Lina Basel-Vanagaite

List of Publications by Year
in descending order

137	7,474	66343	62596
papers	citations	42 h-index	80 g-index

141	141	141	12102
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A nonsense variant in the second exon of the canonical transcript of <scp>NSD1</scp> does not cause Sotos syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 369-372.	1.2	1
2	Residual risk for clinically significant copy number variants in low-risk pregnancies, following exclusion of noninvasive prenatal screeningâ€"detectable findings. American Journal of Obstetrics and Gynecology, 2022, 226, 562.e1-562.e8.	1.3	8
3	<scp>DYRK1B</scp> haploinsufficiency in a family with metabolic syndrome and abnormal cognition. Clinical Genetics, 2022, 101, 265-266.	2.0	4
4	Challenges in variant interpretation in prenatal exome sequencing. European Journal of Medical Genetics, 2022, 65, 104410.	1.3	3
5	Prenatal and postnatal chromosomal microarray analysis in 885 cases of various congenital heart defects. Archives of Gynecology and Obstetrics, 2022, 306, 1007-1013.	1.7	4
6	Diâ€genic inheritance of germline POLE and PMS2 pathogenic variants causes a unique condition associated with pediatric cancer predisposition. Clinical Genetics, 2022, 101, 442-447.	2.0	5
7	In-silico phenotype prediction by normal mode variant analysis in TUBB4A-related disease. Scientific Reports, 2022, 12, 58.	3.3	2
8	Congenital Thrombocytopenia Associated with a Heterozygous Variant in the MEIS1 Gene Encoding a Transcription Factor Essential for Megakaryopoiesis. Platelets, 2022, , 1-4.	2.3	0
9	Pathogenic variantâ€based preconception carrier screening in the <scp>Israeli Jewish</scp> population. Clinical Genetics, 2022, 101, 517-529.	2.0	4
10	Further delineation of auriculocondylar syndrome based on 14 novel cases and reassessment of 25 published cases. Human Mutation, 2022, 43, 582-594.	2.5	6
11	Physician anxiety or maternal choice?. American Journal of Obstetrics and Gynecology, 2022, 226, 600-601.	1.3	1
12	Postpartum women's attitudes to disclosure of adultâ€onset conditions in pregnancy. Prenatal Diagnosis, 2022, 42, 1038-1048.	2.3	3
13	When phenotype does not match genotype: importance of â€œreal-timeâ€•refining of phenotypic information for exome data interpretation. Genetics in Medicine, 2021, 23, 215-221.	2.4	10
14	The role of phenotype-based search approaches using public online databases in diagnostics of Mendelian disorders. Genetics in Medicine, 2021, 23, 1095-1100.	2.4	5
15	Epilepsy and electroencephalogram evolution in YWHAG gene mutation: A new phenotype and review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 901-908.	1.2	6
16	The phenotype of 15 cases with rare 8q24.13â€q24.3 deletionsâ€"A new syndrome or still an enigma?. American Journal of Medical Genetics, Part A, 2021, 185, 1461-1467.	1.2	0
17	Two intronic cisâ€acting variants in both alleles of the POLR3A gene cause progressive spastic ataxia with hypodontia. Clinical Genetics, 2021, 99, 713-718.	2.0	2
18	A recurrent, homozygous EMC10 frameshift variant is associated with a syndrome of developmental delay with variable seizures and dysmorphic features. Genetics in Medicine, 2021, 23, 1158-1162.	2.4	13

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19	Biallelic variants in ETV2 in a family with congenital heart defects, vertebral abnormalities and preaxial polydactyly. European Journal of Medical Genetics, 2021, 64, 104124.	1.3	6
20	Mild Phenotype of Wolfram Syndrome Associated With a Common Pathogenic Variant Is Predicted by a Structural Model of Wolframin. Neurology: Genetics, 2021, 7, e578.	1.9	12
21	Is it time to report carrier state for recessive disorders in every microarray analysis?â€”A pilot model based on hearing loss genes deletions. European Journal of Human Genetics, 2021, 29, 1292-1300.	2.8	1
