

Lina Basel-Vanagaite

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

134
papers

5,537
citations

38
h-index

72
g-index

141
ext. papers

6,621
ext. citations

6.5
avg, IF

4.88
L-index

#	Paper	IF	Citations
134	Prenatal and postnatal chromosomal microarray analysis in 885 cases of various congenital heart defects.. <i>Archives of Gynecology and Obstetrics</i> , 2022 , 1	2.5	1
133	In-silico phenotype prediction by normal mode variant analysis in TUBB4A-related disease.. <i>Scientific Reports</i> , 2022 , 12, 58	4.9	1
132	Congenital Thrombocytopenia Associated with a Heterozygous Variant in the Gene Encoding a Transcription Factor Essential for Megakaryopoiesis.. <i>Platelets</i> , 2022 , 1-4	3.6	
131	Di-genic inheritance of germline POLE and PMS2 pathogenic variants causes a unique condition associated with pediatric cancer predisposition.. <i>Clinical Genetics</i> , 2021 ,	4	1
130	Biallelic Truncating Variants in the Muscular A-Type Lamin-Interacting Protein (MLIP) Gene Cause Myopathy with Hyper-CKemia.. <i>European Journal of Neurology</i> , 2021 ,	6	2
129	Residual risk for clinically significant copy number variants in low-risk pregnancies, following exclusion of noninvasive prenatal screening-detectable findings. <i>American Journal of Obstetrics and Gynecology</i> , 2021 ,	6.4	1
128	DYRK1B haploinsufficiency in a family with metabolic syndrome and abnormal cognition. <i>Clinical Genetics</i> , 2021 , 101, 265	4	
127	Challenges in variant interpretation in prenatal exome sequencing.. <i>European Journal of Medical Genetics</i> , 2021 , 65, 104410	2.6	1
126	A study of normal copy number variations in Israeli population. <i>Human Genetics</i> , 2021 , 140, 553-563	6.3	1
125	Mild Phenotype of Wolfram Syndrome Associated With a Common Pathogenic Variant Is Predicted by a Structural Model of Wolframin. <i>Neurology: Genetics</i> , 2021 , 7, e578	3.8	5
124	Is it time to report carrier state for recessive disorders in every microarray analysis?-A pilot model based on hearing loss genes deletions. <i>European Journal of Human Genetics</i> , 2021 , 29, 1292-1300	5.3	0
123	When phenotype does not match genotype: importance of "real-time" refining of phenotypic information for exome data interpretation. <i>Genetics in Medicine</i> , 2021 , 23, 215-221	8.1	2
122	The role of phenotype-based search approaches using public online databases in diagnostics of Mendelian disorders. <i>Genetics in Medicine</i> , 2021 , 23, 1095-1100	8.1	2
121	Epilepsy and electroencephalogram evolution in YWHAG gene mutation: A new phenotype and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 901-908	2.5	4
120	The phenotype of 15 cases with rare 8q24.13-q24.3 deletions-A new syndrome or still an enigma?. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 1461-1467	2.5	
119	Two intronic cis-acting variants in both alleles of the POLR3A gene cause progressive spastic ataxia with hypodontia. <i>Clinical Genetics</i> , 2021 , 99, 713-718	4	2
118	A recurrent, homozygous EMC10 frameshift variant is associated with a syndrome of developmental delay with variable seizures and dysmorphic features. <i>Genetics in Medicine</i> , 2021 , 23, 1158-1162	8.1	162

