Laurence Olivier-Faivre

List of Publications by Year
in descending order

70 papers	3,998 citations	279798 23 h-index	123424 61 g-index
72 all docs	72 docs citations	72 times ranked	7009 citing authors

#	Article	IF	Citations
1	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
2	Phenotypic spectrum and genomics of undiagnosed arthrogryposis multiplex congenita. Journal of Medical Genetics, 2022, 59, 559-567.	3.2	25
3	Clinical and molecular data in cases of prenatal localized overgrowth disorder: major implication of genetic variants in <scp>PI3Kâ€AKTâ€mTOR</scp> signaling pathway. Ultrasound in Obstetrics and Gynecology, 2022, 59, 532-542.	1.7	6
4	Refining the clinical phenotype associated with missense variants in exons 38 and 39 of KMT2D. American Journal of Medical Genetics, Part A, 2022, , .	1.2	0
5	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
6	Consolidation of the clinical and genetic definition of aSOX4-related neurodevelopmental syndrome. Journal of Medical Genetics, 2022, 59, 1058-1068.	3.2	10
7	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
8	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
9	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
10	Clinical and neuroimaging findings in 33 patients with <scp>MCAP</scp> syndrome: A survey to evaluate relevant endpoints for future clinical trials. Clinical Genetics, 2021, 99, 650-661.	2.0	12
11	Clinical spectrum of MTOR-related hypomelanosis of Ito with neurodevelopmental abnormalities. Genetics in Medicine, 2021, 23, 1484-1491.	2.4	14
12	Broadening the phenotypic spectrum and physiological insights related toEIF2S3variants. Human Mutation, 2021, 42, 827-834.	2.5	5
13	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
14	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
15	Growth charts in Kabuki syndrome 1. American Journal of Medical Genetics, Part A, 2020, 182, 446-453.	1.2	7
16	Incidence of cardiovascular events and risk markers in a prospective study of children diagnosed with Marfan syndrome. Archives of Cardiovascular Diseases, 2020, 113, 40-49.	1.6	12
17	Associations between cognitive performance and the rehabilitation, medical care and social support provided to French children with Prader-Willi syndrome. European Journal of Medical Genetics, 2020, 63, 104064.	1.3	5
18	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

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19	De novo mutations in the X-linked TFE3 gene cause intellectual disability with pigmentary mosaicism and storage disorder-like features. Journal of Medical Genetics, 2020, 57, 808-819.	3.2	11
20	Mandibularâ€pelvicâ€patellar syndrome is a novel PITX1 â€related disorder due to alteration of PITX1 transactivation ability. Human Mutation, 2020, 41, 1499-1506.	2.5	2
21	Neutralization of HSF1 in cells from PIK3CA-related overgrowth spectrum patients blocks abnormal proliferation. Biochemical and Biophysical Research Communications, 2020, 530, 520-526.	2.1	5
22	Cardiomyopathy due to PRDM16 mutation: First description of a fetal presentation, with possible modifier genes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 129-135.	1.6	15
23	Genome sequencing in cytogenetics: Comparison of shortâ€read and linkedâ€read approaches for germline structural variant detection and characterization. Molecular Genetics & Genomic Medicine, 2020, 8, e1114.	1.2	10
24	Novel KIAA1033 / WASHC4 mutations in three patients with syndromic intellectual disability and a review of the literature. American Journal of Medical Genetics, Part A, 2020, 182, 792-797.	1.2	12
25	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
26	<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
27	Compassionate use of everolimus for refractory epilepsy in a patient with MTOR mosaic mutation. European Journal of Medical Genetics, 2020, 63, 104036.	1.3	8
28	Severe gynaecological involvement in Proteus Syndrome. European Journal of Medical Genetics, 2019, 62, 270-272.	1.3	3
29	HIST1H1E heterozygous proteinâ€truncating variants cause a recognizable syndrome with intellectual disability and distinctive facial gestalt: A study to clarify the HIST1H1E syndrome phenotype in 30 individuals. American Journal of Medical Genetics, Part A, 2019, 179, 2049-2055.	1.2	16
30	Deciphering exome sequencing data: Bringing mitochondrial DNA variants to light. Human Mutation, 2019, 40, 2430-2443.	2.5	11
31	De Novo Variants Disturbing the Transactivation Capacity of POU3F3 Cause a Characteristic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 105, 403-412.	6.2	35
32	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
33	Hearing impairment as an early sign of alphaâ€mannosidosis in children with a mild phenotype: Report of seven new cases. American Journal of Medical Genetics, Part A, 2019, 179, 1756-1763.	1.2	13
34	Whole Exome Sequencing Reveals a Large Genetic Heterogeneity and Revisits the Causes of Hypertrophic Cardiomyopathy. Circulation Genomic and Precision Medicine, 2019, 12, e002500.	3.6	9
35	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
36	Whole genome paired-end sequencing elucidates functional and phenotypic consequences of balanced chromosomal rearrangement in patients with developmental disorders. Journal of Medical Genetics, 2019, 56, 526-535.	3.2	46