22	A study of normal copy number variations in Israeli population. Human Genetics, 2021, 140, 553-563.	3.8	1
23	Biallelic truncating variants in the muscular Aâ€type laminâ€interacting protein (MLIP) gene cause myopathy with hyperCKemia. European Journal of Neurology, 2021, , .	3.3	4
24	Phenotype and mutation expansion of the PTPN23 associated disorder characterized by neurodevelopmental delay and structural brain abnormalities. European Journal of Human Genetics, 2020, 28, 76-87.	2.8	21
25	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain, 2020, 143, 55-68.	7.6	38
26	Ten points to consider when providing genetic counseling for variants of incomplete penetrance and variable expressivity detected in a prenatal setting. Acta Obstetricia Et Gynecologica Scandinavica, 2020, 99, 1427-1429.	2.8	3
27	Impact of a national population-based carrier-screening program on spinal muscular atrophy births. Neuromuscular Disorders, 2020, 30, 970-974.	0.6	17
28	Spectrum of genes for inherited hearing loss in the Israeli Jewish population, including the novel human deafness gene <scp>ATOH1</scp>. Clinical Genetics, 2020, 98, 353-364.	2.0	15
29	Based on a cohort of 52,879 microarrays, recurrent intragenic FBN2 deletion encompassing exons 1â€“8 does not cause Beals syndrome. European Journal of Medical Genetics, 2020, 63, 104008.	1.3	3
30	Should We Report 15q11.2 BP1-BP2 Deletions and Duplications in the Prenatal Setting?. Journal of Clinical Medicine, 2020, 9, 2602.	2.4	8
31	Paediatric systemic lupus erythematosus as a manifestation of constitutional mismatch repair deficiency. Journal of Medical Genetics, 2020, 57, 505-508.	3.2	7
32	Bi-allelic ADARB1 Variants Associated with Microcephaly, Intellectual Disability, and Seizures. American Journal of Human Genetics, 2020, 106, 467-483.	6.2	31
33	Lysine acetyltransferase 8 is involved in cerebral development and syndromic intellectual disability. Journal of Clinical Investigation, 2020, 130, 1431-1445.	8.2	40
34	The yield of full BRCA1/2 genotyping in Israeli Arab high-risk breast/ovarian cancer patients. Breast Cancer Research and Treatment, 2019, 178, 231-237.	2.5	7
35	The rare 13q33â€“q34 microdeletions: eight new patients and review of the literature. Human Genetics, 2019, 138, 1145-1153.	3.8	11
36	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogryposis. American Journal of Human Genetics, 2019, 105, 689-705.	6.2	48

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37	De novo variants in PAK1 lead to intellectual disability with macrocephaly and seizures. Brain, 2019, 142, 3351-3359.	7.6	29
38	Chromosomal microarray vs. NIPS: analysis of 5541 low-risk pregnancies. Genetics in Medicine, 2019, 21, 2462-2467.	2.4	55
39	Consensus interpretation of the p.Met34Thr and p.Val37Ile variants in GJB2 by the ClinGen Hearing Loss Expert Panel. Genetics in Medicine, 2019, 21, 2442-2452.	2.4	56
40	Bayesian-based noninvasive prenatal diagnosis of single-gene disorders. Genome Research, 2019, 29, 428-438.	5.5	31
41	Variable Features of Juvenile Polyposis Syndrome With Gastric Involvement Among Patients With a Large Genomic Deletion of BMPR1A. Clinical and Translational Gastroenterology, 2019, 10, e00054.	2.5	9
42	Identifying facial phenotypes of genetic disorders using deep learning. Nature Medicine, 2019, 25, 60-64.	30.7	449
43	Improved diagnostics by exome sequencing following raw data reevaluation by clinical geneticists involved in the medical care of the individuals tested. Genetics in Medicine, 2019, 21, 1443-1451.	2.4	64
44	Whole-exome sequencing reveals POC5 as a novel gene associated with autosomal recessive retinitis pigmentosa. Human Molecular Genetics, 2018, 27, 614-624.	2.9	26