117	Biallelic variants in ETV2 in a family with congenital heart defects, vertebral abnormalities and preaxial polydactyly. <i>European Journal of Medical Genetics</i> , 2021 , 64, 104124	2.6	3
116	A nonsense variant in the second exon of the canonical transcript of NSD1 does not cause Sotos syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021 ,	2.5	
115	Physician anxiety or maternal choice?. <i>American Journal of Obstetrics and Gynecology</i> , 2021 ,	6.4	1
114	Paediatric systemic lupus erythematosus as a manifestation of constitutional mismatch repair deficiency. <i>Journal of Medical Genetics</i> , 2020 , 57, 505-508	5.8	5
113	Bi-allelic ADARB1 Variants Associated with Microcephaly, Intellectual Disability, and Seizures. <i>American Journal of Human Genetics</i> , 2020 , 106, 467-483	11	12
112	Lysine acetyltransferase 8 is involved in cerebral development and syndromic intellectual disability. <i>Journal of Clinical Investigation</i> , 2020 , 130, 1431-1445	15.9	17
111	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. <i>Brain</i> , 2020 , 143, 55-68	11.2	18
110	Impact of a national population-based carrier-screening program on spinal muscular atrophy births. <i>Neuromuscular Disorders</i> , 2020 , 30, 970-974	2.9	4
109	Spectrum of genes for inherited hearing loss in the Israeli Jewish population, including the novel human deafness gene ATOH1. <i>Clinical Genetics</i> , 2020 , 98, 353-364	4	8
108	Based on a cohort of 52,879 microarrays, recurrent intragenic FBN2 deletion encompassing exons 1-8 does not cause Beals syndrome. <i>European Journal of Medical Genetics</i> , 2020 , 63, 104008	2.6	3
107	Should We Report 15q11.2 BP1-BP2 Deletions and Duplications in the Prenatal Setting?. <i>Journal of Clinical Medicine</i> , 2020 , 9,	5.1	3
106	Phenotype and mutation expansion of the PTPN23 associated disorder characterized by neurodevelopmental delay and structural brain abnormalities. <i>European Journal of Human Genetics</i> , 2020 , 28, 76-87	5.3	10
105	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogyrosis. <i>American Journal of Human Genetics</i> , 2019 , 105, 689-705	11	22
104	De novo variants in PAK1 lead to intellectual disability with macrocephaly and seizures. <i>Brain</i> , 2019 , 142, 3351-3359	11.2	9
103	Chromosomal microarray vs. NIPS: analysis of 5541 low-risk pregnancies. <i>Genetics in Medicine</i> , 2019 , 21, 2462-2467	8.1	41
102	Consensus interpretation of the p.Met34Thr and p.Val37Ile variants in GJB2 by the ClinGen Hearing Loss Expert Panel. <i>Genetics in Medicine</i> , 2019 , 21, 2442-2452	8.1	22
101	Bayesian-based noninvasive prenatal diagnosis of single-gene disorders. <i>Genome Research</i> , 2019 , 29, 428-438	9.7	16
100	The yield of full BRCA1/2 genotyping in Israeli Arab high-risk breast/ovarian cancer patients. <i>Breast Cancer Research and Treatment</i> , 2019 , 178, 231-237	4.4	6

99	The rare 13q33-q34 microdeletions: eight new patients and review of the literature. <i>Human Genetics</i> , 2019 , 138, 1145-1153	6.3	5
98	Variable Features of Juvenile Polyposis Syndrome With Gastric Involvement Among Patients With a Large Genomic Deletion of BMPR1A. <i>Clinical and Translational Gastroenterology</i> , 2019 , 10, e00054	4.2	4
97	Identifying facial phenotypes of genetic disorders using deep learning. <i>Nature Medicine</i> , 2019 , 25, 60-64	50.5	229
96	Improved diagnostics by exome sequencing following raw data reevaluation by clinical geneticists involved in the medical care of the individuals tested. <i>Genetics in Medicine</i> , 2019 , 21, 1443-1451	8.1	32
95	Whole-exome sequencing reveals POC5 as a novel gene associated with autosomal recessive retinitis pigmentosa. <i>Human Molecular Genetics</i> , 2018 , 27, 614-624	5.6	14
94	A de novo GABRA2 missense mutation in severe early-onset epileptic encephalopathy with a choreiform movement disorder. <i>European Journal of Paediatric Neurology</i> , 2018 , 22, 516-524	3.8	17
93	When genotype is not predictive of phenotype: implications for genetic counseling based on 21,594 chromosomal microarray analysis examinations. <i>Genetics in Medicine</i> , 2018 , 20, 128-131	8.1	32
92	Cytogenetic analysis in fetuses with late onset abnormal sonographic findings. <i>Journal of Perinatal Medicine</i> , 2018 , 46, 975-982	2.7	9
91	The yield of full BRCA1/2 genotyping in Israeli high-risk breast/ovarian cancer patients who do not carry the predominant mutations. <i>Breast Cancer Research and Treatment</i> , 2018 , 172, 151-157	4.4	9
90	Substrate interaction defects in histidyl-tRNA synthetase linked to dominant axonal peripheral neuropathy. <i>Human Mutation</i> , 2018 , 39, 415-432	4.7	19
89	Kaufman oculocerebrofacial syndrome: Novel UBE3B mutations and clinical features in four unrelated patients. <i>American Journal of Medical Genetics, Part A</i> , 2018 , 176, 187-193	2.5	10
88	Refining the phenotype associated with GNB1 mutations: Clinical data on 18 newly identified patients and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2018 , 176, 2259-2275	2.5	27
87	X-linked elliptocytosis with impaired growth is related to mutated AMMECR1. <i>Gene</i> , 2017 , 606, 47-52	3.8	6
86	Large Intragenic Deletion in DSTYK Underlies Autosomal-Recessive Complicated Spastic Paraparesis, SPG23. <i>American Journal of Human Genetics</i> , 2017 , 100, 364-370	11	22
85	Homozygous mutations in VAMP1 cause a presynaptic congenital myasthenic syndrome. <i>Annals of Neurology</i> , 2017 , 81, 597-603	9.4	26
84	A Biallelic Mutation in the Homologous Recombination Repair Gene SPIDR Is Associated With Human Gonadal Dysgenesis. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017 , 102, 681-688	5.6	30
83	De Novo Mutations in CHD4, an ATP-Dependent Chromatin Remodeler Gene, Cause an Intellectual Disability Syndrome with Distinctive Dismorphisms. <i>American Journal of Human Genetics</i> , 2016 , 99, 934-941	11	68
82	Recognition of the Cornelia de Lange syndrome phenotype with facial dysmorphology novel analysis. <i>Clinical Genetics</i> , 2016 , 89, 557-63	4	56

