

Laurence Olivier-Faivre

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/2284431/publications.pdf>

Version: 2024-02-01

70
papers

3,998
citations

279487

23
h-index

123241

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. <i>Journal of Medical Genetics</i> , 2010, 47, 476-485.	1.5	1,677
2	Diagnostic odyssey in severe neurodevelopmental disorders: toward clinical whole-exome sequencing as a first-line diagnostic test. <i>Clinical Genetics</i> , 2016, 89, 700-707.	1.0	205
3	Truncating mutations in the last exon of NOTCH2 cause a rare skeletal disorder with osteoporosis. <i>Nature Genetics</i> , 2011, 43, 306-308.	9.4	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. <i>Genetics in Medicine</i> , 2018, 20, 645-654.	1.1	146
5	Segmental overgrowth, lipomatosis, arteriovenous malformation and epidermal nevus (SOLAMEN) syndrome is related to mosaic PTEN nullizygoty. <i>European Journal of Human Genetics</i> , 2007, 15, 767-773.	1.4	129
6	Mutations in SLC13A5 Cause Autosomal-Recessive Epileptic Encephalopathy with Seizure Onset in the First Days of Life. <i>American Journal of Human Genetics</i> , 2014, 95, 113-120.	2.6	112
7	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. <i>American Journal of Human Genetics</i> , 2015, 97, 922-932.	2.6	101
8	Lysosomal Signaling Licenses Embryonic Stem Cell Differentiation via Inactivation of Tfe3. <i>Cell Stem Cell</i> , 2019, 24, 257-270.e8.	5.2	97
9	In-Frame Mutations in Exon 1 of SKI Cause Dominant Shprintzen-Goldberg Syndrome. <i>American Journal of Human Genetics</i> , 2012, 91, 950-957.	2.6	95
10	Molecular diagnosis of PIK3CA-related overgrowth spectrum (PROS) in 162 patients and recommendations for genetic testing. <i>Genetics in Medicine</i> , 2017, 19, 989-997.	1.1	90
11	Mutations in the Neuronal Vesicular SNARE VAMP2 Affect Synaptic Membrane Fusion and Impair Human Neurodevelopment. <i>American Journal of Human Genetics</i> , 2019, 104, 721-730.	2.6	88
12	WWOX-related encephalopathies: delineation of the phenotypical spectrum and emerging genotype-phenotype correlation. <i>Journal of Medical Genetics</i> , 2015, 52, 61-70.	1.5	74
13	Phenotype and genotype analysis of a French cohort of 119 patients with CHARGE syndrome. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2017, 175, 417-430.	0.7	65
14	Juvenile Xanthogranuloma and Nevus Anemicus in the Diagnosis of Neurofibromatosis Type 1. <i>JAMA Dermatology</i> , 2014, 150, 42.	2.0	63
15	Targeted panel sequencing in adult patients with left ventricular non-compaction reveals a large genetic heterogeneity. <i>Clinical Genetics</i> , 2019, 95, 356-367.	1.0	56
16	New candidate loci identified by array-CGH in a cohort of 100 children presenting with syndromic obesity. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1965-1975.	0.7	49
17	A Recurrent De Novo PACS2 Heterozygous Missense Variant Causes Neonatal-Onset Developmental Epileptic Encephalopathy, Facial Dysmorphism, and Cerebellar Dysgenesis. <i>American Journal of Human Genetics</i> , 2018, 102, 995-1007.	2.6	49
18	Whole genome paired-end sequencing elucidates functional and phenotypic consequences of balanced chromosomal rearrangement in patients with developmental disorders. <i>Journal of Medical Genetics</i> , 2019, 56, 526-535.	1.5	46

