Ange-Line Bruel

List of Publications by Year
in descending order

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48 papers	1,520 citations	471509 17 h-index	345221 36 g-index
48 all docs	48 docs citations	48 times ranked	3719 citing authors

#	Article	IF	Citations
1	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
2	Clinical whole-exome sequencing for the diagnosis of rare disorders with congenital anomalies and/or intellectual disability: substantial interest of prospective annual reanalysis. Genetics in Medicine, 2018, 20, 645-654.	2.4	146
3	The oral-facial-digital syndrome gene C2CD3 encodes a positive regulator of centriole elongation. Nature Genetics, 2014, 46, 905-911.	21.4	121
4	The ciliopathy-associated CPLANE proteins direct basal body recruitment of intraflagellar transport machinery. Nature Genetics, 2016, 48, 648-656.	21.4	119
5	Fifteen years of research on oralâ€“facialâ€“digital syndromes: from 1 to 16 causal genes. Journal of Medical Genetics, 2017, 54, 371-380.	3.2	85
6	Expanding the Phenotype Associated with NAA10â€Related Nâ€Terminal Acetylation Deficiency. Human Mutation, 2016, 37, 755-764.	2.5	70
7	A Recurrent De Novo PACS2 Heterozygous Missense Variant Causes Neonatal-Onset Developmental Epileptic Encephalopathy, Facial Dysmorphism, and Cerebellar Dysgenesis. American Journal of Human Genetics, 2018, 102, 995-1007.	6.2	49
8	Dominant variants in the splicing factor PUF60 cause a recognizable syndrome with intellectual disability, heart defects and short stature. European Journal of Human Genetics, 2017, 25, 43-51.	2.8	44
9	Increased diagnostic and new genes identification outcome using research reanalysis of singleton exome sequencing. European Journal of Human Genetics, 2019, 27, 1519-1531.	2.8	43
10	Loss-of-Function Mutations in UNC45A Cause a Syndrome Associating Cholestasis, Diarrhea, Impaired Hearing, and Bone Fragility. American Journal of Human Genetics, 2018, 102, 364-374.	6.2	40
11	De novo SMARCA2 variants clustered outside the helicase domain cause a new recognizable syndrome with intellectual disability and blepharophimosis distinct from Nicolaidesâ€“Baraitser syndrome. Genetics in Medicine, 2020, 22, 1838-1850.	2.4	31
12	Okurâ€Chung neurodevelopmental syndrome: Eight additional cases with implications on phenotype and genotype expansion. Clinical Genetics, 2018, 93, 880-890.	2.0	30
13	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356.	6.2	30
14	De novo mutations in MSL3 cause an X-linked syndrome marked by impaired histone H4 lysine 16 acetylation. Nature Genetics, 2018, 50, 1442-1451.	21.4	28
15	TAOK1 is associated with neurodevelopmental disorder and essential for neuronal maturation and cortical development. Human Mutation, 2021, 42, 445-459.	2.5	26
16	Reducing diagnostic turnaround times of exome sequencing for families requiring timely diagnoses. European Journal of Medical Genetics, 2017, 60, 595-604.	1.3	22
17	VariantÂrecurrence in neurodevelopmental disorders: the use of publicly available genomic data identifies clinically relevant pathogenic missense variants. Genetics in Medicine, 2019, 21, 2504-2511.	2.4	21
18	<scp>Nextâ€generation</scp> sequencing approaches and challenges in the diagnosis of developmental anomalies and intellectual disability. Clinical Genetics, 2020, 98, 433-444.	2.0	20

