Julien Buratti

List of Publications by Year
in descending order

27 papers	1,054 citations	623734 14 h-index	454955 30 g-index
32 all docs	32 docs citations	32 times ranked	2498 citing authors

#	Article	IF	Citations
1	Patients with KCNH1-related intellectual disability without distinctive features of Zimmermann-Laband/Temple-Baraitser syndrome. Journal of Medical Genetics, 2022, 59, 505-510.	3.2	5
2	Biallelic variants in <scp>ZNF142</scp> lead to a syndromic neurodevelopmental disorder. Clinical Genetics, 2022, 102, 98-109.	2.0	6
3	De novo variants in SIAH1, encoding an E3 ubiquitin ligase, are associated with developmental delay, hypotonia and dysmorphic features. Journal of Medical Genetics, 2021, 58, 205-212.	3.2	6
4	Identification of biallelic germline variants of SRP68 in a sporadic case with severe congenital neutropenia. Haematologica, 2021, 106, 1216-1219.	3.5	10
5	Functional characterization of ABCC8 variants of unknown significance based on bioinformatics predictions, splicing assays, and protein analyses: Benefits for the accurate diagnosis of congenital hyperinsulinism. Human Mutation, 2021, 42, 408-420.	2.5	6
6	SCN1A-related epilepsy with recessive inheritance: Two further families. European Journal of Paediatric Neurology, 2021, 33, 121-124.	1.6	4
7	Phenotype associated with TAF2 biallelic mutations: A clinical description of four individuals and review of the literature. European Journal of Medical Genetics, 2021, 64, 104323.	1.3	5
8	Involvement of ADGRV1 Gene in Familial Forms of Genetic Generalized Epilepsy. Frontiers in Neurology, 2021, 12, 738272.	2.4	7
9	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
10	Congenital immobility and stiffness related to biallelic ATAD1 variants. Neurology: Genetics, 2020, 6, e520.	1.9	5
11	Mutations disrupting neuritogenesis genes confer risk for cerebral palsy. Nature Genetics, 2020, 52, 1046-1056.	21.4	96
12	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	12.8	99
13	Both rare and common genetic variants contribute to autism in the Faroe Islands. Npj Genomic Medicine, 2019, 4, 1.	3.8	72
14	Novel GABRA2 variants in epileptic encephalopathy and intellectual disability with seizures. Brain, 2019, 142, e15-e15.	7.6	12
15	Variants in TCF20 in neurodevelopmental disability: description of 27 new patients and review of literature. Genetics in Medicine, 2019, 21, 2036-2042.	2.4	23
16	Unstable TTTTA/TTTCA expansions in MARCH6 are associated with Familial Adult Myoclonic Epilepsy type 3. Nature Communications, 2019, 10, 4919.	12.8	111
17	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. Genetics in Medicine, 2019, 21, 837-849.	2.4	47
18	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. Nature Communications, 2018, 9, 4619.	12.8	70

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19	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. American Journal of Human Genetics, 2018, 103, 666-678.	6.2	87
20	Association of modifiers and other genetic factors explain Marfan syndrome clinical variability. European Journal of Human Genetics, 2018, 26, 1759-1772.	2.8	73
21	BCL11B mutations in patients affected by a neurodevelopmental disorder with reduced type 2 innate lymphoid cells. Brain, 2018, 141, 2299-2311.	7.6	81
22	A framework to identify contributing genes in patients with Phelan-McDermid syndrome. Npj Genomic Medicine, 2017, 2, 32.	3.8	58
23	Permanent draft genome sequence of the probiotic strain Propionibacterium freudenreichii CIRM-BIA 129 (ITG P20). Standards in Genomic Sciences, 2016, 11, 6.	1.5	11
24	A de novo microdeletion of SEMA5A in a boy with autism spectrum disorder and intellectual disability. European Journal of Human Genetics, 2016, 24, 838-843.	2.8	40
25	Mutations and genomic islands can explain the strain dependency of sugar utilization in 21 strains of Propionibacterium freudenreichii. BMC Genomics, 2015, 16, 296.	2.8	30
26	Genome Sequence of Lactobacillus delbrueckii subsp. lactis CNRZ327, a Dairy Bacterium with Anti-Inflammatory Properties. Genome Announcements, 2014, 2, .	0.8	4
27	Lactobacillus delbrueckii ssp. lactis and ssp. bulgaricus: a chronicle of evolution in action. BMC Genomics, 2014, 15, 407.	2.8	59