81	A founder mutation in ADAMTSL4 causes early-onset bilateral ectopia lentis among Jews of Bukharian origin. <i>Molecular Genetics and Metabolism</i> , 2016 , 117, 38-41	3.7	6
80	Intra-familial Variation in Clinical Phenotype of CARD14-related Psoriasis. <i>Acta Dermato-Venereologica</i> , 2016 , 96, 885-887	2.2	9
79	Polymicrogyria and myoclonic epilepsy in autosomal recessive cutis laxa type 2A. <i>Neurogenetics</i> , 2016 , 17, 251-257	3	3
78	Homozygous MED25 mutation implicated in eye-intellectual disability syndrome. <i>Human Genetics</i> , 2015 , 134, 577-87	6.3	11
77	A de-novo interstitial microduplication involving 2p16.1-p15 and mirroring 2p16.1-p15 microdeletion syndrome: Clinical and molecular analysis. <i>European Journal of Paediatric Neurology</i> , 2015 , 19, 711-5	3.8	13
76	Increased STAG2 dosage defines a novel cohesinopathy with intellectual disability and behavioral problems. <i>Human Molecular Genetics</i> , 2015 , 24, 7171-81	5.6	24
75	Lethal neonatal rigidity and multifocal seizure syndrome--report of another family with a BRAT1 mutation. <i>European Journal of Paediatric Neurology</i> , 2015 , 19, 240-2	3.8	18
74	Keppen-Lubinsky syndrome is caused by mutations in the inwardly rectifying K ⁺ channel encoded by KCNJ6. <i>American Journal of Human Genetics</i> , 2015 , 96, 295-300	11	72
73	Expanding the clinical and mutational spectrum of Kaufman oculocerebrofacial syndrome with biallelic UBE3B mutations. <i>Human Genetics</i> , 2014 , 133, 939-49	6.3	22
72	Two siblings with early infantile myoclonic encephalopathy due to mutation in the gene encoding mitochondrial glutamate/H ⁺ symporter SLC25A22. <i>European Journal of Paediatric Neurology</i> , 2014 , 18, 801-5	3.8	23
71	Homozygous truncating PTPRF mutation causes athelia. <i>Human Genetics</i> , 2014 , 133, 1041-7	6.3	6
70	Expanding the mutational spectrum of CRLF1 in Crisponi/CISS1 syndrome. <i>Human Mutation</i> , 2014 , 35, 424-33	4.7	17
69	Dominant mutations in GRHL3 cause Van der Woude Syndrome and disrupt oral periderm development. <i>American Journal of Human Genetics</i> , 2014 , 94, 23-32	11	146
68	A novel mutation in the endosomal Na ⁺ /H ⁺ exchanger NHE6 (SLC9A6) causes Christianson syndrome with electrical status epilepticus during slow-wave sleep (ESES). <i>Epilepsy Research</i> , 2014 , 108, 811-5	3	33
67	Broad phenotypic heterogeneity due to a novel SCN1A mutation in a family with genetic epilepsy with febrile seizures plus. <i>Journal of Child Neurology</i> , 2014 , 29, 221-6	2.5	34
66	Exome sequencing reveals SYCE1 mutation associated with autosomal recessive primary ovarian insufficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2014 , 99, E2129-32	5.6	104
65	Identification of a novel mutation in the PNLIP gene in two brothers with congenital pancreatic lipase deficiency. <i>Journal of Lipid Research</i> , 2014 , 55, 307-12	6.3	21
64	A novel splice-site mutation in the AAGAB gene segregates with hereditary punctate palmoplantar keratoderma and congenital dysplasia of the hip in a large family. <i>Clinical and Experimental Dermatology</i> , 2014 , 39, 182-6	1.8	9