45	A de novo GABRA2 missense mutation in severe early-onset epileptic encephalopathy with a choreiform movement disorder. European Journal of Paediatric Neurology, 2018, 22, 516-524.	1.6	23
46	When genotype is not predictive of phenotype: implications for genetic counseling based on 21,594 chromosomal microarray analysis examinations. Genetics in Medicine, 2018, 20, 128-131.	2.4	47
47	Cytogenetic analysis in fetuses with late onset abnormal sonographic findings. Journal of Perinatal Medicine, 2018, 46, 975-982.	1.4	13
48	Substrate interaction defects in histidyl-tRNA synthetase linked to dominant axonal peripheral neuropathy. Human Mutation, 2018, 39, 415-432.	2.5	30
49	Kaufman oculocerebrofacial syndrome: Novel UBE3B mutations and clinical features in four unrelated patients. American Journal of Medical Genetics, Part A, 2018, 176, 187-193.	1.2	13
50	Refining the phenotype associated with GNB1 mutations: Clinical data on 18 newly identified patients and review of the literature. American Journal of Medical Genetics, Part A, 2018, 176, 2259-2275.	1.2	47
51	The yield of full BRCA1/2 genotyping in Israeli high-risk breast/ovarian cancer patients who do not carry the predominant mutations. Breast Cancer Research and Treatment, 2018, 172, 151-157.	2.5	19
52	X-linked elliptocytosis with impaired growth is related to mutated AMMECR1. Gene, 2017, 606, 47-52.	2.2	6
53	Large Intragenic Deletion in DSTYK Underlies Autosomal-Recessive Complicated Spastic Paraparesis, SPG23. American Journal of Human Genetics, 2017, 100, 364-370.	6.2	32
54	Homozygous mutations in <scp>VAMP</scp>1 cause a presynaptic congenital myasthenic syndrome. Annals of Neurology, 2017, 81, 597-603.	5.3	48

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55	A Biallelic Mutation in the Homologous Recombination Repair Gene SPIDR Is Associated With Human Gonadal Dysgenesis. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 681-688.	3.6	47
56	Intra-familial Variation in Clinical Phenotype of CARD14-related Psoriasis. Acta Dermato-Venereologica, 2016, 96, 885-887.	1.3	13
57	Polymicrogyria and myoclonic epilepsy in autosomal recessive cutis laxa type 2A. Neurogenetics, 2016, 17, 251-257.	1.4	4
58	De Novo Mutations in CHD4 , an ATP-Dependent Chromatin Remodeler Gene, Cause an Intellectual Disability Syndrome with Distinctive Dysmorphisms. American Journal of Human Genetics, 2016, 99, 934-941.	6.2	111
59	Recognition of the Cornelia de Lange syndrome phenotype with facial dysmorphology novel analysis. Clinical Genetics, 2016, 89, 557-563.	2.0	77
60	A founder mutation in ADAMTSL4 causes early-onset bilateral ectopia lentis among Jews of Bukharian origin. Molecular Genetics and Metabolism, 2016, 117, 38-41.	1.1	13
61	IncreasedSTAG2dosage defines a novel cohesinopathy with intellectual disability and behavioral problems. Human Molecular Genetics, 2015, 24, 7171-7181.	2.9	28
62	Lethal neonatal rigidity and multifocal seizure syndrome â€“ Report of another family with a BRAT1 mutation. European Journal of Paediatric Neurology, 2015, 19, 240-242.	1.6	28
63	Keppen-Lubinsky Syndrome Is Caused by Mutations in the Inwardly Rectifying K+ Channel Encoded by KCNJ6. American Journal of Human Genetics, 2015, 96, 295-300.	6.2	95
64	Homozygous MED25 mutation implicated in eyeâ€“intellectual disability syndrome. Human Genetics, 2015, 134, 577-587.	3.8	18
65	A de-novo interstitial microduplication involving 2p16.1-p15 and mirroring 2p16.1-p15 microdeletion syndrome: Clinical and molecular analysis. European Journal of Paediatric Neurology, 2015, 19, 711-715.	1.6	16
66	Broad Phenotypic Heterogeneity due to a Novel SCN1A Mutation in a Family With Genetic Epilepsy With Febrile Seizures Plus. Journal of Child Neurology, 2014, 29, 221-226.	1.4	48
67	Exome Sequencing Reveals SYCE1 Mutation Associated With Autosomal Recessive Primary Ovarian Insufficiency. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E2129-E2132.	3.6	128
