Lina Basel-Vanagaite

List of Publications by Citations

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	Positional cloning uncovers mutations in PLCE1 responsible for a nephrotic syndrome variant that may be reversible. *Nature Genetics*, **2006**, 38, 1397-405	36.3	432
133	G protein-coupled receptor-dependent development of human frontal cortex. *Science*, **2004**, 303, 2033-6	33.3	404
132	Impaired glycosylation and cutis laxa caused by mutations in the vesicular H+-ATPase subunit ATP6V0A2. *Nature Genetics*, **2008**, 40, 32-4	36.3	279
131	Identifying facial phenotypes of genetic disorders using deep learning. *Nature Medicine*, **2019**, 25, 60-64	50.5	229
130	tRNA splicing endonuclease mutations cause pontocerebellar hypoplasia. *Nature Genetics*, **2008**, 40, 1113-8	36.3	177
129	Fragile-X carrier screening and the prevalence of premutation and full-mutation carriers in Israel. *American Journal of Human Genetics*, **2001**, 69, 351-60	11	165
128	Clinical, neuroradiological and genetic findings in pontocerebellar hypoplasia. *Brain*, **2011**, 134, 143-56	11.2	161
127	Dominant mutations in GRHL3 cause Van der Woude Syndrome and disrupt oral periderm development. *American Journal of Human Genetics*, **2014**, 94, 23-32	11	146
126	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, 467-77	11	139
125	De novo SCN1A mutations are a major cause of severe myoclonic epilepsy of infancy. *Human Mutation*, **2003**, 21, 615-21	4.7	137
124	Mutated nup62 causes autosomal recessive infantile bilateral striatal necrosis. *Annals of Neurology*, **2006**, 60, 214-22	9.4	128
123	Mutations in PIK3R1 cause SHORT syndrome. *American Journal of Human Genetics*, **2013**, 93, 158-66	11	125
122	Twenty-two novel mutations in the lysosomal alpha-glucosidase gene (GAA) underscore the genotype-phenotype correlation in glycogen storage disease type II. *Human Mutation*, **2004**, 23, 47-56	4.7	125
121	Multiple congenital anomalies-hypotonia-seizures syndrome is caused by a mutation in PIGN. *Journal of Medical Genetics*, **2011**, 48, 383-9	5.8	116
120	Bilateral frontoparietal polymicrogyria: clinical and radiological features in 10 families with linkage to chromosome 16. *Annals of Neurology*, **2003**, 53, 596-606	9.4	110
119	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*, **2006**, 43, 203-10	5.8	108
118	Exome sequencing reveals SYCE1 mutation associated with autosomal recessive primary ovarian insufficiency. *Journal of Clinical Endocrinology and Metabolism*, **2014**, 99, E2129-32	5.6	104