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19	Truncating variants of the DLG4 gene are responsible for intellectual disability with marfanoid features. Clinical Genetics, 2018, 93, 1172-1178.	2.0	19
20	Genotype-first in a cohort of 95 fetuses with multiple congenital abnormalities: when exome sequencing reveals unexpected fetal phenotype-genotype correlations. Journal of Medical Genetics, 2021, 58, 400-413.	3.2	18
21	Kosaki overgrowth syndrome: A novel pathogenic variant in PDGFRB and expansion of the phenotype including cerebrovascular complications. Clinical Genetics, 2020, 98, 19-31.	2.0	17
22	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	2.4	16
23	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 601-617.	6.2	16
24	Autosomal recessive truncating <scp>MAB21L1</scp> mutation associated with a syndromic scrotal agenesis. Clinical Genetics, 2017, 91, 333-338.	2.0	15
25	Expanding the clinical spectrum of recessive truncating mutations ofKLHL7to a Bohring-Opitz-like phenotype. Journal of Medical Genetics, 2017, 54, 830-835.	3.2	15
26	2.5 yearsâ€™ experience of GeneMatcher data-sharing: a powerful tool for identifying new genes responsible for rare diseases. Genetics in Medicine, 2019, 21, 1657-1661.	2.4	14
27	MYT1L-associated neurodevelopmental disorder: description of 40 new cases and literature review of clinical and molecular aspects. Human Genetics, 2022, 141, 65-80.	3.8	14
28	Expanding the phenotype of <scp>ASXL3</scp>â€related syndrome: A comprehensive description of 45 unpublished individuals with inherited and de novo pathogenic variants in <scp>ASXL3</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 3446-3458.	1.2	12
29	Accelerated genome sequencing with controlled costs for infants in intensive care units: a feasibility study in a French hospital network. European Journal of Human Genetics, 2022, 30, 567-576.	2.8	12
30	Deciphering exome sequencing data: Bringing mitochondrial DNA variants to light. Human Mutation, 2019, 40, 2430-2443.	2.5	11
31	Neuropsychological study in 19 French patients with <scp>Whiteâ€Sutton</scp> syndrome and <scp>POGZ</scp> mutations. Clinical Genetics, 2021, 99, 407-417.	2.0	10
32	Second-tier trio exome sequencing after negative solo clinical exome sequencing: an efficient strategy to increase diagnostic yield and decipher molecular bases in undiagnosed developmental disorders. Human Genetics, 2020, 139, 1381-1390.	3.8	8
33	INTUâ€related oralâ€facialâ€digital syndrome type VI: A confirmatory report. Clinical Genetics, 2018, 93, 1205-1209.	2.0	7
34	Excess of de novo variants in genes involved in chromatin remodelling in patients with marfanoid habitus and intellectual disability. Journal of Medical Genetics, 2020, 57, 466-474.	3.2	7
35	The oculoauriculofrontonasal syndrome: Further clinical characterization and additional evidence suggesting a nontraditional mode of inheritance. American Journal of Medical Genetics, Part A, 2018, 176, 2740-2750.	1.2	6
36	High efficiency and clinical relevance of exome sequencing in the daily practice of neurogenetics. Journal of Medical Genetics, 2022, 59, 445-452.	3.2	6

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37	Copy number variants calling from WES data through eXome hidden Markov model (XHMM) identifies additional 2.5% pathogenic genomic imbalances smaller than 30Âkb undetected by arrayâ€CGH. Annals of Human Genetics, 2022, 86, 171-180.	0.8	6
38	Expanding the phenotype of <scp>HNRNPU</scp>â€related neurodevelopmental disorder with emphasis on seizure phenotype and review of literature. American Journal of Medical Genetics, Part A, 2022, 188, 1497-1514.	1.2	6
39	Interest of exome sequencing trioâ€like strategy based on pooled parental DNA for diagnosis and translational research in rare diseases. Molecular Genetics & Genomic Medicine, 2021, 9, e1836.	1.2	5
40	<scp>Skrabanâ€Deardorff</scp> syndrome: Six new cases of <scp>WDR26</scp>â€related disease and expansion of the clinical phenotype. Clinical Genetics, 2021, 99, 732-739.	2.0	4
41	ITSN1: a novel candidate gene involved in autosomal dominant neurodevelopmental disorder spectrum. European Journal of Human Genetics, 2022, 30, 111-116.	2.8	4
42	Unexpected diagnosis of a SHH nonsense variant causing a variable phenotype ranging from familial coloboma and Intellectual disability to isolated microcephaly. Clinical Genetics, 2018, 94, 182-184.	2.0	2
43	The diagnostic rate of inherited metabolic disorders by exome sequencing in a cohort of 547 individuals with developmental disorders. Molecular Genetics and Metabolism Reports, 2021, 29, 100812.	1.1	2
44	Atypical phenotype of a patient with Bardetâ€“Biedl syndrome type 4. Molecular Genetics & Genomic Medicine, 2022, 10, e1869.	1.2	2
45	Hydrothorax in fetal cases of Opitz G/<scp>BBB</scp> diagnosis: Extending the phenotype?. Clinical Genetics, 2020, 98, 620-621.	2.0	1
46	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
47	Toward clinical and molecular dissection of frontonasal dysplasia with facial skin polyps: From Pai syndrome to differential diagnosis through a series of 27 patients. American Journal of Medical Genetics, Part A, 2022, 188, 2036-2047.	1.2	1
48	Same performance of exome sequencing before and after fetal autopsy for congenital abnormalities: toward a paradigm shift in prenatal diagnosis?. European Journal of Human Genetics, 2022, , .	2.8	1