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	The revised Ghent nosology for the Marfan syndrome. Journal of Medical Genetics, 2010, 47, 476-485.	3.2	1,677
2	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
3	Truncating mutations in the last exon of NOTCH2 cause a rare skeletal disorder with osteoporosis. Nature Genetics, 2011, 43, 306-308.	21.4	181
4	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
5	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
6	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
7	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. American Journal of Human Genetics, 2015, 97, 922-932.	6.2	101
8	Lysosomal Signaling Licenses Embryonic Stem Cell Differentiation via Inactivation of Tfe3. Cell Stem Cell, 2019, 24, 257-270.e8.	11.1	97
9	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
10	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
11	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
12	WWOX-related encephalopathies: delineation of the phenotypical spectrum and emerging genotype-phenotype correlation. Journal of Medical Genetics, 2015, 52, 61-70.	3.2	74
13	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
14	Juvenile Xanthogranuloma and Nevus Anemicus in the Diagnosis of Neurofibromatosis Type 1. JAMA Dermatology, 2014, 150, 42.	4.1	63
15	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
16	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
17	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
18	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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19	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
20	NFIB Haploinsufficiency Is Associated with Intellectual Disability and Macrocephaly. American Journal of Human Genetics, 2018, 103, 752-768.	6.2	40
21	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
22	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
23	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
24	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
25	Phenotypic spectrum and genomics of undiagnosed arthrogryposis multiplex congenita. Journal of Medical Genetics, 2022, 59, 559-567.	3.2	25
26	Systematic molecular and cytogenetic screening of 100 patients with marfanoid syndromes and intellectual disability. Clinical Genetics, 2013, 84, 507-521.	2.0	23
27	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
28	<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
29	Truncating variants of the DLG4 gene are responsible for intellectual disability with marfanoid features. Clinical Genetics, 2018, 93, 1172-1178.	2.0	19
30	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
31	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
32	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
33	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
34	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
35	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
36	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

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37	Clinical spectrum of MTOR-related hypomelanosis of Ito with neurodevelopmental abnormalities. Genetics in Medicine, 2021, 23, 1484-1491.	2.4	14
38	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
39	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
40	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
41	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
42	6q16.3q23.3 duplication associated with Prader-Willi-like syndrome. Molecular Cytogenetics, 2015, 8, 42.	0.9	11
43	Deciphering exome sequencing data: Bringing mitochondrial DNA variants to light. Human Mutation, 2019, 40, 2430-2443.	2.5	11
44	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
45	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
46	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
47	Consolidation of the clinical and genetic definition of aSOX4-related neurodevelopmental syndrome. Journal of Medical Genetics, 2022, 59, 1058-1068.	3.2	10
48	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
49	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
50	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
51	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
52	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
53	Growth charts in Kabuki syndrome 1. American Journal of Medical Genetics, Part A, 2020, 182, 446-453.	1.2	7
54	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

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55	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
56	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
57	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
58	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
59	Malignant transformation of presacral mass in Currarino syndrome. Pediatric Blood and Cancer, 2019, 66, e27659.	1.5	5
60	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
61	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
62	Broadening the phenotypic spectrum and physiological insights related toEIF2S3variants. Human Mutation, 2021, 42, 827-834.	2.5	5
63	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
64	Time-based prospective memory in children and adolescents with 22q11.2 deletion syndrome. Clinical Neuropsychologist, 2018, 32, 981-992.	2.3	3
65	Severe gynaecological involvement in Proteus Syndrome. European Journal of Medical Genetics, 2019, 62, 270-272.	1.3	3
66	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
67	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
68	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
69	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
70	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