63	Biallelic SZT2 mutations cause infantile encephalopathy with epilepsy and dysmorphic corpus callosum. <i>American Journal of Human Genetics</i> , 2013 , 93, 524-9	11	51
62	Microcephaly thin corpus callosum intellectual disability syndrome caused by mutated TAF2. <i>Pediatric Neurology</i> , 2013 , 49, 411-416.e1	2.9	19
61	Mutation spectrum in RAB3GAP1, RAB3GAP2, and RAB18 and genotype-phenotype correlations in warburg micro syndrome and Martsolf syndrome. <i>Human Mutation</i> , 2013 , 34, 686-96	4.7	86
60	Mutations in PIK3R1 cause SHORT syndrome. <i>American Journal of Human Genetics</i> , 2013 , 93, 158-66	11	125
59	A novel mutation in the GAN gene causes an intermediate form of giant axonal neuropathy in an Arab-Israeli family. <i>European Journal of Paediatric Neurology</i> , 2013 , 17, 259-64	3.8	6
58	Mutations in GMPPA cause a glycosylation disorder characterized by intellectual disability and autonomic dysfunction. <i>American Journal of Human Genetics</i> , 2013 , 93, 727-34	11	45
57	Heterogeneity of mutational mechanisms and modes of inheritance in auriculocondylar syndrome. <i>Journal of Medical Genetics</i> , 2013 , 50, 174-86	5.8	32
56	Phenotypic psychiatric characterization of children with Williams syndrome and response of those with ADHD to methylphenidate treatment. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2012 , 159B, 13-20	3.5	23
55	High frequency of autosomal-recessive DFNB59 hearing loss in an isolated Arab population in Israel. <i>Clinical Genetics</i> , 2012 , 82, 271-6	4	15
54	eIF2 ϵ mutation that disrupts eIF2 complex integrity links intellectual disability to impaired translation initiation. <i>Molecular Cell</i> , 2012 , 48, 641-6	17.6	55
53	Microcephaly-thin corpus callosum syndrome maps to 8q23.2-q24.12. <i>Pediatric Neurology</i> , 2012 , 46, 363-8.9	10	
52	Genotype-phenotype correlation in 22q11.2 deletion syndrome. <i>BMC Medical Genetics</i> , 2012 , 13, 122	2.1	66
51	Deficiency for the ubiquitin ligase UBE3B in a blepharophimosis-ptosis-intellectual-disability syndrome. <i>American Journal of Human Genetics</i> , 2012 , 91, 998-1010	11	62
50	Transient infantile hypertriglyceridemia, fatty liver, and hepatic fibrosis caused by mutated GPD1, encoding glycerol-3-phosphate dehydrogenase 1. <i>American Journal of Human Genetics</i> , 2012 , 90, 49-60	11	58
49	New syndrome of congenital circumferential skin folds associated with multiple congenital anomalies. <i>Pediatric Dermatology</i> , 2012 , 29, 89-95	1.9	5
48	A comparative study of hearing loss in two microdeletion syndromes: velocardiofacial (22q11.2 deletion) and Williams (7q11.23 deletion) syndromes. <i>Journal of Pediatrics</i> , 2011 , 158, 301-6	3.6	21
47	Three sibs with microcephaly, clubfeet and agenesis of corpus callosum: a new genetic syndrome?. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 1060-5	2.5	1
46	X-linked mental retardation with alacrima and achalasia-Triple A syndrome or a new syndrome?. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 1959-63	2.5	2