117	A truncating mutation of TRAPPC9 is associated with autosomal-recessive intellectual disability and postnatal microcephaly. *American Journal of Human Genetics*, **2009**, 85, 897-902	11	104
116	Loss-of-function mutations in ATP6V0A2 impair vesicular trafficking, tropoelastin secretion and cell survival. *Human Molecular Genetics*, **2009**, 18, 2149-65	5.6	100
115	An autosomal recessive form of bilateral frontoparietal polymicrogyria maps to chromosome 16q12.2-21. *American Journal of Human Genetics*, **2002**, 70, 1028-33	11	97
114	Mutation spectrum in RAB3GAP1, RAB3GAP2, and RAB18 and genotype-phenotype correlations in warburg micro syndrome and Martsolf syndrome. *Human Mutation*, **2013**, 34, 686-96	4.7	86
113	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	11	72
112	RIN2 deficiency results in macrocephaly, alopecia, cutis laxa, and scoliosis: MACS syndrome. *American Journal of Human Genetics*, **2009**, 85, 254-63	11	70
111	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	11	68
110	Genotype-phenotype correlation in 22q11.2 deletion syndrome. *BMC Medical Genetics*, **2012**, 13, 122	2.1	66
109	Deficiency for the ubiquitin ligase UBE3B in a blepharophimosis-ptosis-intellectual-disability syndrome. *American Journal of Human Genetics*, **2012**, 91, 998-1010	11	62
108	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	11	58
107	Recognition of the Cornelia de Lange syndrome phenotype with facial dysmorphology novel analysis. *Clinical Genetics*, **2016**, 89, 557-63	4	56
106	eIF2 mutation that disrupts eIF2 complex integrity links intellectual disability to impaired translation initiation. *Molecular Cell*, **2012**, 48, 641-6	17.6	55
105	Ethnically diverse causes of Walker-Warburg syndrome (WWS): FCMD mutations are a more common cause of WWS outside of the Middle East. *Human Mutation*, **2008**, 29, E231-41	4.7	54
104	Biallelic SZT2 mutations cause infantile encephalopathy with epilepsy and dysmorphic corpus callosum. *American Journal of Human Genetics*, **2013**, 93, 524-9	11	51
103	Etiological heterogeneity of familial periventricular heterotopia and hydrocephalus. *Brain and Development*, **2004**, 26, 326-34	2.2	49
102	Terminal osseous dysplasia is caused by a single recurrent mutation in the FLNA gene. *American Journal of Human Genetics*, **2010**, 87, 146-53	11	48
101	Case series: 2q33.1 microdeletion syndrome--further delineation of the phenotype. *Journal of Medical Genetics*, **2011**, 48, 290-8	5.8	46
100	Mutations in GMPPA cause a glycosylation disorder characterized by intellectual disability and autonomic dysfunction. *American Journal of Human Genetics*, **2013**, 93, 727-34	11	45

99	Autosomal recessive ichthyosis with hypotrichosis syndrome: further delineation of the phenotype. *Clinical Genetics*, **2008**, 74, 47-53	4	43
98	Chromosomal microarray vs. NIPS: analysis of 5541 low-risk pregnancies. *Genetics in Medicine*, **2019**, 21, 2462-2467	8.1	41
97	Expanding the clinical phenotype of autosomal dominant dyskeratosis congenita caused by TERT mutations. *Haematologica*, **2008**, 93, 943-4	6.6	38
96	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, 221-6	2.5	34
95	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, 811-5	3	33
94	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	8.1	32
93	Heterogeneity of mutational mechanisms and modes of inheritance in auriculocondylar syndrome. *Journal of Medical Genetics*, **2013**, 50, 174-86	5.8	32
92	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	8.1	32
91	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	5.6	30
90	Genetic screening for autosomal recessive nonsyndromic mental retardation in an isolated population in Israel. *European Journal of Human Genetics*, **2007**, 15, 250-3	5.3	30
89	Seventeen novel mutations that cause profound biotinidase deficiency. *Molecular Genetics and Metabolism*, **2002**, 77, 108-11	3.7	29
88	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	2.5	27
87	Homozygous mutations in VAMP1 cause a presynaptic congenital myasthenic syndrome. *Annals of Neurology*, **2017**, 81, 597-603	9.4	26
86	Increased STAG2 dosage defines a novel cohesinopathy with intellectual disability and behavioral problems. *Human Molecular Genetics*, **2015**, 24, 7171-81	5.6	24
85	Psychiatric morbidity with focus on obsessive-compulsive disorder in an Israeli cohort of adolescents with mild to moderate mental retardation. *Journal of Neural Transmission*, **2008**, 115, 929-36	4.3	24
84	Expanding the phenotypic spectrum of L1CAM-associated disease. *Clinical Genetics*, **2006**, 69, 414-9	4	24
83	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, 801-5	3.8	23
82	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	3.5	23

