	132
137	Clinical utility gene card for: DiGeorge syndrome, velocardiofacial syndrome, Shprintzen syndrome, chromosome 22q11.2 deletion syndrome (22q11.2, TBX1). European Journal of Human Genetics, 2010, 18, 1071-1071.	1.4	6
138	Mutations in GRIN2A and GRIN2B encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes. Nature Genetics, 2010, 42, 1021-1026.	9.4	431
139	Disruption of ST5 is associated with mental retardation and multiple congenital anomalies. Journal of Medical Genetics, 2010, 47, 91-98.	1.5	12
140	Unmasking of a Recessive SCARF2 Mutation by a 22q11.12 de novo Deletion in a Patient with Van den Ende-Gupta Syndrome. Molecular Syndromology, 2010, 1, 239-245.	0.3	32
141	Systematic survey of variants in TBX1 in non-syndromic tetralogy of Fallot identifies a novel 57 base pair deletion that reduces transcriptional activity but finds no evidence for association with common variants. Heart, 2010, 96, 1651-1655.	1.2	61
142	Comprehensive genotype-phenotype analysis in 230 patients with tetralogy of Fallot. Journal of Medical Genetics, 2010, 47, 321-331.	1.5	126
143	Microcephalin and pericentrin regulate mitotic entry via centrosome-associated Chk1. Journal of Cell Biology, 2009, 185, 1149-1157.	2.3	83
144	Independent NF1 and PTPN11 mutations in a family with neurofibromatosisâ€Noonan syndrome. American Journal of Medical Genetics, Part A, 2009, 149A, 1263-1267.	0.7	55

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145	Pulmonary hypoplasiaâ€“diaphragmatic herniaâ€“anophthalmiaâ€“cardiac defect (PDAC) syndrome due to STRA6 mutationsâ€”What are the minimal criteria?. American Journal of Medical Genetics, Part A, 2009, 149A, 2457-2463.	0.7	35
146	Chromosomale Ursachen der geistigen Behinderung. Medizinische Genetik, 2009, 21, 237-245.	0.1	0
147	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.	1.4	35
148	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.	1.4	96
149	Lethal cutis laxa with contractural arachnodactyly, overgrowth and soft tissue bleeding due to a novel homozygous fibulinâ€4 gene mutation. Clinical Genetics, 2009, 76, 276-281.	1.0	50
150	CNTNAP2 and NRXN1 Are Mutated in Autosomal-Recessive Pitt-Hopkins-like Mental Retardation and Determine the Level of a Common Synaptic Protein in Drosophila. American Journal of Human Genetics, 2009, 85, 655-666.	2.6	573
151	Dosage-Dependent Severity of the Phenotype in Patients with Mental Retardation Due to a Recurrent Copy-Number Gain at Xq28 Mediated by an Unusual Recombination. American Journal of Human Genetics, 2009, 85, 809-822.	2.6	70
152	Further clinical and molecular delineation of the 9q subtelomeric deletion syndrome supports a major contribution of EHMT1 haploinsufficiency to the core phenotype. Journal of Medical Genetics, 2009, 46, 598-606.	1.5	194
153	Mild clinical phenotype and subtle radiographic findings in an infant with cartilage-hair hypoplasia. Turkish Journal of Pediatrics, 2009, 51, 493-6.	0.3	6
154	Spontaneous Development and Rupture of Pulmonary Artery Aneurysm: A Rare Complication in an Infant with Peripheral Pulmonary Artery Stenoses Due to Mutation of the Elastin Gene. Pediatric Cardiology, 2008, 29, 438-441.	0.6	5
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156	Clinical and genetic distinction of Schimke immunoâ€osseous dysplasia and cartilageâ€hair hypoplasia. American Journal of Medical Genetics, Part A, 2008, 146A, 2013-2017.	0.7	18
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