Boris Keren

List of Publications by Year in descending order

49 papers	1,679 citations	361413 20 h-index	330143 37 g-index
54 all docs	54 docs citations	54 times ranked	3972 citing authors

#	Article	IF	Citations
1	Recurrent de novo missense variants across multiple histone H4 genes underlie a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 750-758.	6.2	13
2	Heterozygous variants in CTR9, which encodes a major component of the PAF1 complex, are associated with a neurodevelopmental disorder. Genetics in Medicine, 2022, , .	2.4	1
3	SEMA6B variants cause intellectual disability and alter dendritic spine density and axon guidance. Human Molecular Genetics, 2022, 31, 3325-3340.	2.9	5
4	Biallelic variants in <scp>ZNF142</scp> lead to a syndromic neurodevelopmental disorder. Clinical Genetics, 2022, 102, 98-109.	2.0	6
5	Phenotypic expansion of the <scp>BPTF</scp>â€related neurodevelopmental disorder with dysmorphic facies and distal limb anomalies. American Journal of Medical Genetics, Part A, 2021, 185, 1366-1378.	1.2	8
6	Comprehensive study of 28 individuals with SIN3A-related disorder underscoring the associated mild cognitive and distinctive facial phenotype. European Journal of Human Genetics, 2021, 29, 625-636.	2.8	17
7	Demyelinating motor neuropathy associated with a homozygous <scp>GPT2</scp> pathogenic variant. Muscle and Nerve, 2021, 63, E41-E44.	2.2	2
8	Dysregulation of the NRG1/ERBB pathway causes a developmental disorder with gastrointestinal dysmotility in humans. Journal of Clinical Investigation, 2021, 131, .	8.2	24
9	Monoallelic and bi-allelic variants in NCDN cause neurodevelopmental delay, intellectual disability, and epilepsy. American Journal of Human Genetics, 2021, 108, 739-748.	6.2	15
10	Recurrence of an early postzygotic rescue of an inherited unbalanced translocation resulting in mosaic segmental uniparental isodisomy of chromosome 11q in siblings. American Journal of Medical Genetics, Part A, 2021, 185, 3057-3061.	1.2	1
11	Variants in the degron of AFF3 are associated with intellectual disability, mesomelic dysplasia, horseshoe kidney, and epileptic encephalopathy. American Journal of Human Genetics, 2021, 108, 857-873.	6.2	19
12	Elp2 mutations perturb the epitranscriptome and lead to a complex neurodevelopmental phenotype. Nature Communications, 2021, 12, 2678.	12.8	26
13	CSNK2B: A broad spectrum of neurodevelopmental disability and epilepsy severity. Epilepsia, 2021, 62, e103-e109.	5.1	13
14	SCN1A-related epilepsy with recessive inheritance: Two further families. European Journal of Paediatric Neurology, 2021, 33, 121-124.	1.6	4
15	Integrative approach to interpret DYRK1A variants, leading to a frequent neurodevelopmental disorder. Genetics in Medicine, 2021, 23, 2150-2159.	2.4	21
16	Dysfunctional Homozygous VRK1-D263G Variant Impairs the Assembly of Cajal Bodies and DNA Damage Response in Hereditary Spastic Paraplegia. Neurology: Genetics, 2021, 7, e624.	1.9	2
17	Tremor-like subcortical myoclonus in STXBP1 encephalopathy. European Journal of Paediatric Neurology, 2021, 34, 62-66.	1.6	6
18	Expanding the Phenotypic Spectrum of GPI Anchoring Deficiency Due to Biallelic Variants in GPAA1. Neurology: Genetics, 2021, 7, e631.	1.9	2