81	Clinical and brain imaging heterogeneity of severe microcephaly. *Pediatric Neurology*, **2010**, 43, 7-16	2.9	23
80	Large Intragenic Deletion in DSTYK Underlies Autosomal-Recessive Complicated Spastic Paraparesis, SPG23. *American Journal of Human Genetics*, **2017**, 100, 364-370	11	22
79	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogryposis. *American Journal of Human Genetics*, **2019**, 105, 689-705	11	22
78	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	8.1	22
77	Expanding the clinical and mutational spectrum of Kaufman oculocerebrofacial syndrome with biallelic UBE3B mutations. *Human Genetics*, **2014**, 133, 939-49	6.3	22
76	Acute lymphoblastic leukemia in Weaver syndrome. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 383-6	2.5	22
75	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, 53-6		22
74	Genetics of autosomal recessive non-syndromic mental retardation: recent advances. *Clinical Genetics*, **2007**, 72, 167-74	4	22
73	Identification of a novel mutation in the PNLIP gene in two brothers with congenital pancreatic lipase deficiency. *Journal of Lipid Research*, **2014**, 55, 307-12	6.3	21
72	A comparative study of hearing loss in two microdeletion syndromes: velocardiofacial (22q11.2 deletion) and Williams (7q11.23 deletion) syndromes. *Journal of Pediatrics*, **2011**, 158, 301-6	3.6	21
71	Microcephaly thin corpus callosum intellectual disability syndrome caused by mutated TAF2. *Pediatric Neurology*, **2013**, 49, 411-416.e1	2.9	19
70	Substrate interaction defects in histidyl-tRNA synthetase linked to dominant axonal peripheral neuropathy. *Human Mutation*, **2018**, 39, 415-432	4.7	19
69	Lethal neonatal rigidity and multifocal seizure syndrome--report of another family with a BRAT1 mutation. *European Journal of Paediatric Neurology*, **2015**, 19, 240-2	3.8	18
68	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. *Brain*, **2020**, 143, 55-68	11.2	18
67	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	3.8	17
66	Expanding the mutational spectrum of CRLF1 in Crisponi/CISS1 syndrome. *Human Mutation*, **2014**, 35, 424-33	4.7	17
65	Lysine acetyltransferase 8 is involved in cerebral development and syndromic intellectual disability. *Journal of Clinical Investigation*, **2020**, 130, 1431-1445	15.9	17
64	Bayesian-based noninvasive prenatal diagnosis of single-gene disorders. *Genome Research*, **2019**, 29, 428-438	9.7	16

63	An emerging 1q21.1 deletion-associated neurodevelopmental phenotype. *Journal of Child Neurology*, **2011**, 26, 113-6	2.5	16
62	High frequency of autosomal-recessive DFNB59 hearing loss in an isolated Arab population in Israel. *Clinical Genetics*, **2012**, 82, 271-6	4	15
61	CDH3-Related Syndromes: Report on a New Mutation and Overview of the Genotype-Phenotype Correlations. *Molecular Syndromology*, **2010**, 1, 223-230	1.5	15
60	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	11	15
59	Whole-exome sequencing reveals POC5 as a novel gene associated with autosomal recessive retinitis pigmentosa. *Human Molecular Genetics*, **2018**, 27, 614-624	5.6	14
58	Yunis-Varon syndrome: further delineation of the phenotype. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 532-7	2.5	14
57	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, 711-5	3.8	13
56	Bi-allelic ADARB1 Variants Associated with Microcephaly, Intellectual Disability, and Seizures. *American Journal of Human Genetics*, **2020**, 106, 467-483	11	12
55	Homozygous MED25 mutation implicated in eye-intellectual disability syndrome. *Human Genetics*, **2015**, 134, 577-87	6.3	11
54	Allele dosage-dependent penetrance of RET proto-oncogene in an Israeli-Arab inbred family segregating Hirschsprung disease. *European Journal of Human Genetics*, **2007**, 15, 242-5	5.3	11
53	Microcephaly-thin corpus callosum syndrome maps to 8q23.2-q24.12. *Pediatric Neurology*, **2012**, 46, 363-8	2.9	10
52	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	5.3	10
51	Kaufman oculocerebrofacial syndrome: Novel UBE3B mutations and clinical features in four unrelated patients. *American Journal of Medical Genetics, Part A*, **2018**, 176, 187-193	2.5	10
50	De novo variants in PAK1 lead to intellectual disability with macrocephaly and seizures. *Brain*, **2019**, 142, 3351-3359	11.2	9
49	Cytogenetic analysis in fetuses with late onset abnormal sonographic findings. *Journal of Perinatal Medicine*, **2018**, 46, 975-982	2.7	9
48	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	4.4	9
47	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. *Clinical and Experimental Dermatology*, **2014**, 39, 182-6	1.8	9
46	Intra-familial Variation in Clinical Phenotype of CARD14-related Psoriasis. *Acta Dermato-Venereologica*, **2016**, 96, 885-887	2.2	9