68	Identification of a novel mutation in the PNLIP gene in two brothers with congenital pancreatic lipase deficiency. Journal of Lipid Research, 2014, 55, 307-312.	4.2	29
69	A novel splice-site mutation in theAAGABgene segregates with hereditary punctate palmoplantar keratoderma and congenital dysplasia of the hip in a large family. Clinical and Experimental Dermatology, 2014, 39, 182-186.	1.3	14
70	Expanding the clinical and mutational spectrum of Kaufman oculocerebrofacial syndrome with biallelic UBE3B mutations. Human Genetics, 2014, 133, 939-949.	3.8	29
71	Two siblings with early infantile myoclonic encephalopathy due to mutation in the gene encoding mitochondrial glutamate/H+ symporter SLC25A22. European Journal of Paediatric Neurology, 2014, 18, 801-805.	1.6	26
72	Homozygous truncating PTPRF mutation causes athelia. Human Genetics, 2014, 133, 1041-1047.	3.8	10

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73	Expanding the Mutational Spectrum ofCRLF1in Crisponi/CISS1 Syndrome. Human Mutation, 2014, 35, 424-433.	2.5	21
74	Dominant Mutations in GRHL3 Cause Van der Woude Syndrome and Disrupt Oral Periderm Development. American Journal of Human Genetics, 2014, 94, 23-32.	6.2	195
75	A novel mutation in the endosomal Na+/H+ exchanger NHE6 (SLC9A6) causes Christianson syndrome with electrical status epilepticus during slow-wave sleep (ESES). Epilepsy Research, 2014, 108, 811-815.	1.6	40
76	Biallelic SZT2 Mutations Cause Infantile Encephalopathy with Epilepsy and Dysmorphic Corpus Callosum. American Journal of Human Genetics, 2013, 93, 524-529.	6.2	81
77	Microcephaly Thin Corpus Callosum Intellectual Disability Syndrome Caused by Mutated TAF2. Pediatric Neurology, 2013, 49, 411-416.e1.	2.1	36
78	Mutation Spectrum inRAB3GAP1,RAB3GAP2, andRAB18and Genotype-Phenotype Correlations in Warburg Micro Syndrome and Martsolf Syndrome. Human Mutation, 2013, 34, 686-696.	2.5	114
79	Mutations in PIK3R1 Cause SHORT Syndrome. American Journal of Human Genetics, 2013, 93, 158-166.	6.2	156
80	A novel mutation in the GAN gene causes an intermediate form of giant axonal neuropathy in an Arabâ€“Israeli family. European Journal of Paediatric Neurology, 2013, 17, 259-264.	1.6	9
81	Mutations in GMPPA Cause a Glycosylation Disorder Characterized by Intellectual Disability and Autonomic Dysfunction. American Journal of Human Genetics, 2013, 93, 727-734.	6.2	57
82	Heterogeneity of mutational mechanisms and modes of inheritance in auriculocondylar syndrome. Journal of Medical Genetics, 2013, 50, 174-186.	3.2	44
83	High frequency of autosomalâ€recessive DFNB59 hearing loss in an isolated Arab population in Israel. Clinical Genetics, 2012, 82, 271-276.	2.0	18
84	eIF2Î³ Mutation that Disrupts eIF2 Complex Integrity Links Intellectual Disability to Impaired Translation Initiation. Molecular Cell, 2012, 48, 641-646.	9.7	63
85	Microcephaly-Thin Corpus Callosum Syndrome Maps to 8q23.2-q24.12. Pediatric Neurology, 2012, 46, 363-368.	2.1	14
86	Genotype-phenotype correlation in 22q11.2 deletion syndrome. BMC Medical Genetics, 2012, 13, 122.	2.1	83
87	Deficiency for the Ubiquitin Ligase UBE3B in a Blepharophimosis-Ptosis-Intellectual-Disability Syndrome. American Journal of Human Genetics, 2012, 91, 998-1010.	6.2	82
88	Transient Infantile Hypertriglyceridemia, Fatty Liver, and Hepatic Fibrosis Caused by Mutated GPD1, Encoding Glycerol-3-Phosphate Dehydrogenase 1. American Journal of Human Genetics, 2012, 90, 49-60.	6.2	74
89	New Syndrome of Congenital Circumferential Skin Folds Associated with Multiple Congenital Anomalies. Pediatric Dermatology, 2012, 29, 89-95.	0.9	7
90	Phenotypic psychiatric characterization of children with Williams syndrome and response of those with ADHD to methylphenidate treatment. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2012, 159B, 13-20.	1.7	32