45	An emerging 1q21.1 deletion-associated neurodevelopmental phenotype. <i>Journal of Child Neurology</i> , 2011 , 26, 113-6	2.5	16
44	Case series: 2q33.1 microdeletion syndrome--further delineation of the phenotype. <i>Journal of Medical Genetics</i> , 2011 , 48, 290-8	5.8	46
43	Clinical, neuroradiological and genetic findings in pontocerebellar hypoplasia. <i>Brain</i> , 2011 , 134, 143-56	11.2	161
42	Multiple congenital anomalies-hypotonia-seizures syndrome is caused by a mutation in PIGN. <i>Journal of Medical Genetics</i> , 2011 , 48, 383-9	5.8	116
41	CDH3-Related Syndromes: Report on a New Mutation and Overview of the Genotype-Phenotype Correlations. <i>Molecular Syndromology</i> , 2010 , 1, 223-230	1.5	15
40	Clinical and brain imaging heterogeneity of severe microcephaly. <i>Pediatric Neurology</i> , 2010 , 43, 7-16	2.9	23
39	Terminal osseous dysplasia is caused by a single recurrent mutation in the FLNA gene. <i>American Journal of Human Genetics</i> , 2010 , 87, 146-53	11	48
38	SOBP is mutated in syndromic and nonsyndromic intellectual disability and is highly expressed in the brain limbic system. <i>American Journal of Human Genetics</i> , 2010 , 87, 694-700	11	15
37	Acute lymphoblastic leukemia in Weaver syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2010 , 152A, 383-6	2.5	22
36	Familial hydrocephalus with normal cognition and distinctive radiological features. <i>American Journal of Medical Genetics, Part A</i> , 2010 , 152A, 2743-8	2.5	5
35	Loss-of-function mutations in ATP6V0A2 impair vesicular trafficking, tropoelastin secretion and cell survival. <i>Human Molecular Genetics</i> , 2009 , 18, 2149-65	5.6	100
34	Keppen-Lubinsky syndrome: Expanding the phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2009 , 149A, 1827-9	2.5	8
33	RIN2 deficiency results in macrocephaly, alopecia, cutis laxa, and scoliosis: MACS syndrome. <i>American Journal of Human Genetics</i> , 2009 , 85, 254-63	11	70
32	A truncating mutation of TRAPPC9 is associated with autosomal-recessive intellectual disability and postnatal microcephaly. <i>American Journal of Human Genetics</i> , 2009 , 85, 897-902	11	104
31	Impaired glycosylation and cutis laxa caused by mutations in the vesicular H ⁺ -ATPase subunit ATP6V0A2. <i>Nature Genetics</i> , 2008 , 40, 32-4	36.3	279
30	tRNA splicing endonuclease mutations cause pontocerebellar hypoplasia. <i>Nature Genetics</i> , 2008 , 40, 1113-63	36.3	177
29	Autosomal recessive ichthyosis with hypotrichosis syndrome: further delineation of the phenotype. <i>Clinical Genetics</i> , 2008 , 74, 47-53	4	43
28	Genetic carrier screening for spinal muscular atrophy and spinal muscular atrophy with respiratory distress 1 in an isolated population in Israel. <i>Genetic Testing and Molecular Biomarkers</i> , 2008 , 12, 53-6		22