45	Keppen-Lubinsky syndrome: Expanding the phenotype. *American Journal of Medical Genetics, Part A* , **2009**, 149A, 1827-9	2.5	8
44	Amniotic trisomy 11 mosaicism--is it a benign finding?. *Prenatal Diagnosis*, **2006**, 26, 778-81	3.2	8
43	Spectrum of genes for inherited hearing loss in the Israeli Jewish population, including the novel human deafness gene ATOH1. *Clinical Genetics*, **2020**, 98, 353-364	4	8
42	X-linked elliptocytosis with impaired growth is related to mutated AMMECR1. *Gene*, **2017**, 606, 47-52	3.8	6
41	A founder mutation in ADAMTSL4 causes early-onset bilateral ectopia lentis among Jews of Bukharian origin. *Molecular Genetics and Metabolism*, **2016**, 117, 38-41	3.7	6
40	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	4.4	6
39	Homozygous truncating PTPRF mutation causes athelia. *Human Genetics*, **2014**, 133, 1041-7	6.3	6
38	A novel mutation in the GAN gene causes an intermediate form of giant axonal neuropathy in an Arab-Israeli family. *European Journal of Paediatric Neurology*, **2013**, 17, 259-64	3.8	6
37	Autosomal recessive mental retardation syndrome with anterior maxillary protrusion and strabismus: MRAMS syndrome. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 1687-91	2.5	6
36	Autosomal dominant isolated question mark ear. *American Journal of Medical Genetics, Part A*, **2008** , 146A, 2280-3	2.5	6
35	Paediatric systemic lupus erythematosus as a manifestation of constitutional mismatch repair deficiency. *Journal of Medical Genetics*, **2020**, 57, 505-508	5.8	5
34	The rare 13q33-q34 microdeletions: eight new patients and review of the literature. *Human Genetics*, **2019**, 138, 1145-1153	6.3	5
33	New syndrome of congenital circumferential skin folds associated with multiple congenital anomalies. *Pediatric Dermatology*, **2012**, 29, 89-95	1.9	5
32	Familial hydrocephalus with normal cognition and distinctive radiological features. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 2743-8	2.5	5
31	Mild Phenotype of Wolfram Syndrome Associated With a Common Pathogenic Variant Is Predicted by a Structural Model of Wolframin. *Neurology: Genetics*, **2021**, 7, e578	3.8	5
30	Clinical approaches to genetic mental retardation. *Israel Medical Association Journal*, **2008**, 10, 821-6	0.9	5
29	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		4
28	New syndrome of simplified gyral pattern, micromelia, dysmorphic features and early death. *American Journal of Medical Genetics Part A*, **2003**, 119A, 200-6		4