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91	A Comparative Study of Hearing Loss in Two Microdeletion Syndromes: Velocardiofacial (22q11.2) Tj ETQq1 1 0.784314 rgBT /Overlo	1.8	26
92	Three sibs with microcephaly, clubfeet and agenesis of corpus callosum: A new genetic syndrome?. American Journal of Medical Genetics, Part A, 2011, 155, 1060-1065.	1.2	2
93	Xâ€linked mental retardation with alacrima and achalasiaâ€"Triple A syndrome or a new syndrome?. American Journal of Medical Genetics, Part A, 2011, 155, 1959-1963.	1.2	4
94	An Emerging 1q21.1 Deletion-Associated Neurodevelopmental Phenotype. Journal of Child Neurology, 2011, 26, 113-116.	1.4	19
95	Case series: 2q33.1 microdeletion syndrome--further delineation of the phenotype. Journal of Medical Genetics, 2011, 48, 290-298.	3.2	59
96	Clinical, neuroradiological and genetic findings in pontocerebellar hypoplasia. Brain, 2011, 134, 143-156.	7.6	200
97	Multiple congenital anomalies-hypotonia-seizures syndrome is caused by a mutation in PIGN. Journal of Medical Genetics, 2011, 48, 383-389.	3.2	138
98	Terminal Osseous Dysplasia Is Caused by a Single Recurrent Mutation in the FLNA Gene. American Journal of Human Genetics, 2010, 87, 146-153.	6.2	50
99	SOBP Is Mutated in Syndromic and Nonsyndromic Intellectual Disability and Is Highly Expressed in the Brain Limbic System. American Journal of Human Genetics, 2010, 87, 694-700.	6.2	20
100	Acute lymphoblastic leukemia in Weaver syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 383-386.	1.2	26
101	Familial hydrocephalus with normal cognition and distinctive radiological features. American Journal of Medical Genetics, Part A, 2010, 152A, 2743-2748.	1.2	8
102	<i>CDH3</i>-Related Syndromes: Report on a New Mutation and Overview of the Genotype-Phenotype Correlations. Molecular Syndromology, 2010, 1, 223-230.	0.8	19
103	Clinical and Brain Imaging Heterogeneity of Severe Microcephaly. Pediatric Neurology, 2010, 43, 7-16.	2.1	29
104	Loss-of-function mutations in ATP6V0A2 impair vesicular trafficking, tropoelastin secretion and cell survival. Human Molecular Genetics, 2009, 18, 2149-2165.	2.9	115
105	Keppenâ€"Lubinsky syndrome: Expanding the phenotype. American Journal of Medical Genetics, Part A, 2009, 149A, 1827-1829.	1.2	10
106	RIN2 Deficiency Results in Macrocephaly, Alopecia, Cutis Laxa, and Scoliosis: MACS Syndrome. American Journal of Human Genetics, 2009, 85, 254-263.	6.2	89
107	A Truncating Mutation of TRAPPC9 Is Associated with Autosomal-Recessive Intellectual Disability and Postnatal Microcephaly. American Journal of Human Genetics, 2009, 85, 897-902.	6.2	134
108	Psychiatric morbidity with focus on obsessiveâ€"compulsive disorder in an Israeli cohort of adolescents with mild to moderate mental retardation. Journal of Neural Transmission, 2008, 115, 929-936.	2.8	28

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109	Ethnically diverse causes of Walker-Warburg syndrome (WWS):FCMDmutations are a more common cause of WWS outside of the Middle East. Human Mutation, 2008, 29, E231-E241.	2.5	67
110	Yunisâ€“Varon syndrome: Further delineation of the phenotype. American Journal of Medical Genetics, Part A, 2008, 146A, 532-537.	1.2	18
111	Autosomal dominant isolated question mark ear. American Journal of Medical Genetics, Part A, 2008, 146A, 2280-2283.	1.2	12
112	Impaired glycosylation and cutis laxa caused by mutations in the vesicular H+-ATPase subunit ATP6V0A2. Nature Genetics, 2008, 40, 32-34.	21.4	330
113	tRNA splicing endonuclease mutations cause pontocerebellar hypoplasia. Nature Genetics, 2008, 40, 1113-1118.	21.4	217
114	Autosomal recessive ichthyosis with hypotrichosis syndrome: further delineation of the phenotype. Clinical Genetics, 2008, 74, 47-53.	2.0	47