27	Expanding the clinical phenotype of autosomal dominant dyskeratosis congenita caused by TERT mutations. <i>Haematologica</i> , 2008 , 93, 943-4	6.6	38
26	Psychiatric morbidity with focus on obsessive-compulsive disorder in an Israeli cohort of adolescents with mild to moderate mental retardation. <i>Journal of Neural Transmission</i> , 2008 , 115, 929-36	4.3	24
25	Ethnically diverse causes of Walker-Warburg syndrome (WWS): FCMD mutations are a more common cause of WWS outside of the Middle East. <i>Human Mutation</i> , 2008 , 29, E231-41	4.7	54
24	Yunis-Varon syndrome: further delineation of the phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2008 , 146A, 532-7	2.5	14
23	Autosomal dominant isolated question mark ear. <i>American Journal of Medical Genetics, Part A</i> , 2008 , 146A, 2280-3	2.5	6
22	Clinical approaches to genetic mental retardation. <i>Israel Medical Association Journal</i> , 2008 , 10, 821-6	0.9	5
21	Autosomal recessive mental retardation syndrome with anterior maxillary protrusion and strabismus: MRAMS syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2007 , 143A, 1687-91	2.5	6
20	Allele dosage-dependent penetrance of RET proto-oncogene in an Israeli-Arab inbred family segregating Hirschsprung disease. <i>European Journal of Human Genetics</i> , 2007 , 15, 242-5	5.3	11
19	Genetic screening for autosomal recessive nonsyndromic mental retardation in an isolated population in Israel. <i>European Journal of Human Genetics</i> , 2007 , 15, 250-3	5.3	30
18	Genetics of autosomal recessive non-syndromic mental retardation: recent advances. <i>Clinical Genetics</i> , 2007 , 72, 167-74	4	22
17	Autosomal recessive ichthyosis with hypotrichosis caused by a mutation in ST14, encoding type II transmembrane serine protease matriptase. <i>American Journal of Human Genetics</i> , 2007 , 80, 467-77	11	139
16	Mutated nup62 causes autosomal recessive infantile bilateral striatal necrosis. <i>Annals of Neurology</i> , 2006 , 60, 214-22	9.4	128
15	Amniotic trisomy 11 mosaicism--is it a benign finding?. <i>Prenatal Diagnosis</i> , 2006 , 26, 778-81	3.2	8
14	Expanding the phenotypic spectrum of L1CAM-associated disease. <i>Clinical Genetics</i> , 2006 , 69, 414-9	4	24
13	Positional cloning uncovers mutations in PLCE1 responsible for a nephrotic syndrome variant that may be reversible. <i>Nature Genetics</i> , 2006 , 38, 1397-405	36.3	432
12	The CC2D1A, a member of a new gene family with C2 domains, is involved in autosomal recessive non-syndromic mental retardation. <i>Journal of Medical Genetics</i> , 2006 , 43, 203-10	5.8	108
11	G protein-coupled receptor-dependent development of human frontal cortex. <i>Science</i> , 2004 , 303, 2033-6	33.3	404
10	Etiological heterogeneity of familial periventricular heterotopia and hydrocephalus. <i>Brain and Development</i> , 2004 , 26, 326-34	2.2	49

9	Twenty-two novel mutations in the lysosomal alpha-glucosidase gene (GAA) underscore the genotype-phenotype correlation in glycogen storage disease type II. <i>Human Mutation</i> , 2004 , 23, 47-56	4-7	125
8	De novo SCN1A mutations are a major cause of severe myoclonic epilepsy of infancy. <i>Human Mutation</i> , 2003 , 21, 615-21	4-7	137
7	New syndrome of simplified gyral pattern, micromelia, dysmorphic features and early death. <i>American Journal of Medical Genetics Part A</i> , 2003 , 119A, 200-6		4
6	Bilateral frontoparietal polymicrogyria: clinical and radiological features in 10 families with linkage to chromosome 16. <i>Annals of Neurology</i> , 2003 , 53, 596-606	9-4	110
5	Branchial cyst, sensorineural deafness, congenital heart defect, and skeletal abnormalities: Branchio-oto-cardio-skeletal (BOCS) syndrome?. <i>American Journal of Medical Genetics Part A</i> , 2002 , 113, 78-81		4
4	An autosomal recessive form of bilateral frontoparietal polymicrogyria maps to chromosome 16q12.2-21. <i>American Journal of Human Genetics</i> , 2002 , 70, 1028-33	11	97
3	Seventeen novel mutations that cause profound biotinidase deficiency. <i>Molecular Genetics and Metabolism</i> , 2002 , 77, 108-11	3-7	29
2	Fragile-X carrier screening and the prevalence of premutation and full-mutation carriers in Israel. <i>American Journal of Human Genetics</i> , 2001 , 69, 351-60	11	165
1	Spectrum of genes for inherited hearing loss in the Israeli Jewish population, including the novel human deafness gene ATOH1		1