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37	Mutations in the Neuronal Vesicular SNARE VAMP2 Affect Synaptic Membrane Fusion and Impair Human Neurodevelopment. American Journal of Human Genetics, 2019, 104, 721-730.	6.2	88
38	Malignant transformation of presacral mass in Currarino syndrome. Pediatric Blood and Cancer, 2019, 66, e27659.	1.5	5
39	Secondary findings from whole-exome/genome sequencing evaluating stakeholder perspectives. A review of the literature. European Journal of Medical Genetics, 2019, 62, 103529.	1.3	33
40	Lysosomal Signaling Licenses Embryonic Stem Cell Differentiation via Inactivation of Tfe3. Cell Stem Cell, 2019, 24, 257-270.e8.	11.1	97
41	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
42	Targeted panel sequencing in adult patients with left ventricular nonâ€compaction reveals a large genetic heterogeneity. Clinical Genetics, 2019, 95, 356-367.	2.0	56
43	Truncating variants of the DLG4 gene are responsible for intellectual disability with marfanoid features. Clinical Genetics, 2018, 93, 1172-1178.	2.0	19
44	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
45	Delineating the psychiatric and behavioral phenotype of recurrent 2q13 deletions and duplications. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 397-405.	1.7	16
46	Recessive loss of function PIGN alleles, including an intragenic deletion with founder effect in La RÃ©union Island, in patients with Fryns syndrome. European Journal of Human Genetics, 2018, 26, 340-349.	2.8	27
47	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
48	Time-based prospective memory in children and adolescents with 22q11.2 deletion syndrome. Clinical Neuropsychologist, 2018, 32, 981-992.	2.3	3
49	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
50	Further delineation of the clinical spectrum of de novo TRIM8 truncating mutations. American Journal of Medical Genetics, Part A, 2018, 176, 2470-2478.	1.2	19
51	Extending the ALDH18A1 clinical spectrum to severe autosomal recessive fetal cutis laxa with corpus callosum agenesis. American Journal of Medical Genetics, Part A, 2018, 176, 2509-2512.	1.2	9
52	TBL1XR1 mutations in Pierpont syndrome are not restricted to the recurrent p.Tyr446Cys mutation. American Journal of Medical Genetics, Part A, 2018, 176, 2813-2818.	1.2	10
53	NFIB Haploinsufficiency Is Associated with Intellectual Disability and Macrocephaly. American Journal of Human Genetics, 2018, 103, 752-768.	6.2	40
54	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

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55	Molecular diagnosis of PIK3CA-related overgrowth spectrum (PROS) in 162 patients and recommendations for genetic testing. Genetics in Medicine, 2017, 19, 989-997.	2.4	90
56	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
57	Phenotype and genotype analysis of a French cohort of 119 patients with CHARGE syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 417-430.	1.6	65
58	Clinical delineation of a subtype of frontonasal dysplasia with creased nasal ridge and upper limb anomalies: Report of six unrelated patients. American Journal of Medical Genetics, Part A, 2017, 173, 3136-3142.	1.2	9
59	Diagnostic odyssey in severe neurodevelopmental disorders: toward clinical wholeâ€exome sequencing as a firstâ€line diagnostic test. Clinical Genetics, 2016, 89, 700-707.	2.0	205
60	6q16.3q23.3 duplication associated with Prader-Willi-like syndrome. Molecular Cytogenetics, 2015, 8, 42.	0.9	11
61	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. American Journal of Human Genetics, 2015, 97, 922-932.	6.2	101
62	WWOX-related encephalopathies: delineation of the phenotypical spectrum and emerging genotype-phenotype correlation. Journal of Medical Genetics, 2015, 52, 61-70.	3.2	74
63	Juvenile Xanthogranuloma and Nevus Anemicus in the Diagnosis of Neurofibromatosis Type 1. JAMA Dermatology, 2014, 150, 42.	4.1	63
64	Mutations in SLC13A5 Cause Autosomal-Recessive Epileptic Encephalopathy with Seizure Onset in the First Days of Life. American Journal of Human Genetics, 2014, 95, 113-120.	6.2	112
65	New candidate loci identified by arrayâ€CGH in a cohort of 100 children presenting with syndromic obesity. American Journal of Medical Genetics, Part A, 2014, 164, 1965-1975.	1.2	49
66	Systematic molecular and cytogenetic screening of 100 patients with marfanoid syndromes and intellectual disability. Clinical Genetics, 2013, 84, 507-521.	2.0	23
67	In-Frame Mutations in Exon 1 of SKI Cause Dominant Shprintzen-Goldberg Syndrome. American Journal of Human Genetics, 2012, 91, 950-957.	6.2	95
68	Truncating mutations in the last exon of NOTCH2 cause a rare skeletal disorder with osteoporosis. Nature Genetics, 2011, 43, 306-308.	21.4	181
69	The revised Ghent nosology for the Marfan syndrome. Journal of Medical Genetics, 2010, 47, 476-485.	3.2	1,677
70	Segmental overgrowth, lipomatosis, arteriovenous malformation and epidermal nevus (SOLAMEN) syndrome is related to mosaic PTEN nullizygosity. European Journal of Human Genetics, 2007, 15, 767-773.	2.8	129