Thomas Besnard

List of Publications by Year
in descending order

26	1,245	471371 17	501076 28
papers	citations	h-index	g-index

31	31	31	3296
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	2.6	136
2	Usher syndrome type 2 caused by activation of an USH2A pseudoexon: Implications for diagnosis and therapy. Human Mutation, 2012, 33, 104-108.	1.1	102
3	Germline De Novo Mutations in GNB1 Cause Severe Neurodevelopmental Disability, Hypotonia, and Seizures. American Journal of Human Genetics, 2016, 98, 1001-1010.	2.6	102
4	Complex Compound Inheritance of Lethal Lung Developmental Disorders Due to Disruption of the TBX-FGF Pathway. American Journal of Human Genetics, 2019, 104, 213-228.	2.6	90
5	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 100, 352-363.	2.6	86
6	Identification of a new VHL exon and complex splicing alterations in familial erythrocytosis or von Hippel-Lindau disease. Blood, 2018, 132, 469-483.	0.6	70
7	De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 101, 716-724.	2.6	66
8	Non-USH2A mutations in USH2 patients. Human Mutation, 2012, 33, 504-510.	1.1	57
9	Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. American Journal of Human Genetics, 2017, 100, 676-688.	2.6	54
10	Experience of targeted Usher exome sequencing as a clinical test. Molecular Genetics & Genomic Medicine, 2014, 2, 30-43.	0.6	53
11	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. American Journal of Human Genetics, 2018, 102, 744-759.	2.6	51
12	Four-Year Follow-up of Diagnostic Service in USH1 Patients. , 2011, 52, 4063.		47
13	De Novo Truncating Variants in SON Cause Intellectual Disability, Congenital Malformations, and Failure to Thrive. American Journal of Human Genetics, 2016, 99, 720-727.	2.6	45
14	Expanding the clinical spectrum of hereditary fibrosing poikiloderma with tendon contractures, myopathy and pulmonary fibrosis due to FAM111B mutations. Orphanet Journal of Rare Diseases, 2015, 10, 135.	1.2	42
15	De Novo Truncating Mutations in the Kinetochore-Microtubules Attachment GeneCHAMP1Cause Syndromic Intellectual Disability. Human Mutation, 2016, 37, 354-358.	1.1	40
16	Biallelic pathogenic variants in the lanosterol synthase gene LSS involved in the cholesterol biosynthesis cause alopecia with intellectual disability, a rare recessive neuroectodermal syndrome. Genetics in Medicine, 2019, 21, 2025-2035.	1.1	40
17	Haploinsufficiency of the E3 ubiquitin-protein ligase gene TRIP12 causes intellectual disability with or without autism spectrum disorders, speech delay, and dysmorphic features. Human Genetics, 2017, 136, 377-386.	1.8	36
18	Nasal epithelial cells are a reliable source to study splicing variants in Usher syndrome. Human Mutation, 2010, 31, 734-741.	1.1	29

#	Article	IF	Citations
19	A new mutation of <scp>ANO</scp>6 in two familial cases of Scott syndrome. British Journal of Haematology, 2018, 180, 750-752.	1.2	15
20	Haploinsufficiency of the Sin3/HDAC corepressor complex member SIN3B causes a syndromic intellectual disability/autism spectrum disorder. American Journal of Human Genetics, 2021, 108, 929-941.	2.6	15
21	The contribution of GPR98 and DFNB31 genes to a Spanish Usher syndrome type 2 cohort. Molecular Vision, 2013, 19, 367-73.	1.1	13
22	Stankiewicz-Isidor syndrome: expanding the clinical and molecular phenotype. Genetics in Medicine, 2022, 24, 179-191.	1.1	9
23	CUGC for hereditary fibrosing poikiloderma with tendon contractures, myopathy, and pulmonary fibrosis (POIKTMP). European Journal of Human Genetics, 2016, 24, 779-779.	1.4	8
24	New splicing pathogenic variant in EBP causing extreme familial variability of Conradiâ€“HÃ¼nermannâ€“Happle Syndrome. European Journal of Human Genetics, 2018, 26, 1784-1790.	1.4	7
25	Rare germline heterozygous missense variants in BRCA1-associated protein 1, BAP1, cause a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2022, 109, 361-372.	2.6	6
26	Lossâ€ofâ€function variants in ARHGEF9 are associated with an Xâ€linked intellectual disability dominant disorder. Human Mutation, 2021, 42, 498-505.	1.1	1