27	Impact of a national population-based carrier-screening program on spinal muscular atrophy births. *Neuromuscular Disorders*, **2020**, 30, 970-974	2.9	4
26	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	4.2	4
25	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	2.5	4
24	Based on a cohort of 52,879 microarrays, recurrent intragenic FBN2 deletion encompassing exons 1-8 does not cause Beals syndrome. *European Journal of Medical Genetics*, **2020**, 63, 104008	2.6	3
23	Should We Report 15q11.2 BP1-BP2 Deletions and Duplications in the Prenatal Setting?. *Journal of Clinical Medicine*, **2020**, 9,	5.1	3
22	Polymicrogyria and myoclonic epilepsy in autosomal recessive cutis laxa type 2A. *Neurogenetics*, **2016**, 17, 251-257	3	3
21	Biallelic variants in ETV2 in a family with congenital heart defects, vertebral abnormalities and preaxial polydactyly. *European Journal of Medical Genetics*, **2021**, 64, 104124	2.6	3
20	X-linked mental retardation with alacrima and achalasia-Triple A syndrome or a new syndrome?. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1959-63	2.5	2
19	Biallelic Truncating Variants in the Muscular A-Type Lamin-Interacting Protein (MLIP) Gene Cause Myopathy with Hyper-CKemia.. *European Journal of Neurology*, **2021**,	6	2
18	When phenotype does not match genotype: importance of "real-time" refining of phenotypic information for exome data interpretation. *Genetics in Medicine*, **2021**, 23, 215-221	8.1	2
17	The role of phenotype-based search approaches using public online databases in diagnostics of Mendelian disorders. *Genetics in Medicine*, **2021**, 23, 1095-1100	8.1	2
16	Two intronic cis-acting variants in both alleles of the POLR3A gene cause progressive spastic ataxia with hypodontia. *Clinical Genetics*, **2021**, 99, 713-718	4	2
15	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	8.1	2
14	Three sibs with microcephaly, clubfeet and agenesis of corpus callosum: a new genetic syndrome?. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1060-5	2.5	1
13	Prenatal and postnatal chromosomal microarray analysis in 885 cases of various congenital heart defects.. *Archives of Gynecology and Obstetrics*, **2022**, 1	2.5	1
12	Di-genic inheritance of germline POLE and PMS2 pathogenic variants causes a unique condition associated with pediatric cancer predisposition.. *Clinical Genetics*, **2021**,	4	1
11	In-silico phenotype prediction by normal mode variant analysis in TUBB4A-related disease.. *Scientific Reports*, **2022**, 12, 58	4.9	1
10	Residual risk for clinically significant copy number variants in low-risk pregnancies, following exclusion of noninvasive prenatal screening-detectable findings. *American Journal of Obstetrics and Gynecology*, **2021**,	6.4	1

9	Challenges in variant interpretation in prenatal exome sequencing.. *European Journal of Medical Genetics*, **2021**, 65, 104410	2.6	1
8	A study of normal copy number variations in Israeli population. *Human Genetics*, **2021**, 140, 553-563	6.3	1
7	Spectrum of genes for inherited hearing loss in the Israeli Jewish population, including the novel human deafness gene ATOH1		1
6	Physician anxiety or maternal choice?. *American Journal of Obstetrics and Gynecology*, **2021**,	6.4	1
5	Is it time to report carrier state for recessive disorders in every microarray analysis?-A pilot model based on hearing loss genes deletions. *European Journal of Human Genetics*, **2021**, 29, 1292-1300	5.3	0
4	Congenital Thrombocytopenia Associated with a Heterozygous Variant in the Gene Encoding a Transcription Factor Essential for Megakaryopoiesis.. *Platelets*, **2022**, 1-4	3.6	
3	DYRK1B haploinsufficiency in a family with metabolic syndrome and abnormal cognition. *Clinical Genetics*, **2021**, 101, 265	4	
2	The phenotype of 15 cases with rare 8q24.13-q24.3 deletions-A new syndrome or still an enigma?. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1461-1467	2.5	
1	A nonsense variant in the second exon of the canonical transcript of NSD1 does not cause Sotos syndrome. *American Journal of Medical Genetics, Part A*, **2021**,	2.5	