115	Genetic Carrier Screening for Spinal Muscular Atrophy and Spinal Muscular Atrophy with Respiratory Distress 1 in an Isolated Population in Israel. Genetic Testing and Molecular Biomarkers, 2008, 12, 53-56.	1.7	27
116	Expanding the clinical phenotype of autosomal dominant dyskeratosis congenita caused by TERT mutations. Haematologica, 2008, 93, 943-944.	3.5	43
117	Clinical approaches to genetic mental retardation. Israel Medical Association Journal, 2008, 10, 821-6.	0.1	5
118	Autosomal Recessive Ichthyosis with Hypotrichosis Caused by a Mutation in ST14, Encoding Type II Transmembrane Serine Protease Matriptase. American Journal of Human Genetics, 2007, 80, 467-477.	6.2	164
119	Autosomal recessive mental retardation syndrome with anterior maxillary protrusion and strabismus: MRAMS syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 1687-1691.	1.2	7
120	Allele dosage-dependent penetrance of RET proto-oncogene in an Israeli-Arab inbred family segregating Hirschsprung disease. European Journal of Human Genetics, 2007, 15, 242-245.	2.8	13
121	Genetic screening for autosomal recessive nonsyndromic mental retardation in an isolated population in Israel. European Journal of Human Genetics, 2007, 15, 250-253.	2.8	36
122	Genetics of autosomal recessive nonâ€syndromic mental retardation: recent advances. Clinical Genetics, 2007, 72, 167-174.	2.0	23
123	Amniotic trisomy 11 mosaicismâ€”is it a benign finding?. Prenatal Diagnosis, 2006, 26, 778-781.	2.3	9
124	Expanding the phenotypic spectrum of L1CAM-associated disease. Clinical Genetics, 2006, 69, 414-419.	2.0	31
125	Positional cloning uncovers mutations in PLCE1 responsible for a nephrotic syndrome variant that may be reversible. Nature Genetics, 2006, 38, 1397-1405.	21.4	510
126	Mutatednup62causes autosomal recessive infantile bilateral striatal necrosis. Annals of Neurology, 2006, 60, 214-222.	5.3	271

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127	The CC2D1A, a member of a new gene family with C2 domains, is involved in autosomal recessive non-syndromic mental retardation. Journal of Medical Genetics, 2005, 43, 203-210.	3.2	139
128	G Protein-Coupled Receptor-Dependent Development of Human Frontal Cortex. Science, 2004, 303, 2033-2036.	12.6	498
129	Etiological heterogeneity of familial periventricular heterotopia and hydrocephalus. Brain and Development, 2004, 26, 326-334.	1.1	56
130	Twenty-two novel mutations in the lysosomal ?-glucosidase gene (GAA) underscore the genotype-phenotype correlation in glycogen storage disease type II. Human Mutation, 2004, 23, 47-56.	2.5	142
131	De novoSCN1Amutations are a major cause of severe myoclonic epilepsy of infancy. Human Mutation, 2003, 21, 615-621.	2.5	170
132	New syndrome of simplified gyral pattern, micromelia, dysmorphic features and early death. American Journal of Medical Genetics Part A, 2003, 119A, 200-206.	2.4	5
133	Bilateral frontoparietal polymicrogyria: Clinical and radiological features in 10 families with linkage to chromosome 16. Annals of Neurology, 2003, 53, 596-606.	5.3	120
134	An Autosomal Recessive Form of Bilateral Frontoparietal Polymicrogyria Maps to Chromosome 16q12.2-21. American Journal of Human Genetics, 2002, 70, 1028-1033.	6.2	113
135	Seventeen novel mutations that cause profound biotinidase deficiency. Molecular Genetics and Metabolism, 2002, 77, 108-111.	1.1	33
136	Branchial cyst, sensorineural deafness, congenital heart defect, and skeletal abnormalities: Branchio-oto-cardio-skeletal (BOCS) syndrome?. American Journal of Medical Genetics Part A, 2002, 113, 78-81.	2.4	4
137	Fragile-X Carrier Screening and the Prevalence of Premutation and Full-Mutation Carriers in Israel. American Journal of Human Genetics, 2001, 69, 351-360.	6.2	180