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19	The complete loss of function of the SMS gene results in a severe form of Snyder-Robinson syndrome. European Journal of Medical Genetics, 2020, 63, 103777.	1.3	17
20	The CHD4-related syndrome: a comprehensive investigation of the clinical spectrum, genotypeâ€“phenotype correlations, and molecular basis. Genetics in Medicine, 2020, 22, 389-397.	2.4	53
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.	1.3	42
22	Expanding the genetic and phenotypic relevance of KCNB1 variants in developmental and epileptic encephalopathies: 27 new patients and overview of the literature. Human Mutation, 2020, 41, 69-80.	2.5	33
23	Developmental and epilepsy spectrum of KCNB1 encephalopathy with longâ€term outcome. Epilepsia, 2020, 61, 2461-2473.	5.1	17
24	Missense variant contribution to USP9X-female syndrome. Npj Genomic Medicine, 2020, 5, 53.	3.8	17
25	De Novo Variants in CNOT1, a Central Component of the CCR4-NOT Complex Involved in Gene Expression and RNA and Protein Stability, Cause Neurodevelopmental Delay. American Journal of Human Genetics, 2020, 107, 164-172.	6.2	37
26	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 356-370.	6.2	171
27	Increased diagnostic yield in complex dystonia through exome sequencing. Parkinsonism and Related Disorders, 2020, 74, 50-56.	2.2	34
28	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nature Communications, 2020, 11, 595.	12.8	35
29	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
30	Haploinsufficiency of the Notch Ligand DLL1 Causes Variable Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 105, 631-639.	6.2	42
31	SETD2 related overgrowth syndrome: Presentation of four new patients and review of the literature. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 509-518.	1.6	24
32	The epileptology of GNB5 encephalopathy. Epilepsia, 2019, 60, e121-e127.	5.1	13
33	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	6.2	56
34	Novel GABRA2 variants in epileptic encephalopathy and intellectual disability with seizures. Brain, 2019, 142, e15-e15.	7.6	12
35	Biallelic variants in LARS2 and KARS cause deafness and (ovario)leukodystrophy. Neurology, 2019, 92, e1225-e1237.	1.1	32
36	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. Genetics in Medicine, 2019, 21, 837-849.	2.4	47

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37	Subcortical Myoclonus and Associated Dystonia in 22q11.2 Deletion Syndrome. Tremor and Other Hyperkinetic Movements, 2019, 10, .	2.0	2
38	KIF1C Variants Are Associated with Hypomyelination, Ataxia, Tremor, and Dystonia in Fraternal Twins. Tremor and Other Hyperkinetic Movements, 2019, 9, .	2.0	1
39	Defining the phenotypic spectrum of SLC6A1 mutations. Epilepsia, 2018, 59, 389-402.	5.1	99
40	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. Nature Communications, 2018, 9, 4619.	12.8	70
41	De novo mutations in the GTP/GDP-binding region of RALA, a RAS-like small GTPase, cause intellectual disability and developmental delay. PLoS Genetics, 2018, 14, e1007671.	3.5	16
42	A homozygous KAT2B variant modulates the clinical phenotype of ADD3 deficiency in humans and flies. PLoS Genetics, 2018, 14, e1007386.	3.5	17
43	Mutation spectrum in the ABCC6 gene and genotypeâ€“phenotype correlations in a French cohort with pseudoxanthoma elasticum. Genetics in Medicine, 2017, 19, 909-917.	2.4	54
44	Copy Number Variations Found in Patients with a Corpus Callosum Abnormality and Intellectual Disability. Journal of Pediatrics, 2017, 185, 160-166.e1.	1.8	25
45	A framework to identify contributing genes in patients with Phelan-McDermid syndrome. Npj Genomic Medicine, 2017, 2, 32.	3.8	58
46	The Number of Genomic Copies at the 16p11.2 Locus Modulates Language, Verbal Memory, and Inhibition. Biological Psychiatry, 2016, 80, 129-139.	1.3	78
47	Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. JAMA Psychiatry, 2016, 73, 20.	11.0	195
48	The advantages of SNP arrays over CGH arrays. Molecular Cytogenetics, 2014, 7, I31.	0.9	6
49	CNS malformations in Knobloch syndrome with splice mutation inCOL18A1 gene. American Journal of Medical Genetics, Part A, 2007, 143A, 1514-1518.	1.2	34