Thomas Besnard

List of Publications by Year
in descending order

26 papers	1,245 citations	471509 17 h-index	501196 28 g-index
31 all docs	31 docs citations	31 times ranked	3296 citing authors

#	Article	IF	Citations
1	Stankiewicz-Isidor syndrome: expanding the clinical and molecular phenotype. Genetics in Medicine, 2022, 24, 179-191.	2.4	9
2	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.	6.2	6
3	Lossâ€ofâ€function variants in ARHGEF9 are associated with an Xâ€linked intellectual disability dominant disorder. Human Mutation, 2021, 42, 498-505.	2.5	1
4	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.	6.2	15
5	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.	2.4	40
6	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.	6.2	90
7	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.	6.2	51
8	A new mutation of <scp>ANO</scp>6 in two familial cases of Scott syndrome. British Journal of Haematology, 2018, 180, 750-752.	2.5	15
9	New splicing pathogenic variant in EBP causing extreme familial variability of Conradiâ€“HÃ¼nermannâ€“Happle Syndrome. European Journal of Human Genetics, 2018, 26, 1784-1790.	2.8	7
10	Identification of a new VHL exon and complex splicing alterations in familial erythrocytosis or von Hippel-Lindau disease. Blood, 2018, 132, 469-483.	1.4	70
11	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 100, 352-363.	6.2	86
12	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.	3.8	36
13	Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. American Journal of Human Genetics, 2017, 100, 676-688.	6.2	54
14	De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 101, 716-724.	6.2	66
15	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	6.2	136
16	Germline De Novo Mutations in GNB1 Cause Severe Neurodevelopmental Disability, Hypotonia, and Seizures. American Journal of Human Genetics, 2016, 98, 1001-1010.	6.2	102
17	De Novo Truncating Variants in SON Cause Intellectual Disability, Congenital Malformations, and Failure to Thrive. American Journal of Human Genetics, 2016, 99, 720-727.	6.2	45
18	CUGC for hereditary fibrosing poikiloderma with tendon contractures, myopathy, and pulmonary fibrosis (POIKTMP). European Journal of Human Genetics, 2016, 24, 779-779.	2.8	8

#	Article	IF	Citations
19	De Novo Truncating Mutations in the Kinetochore-Microtubules Attachment GeneCHAMP1Cause Syndromic Intellectual Disability. Human Mutation, 2016, 37, 354-358.	2.5	40
20	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.	2.7	42
21	Experience of targeted Usher exome sequencing as a clinical test. Molecular Genetics & Genomic Medicine, 2014, 2, 30-43.	1.2	53
22	The contribution of GPR98 and DFNB31 genes to a Spanish Usher syndrome type 2 cohort. Molecular Vision, 2013, 19, 367-73.	1.1	13
23	Usher syndrome type 2 caused by activation of an USH2A pseudoexon: Implications for diagnosis and therapy. Human Mutation, 2012, 33, 104-108.	2.5	102
24	Non-USH2A mutations in USH2 patients. Human Mutation, 2012, 33, 504-510.	2.5	57
25	Four-Year Follow-up of Diagnostic Service in USH1 Patients. , 2011, 52, 4063.		47
26	Nasal epithelial cells are a reliable source to study splicing variants in Usher syndrome. Human Mutation, 2010, 31, 734-741.	2.5	29