#	ARTICLE	IF	CITATIONS
19	Increased diagnostic and new genes identification outcome using research reanalysis of singleton exome sequencing. <i>European Journal of Human Genetics</i> , 2019, 27, 1519-1531.	1.4	43
20	NFIB Haploinsufficiency Is Associated with Intellectual Disability and Macrocephaly. <i>American Journal of Human Genetics</i> , 2018, 103, 752-768.	2.6	40
21	De Novo Variants Disturbing the Transactivation Capacity of POU3F3 Cause a Characteristic Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2019, 105, 403-412.	2.6	35
22	Secondary findings from whole-exome/genome sequencing evaluating stakeholder perspectives. A review of the literature. <i>European Journal of Medical Genetics</i> , 2019, 62, 103529.	0.7	33
23	De novo mutations in MSL3 cause an X-linked syndrome marked by impaired histone H4 lysine 16 acetylation. <i>Nature Genetics</i> , 2018, 50, 1442-1451.	9.4	28
24	Recessive loss of function PIGN alleles, including an intragenic deletion with founder effect in La Réunion Island, in patients with Fryns syndrome. <i>European Journal of Human Genetics</i> , 2018, 26, 340-349.	1.4	27
25	Phenotypic spectrum and genomics of undiagnosed arthrogryposis multiplex congenita. <i>Journal of Medical Genetics</i> , 2022, 59, 559-567.	1.5	25
26	Systematic molecular and cytogenetic screening of 100 patients with marfanoid syndromes and intellectual disability. <i>Clinical Genetics</i> , 2013, 84, 507-521.	1.0	23
27	Variant recurrence in neurodevelopmental disorders: the use of publicly available genomic data identifies clinically relevant pathogenic missense variants. <i>Genetics in Medicine</i> , 2019, 21, 2504-2511.	1.1	21
28	Next-generation sequencing approaches and challenges in the diagnosis of developmental anomalies and intellectual disability. <i>Clinical Genetics</i> , 2020, 98, 433-444.	1.0	20
29	Truncating variants of the <i>DLG4</i> gene are responsible for intellectual disability with marfanoid features. <i>Clinical Genetics</i> , 2018, 93, 1172-1178.	1.0	19
30	Further delineation of the clinical spectrum of de novo <i>TRIM8</i> truncating mutations. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2470-2478.	0.7	19
31	Genotype-first in a cohort of 95 fetuses with multiple congenital abnormalities: when exome sequencing reveals unexpected fetal phenotype-genotype correlations. <i>Journal of Medical Genetics</i> , 2021, 58, 400-413.	1.5	18
32	Delineating the psychiatric and behavioral phenotype of recurrent 2q13 deletions and duplications. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2018, 177, 397-405.	1.1	16
33	<i>HIST1H1E</i> heterozygous protein-truncating variants cause a recognizable syndrome with intellectual disability and distinctive facial gestalt: A study to clarify the <i>HIST1H1E</i> syndrome phenotype in 30 individuals. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2049-2055.	0.7	16
34	Expanding the clinical spectrum of recessive truncating mutations of <i>KLHL7</i> to a Bohring-Opitz-like phenotype. <i>Journal of Medical Genetics</i> , 2017, 54, 830-835.	1.5	15
35	Cardiomyopathy due to <i>PRDM16</i> mutation: First description of a fetal presentation, with possible modifier genes. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2020, 184, 129-135.	0.7	15
36	2.5 years' experience of GeneMatcher data-sharing: a powerful tool for identifying new genes responsible for rare diseases. <i>Genetics in Medicine</i> , 2019, 21, 1657-1661.	1.1	14

#	ARTICLE	IF	CITATIONS
37	Clinical spectrum of MTOR-related hypomelanosis of Ito with neurodevelopmental abnormalities. <i>Genetics in Medicine</i> , 2021, 23, 1484-1491.	1.1	14
38	Hearing impairment as an early sign of alpha β -mannosidosis in children with a mild phenotype: Report of seven new cases. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1756-1763.	0.7	13
39	Incidence of cardiovascular events and risk markers in a prospective study of children diagnosed with Marfan syndrome. <i>Archives of Cardiovascular Diseases</i> , 2020, 113, 40-49.	0.7	12
40	Novel KIAA1033 / WASHC4 mutations in three patients with syndromic intellectual disability and a review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 792-797.	0.7	12
41	Clinical and neuroimaging findings in 33 patients with <sc>MCAP</sc> syndrome: A survey to evaluate relevant endpoints for future clinical trials. <i>Clinical Genetics</i> , 2021, 99, 650-661.	1.0	12
42	6q16.3q23.3 duplication associated with Prader-Willi-like syndrome. <i>Molecular Cytogenetics</i> , 2015, 8, 42.	0.4	11
43	Deciphering exome sequencing data: Bringing mitochondrial DNA variants to light. <i>Human Mutation</i> , 2019, 40, 2430-2443.	1.1	11
44	<i>De novo</i> mutations in the X-linked <i>TFE3</i> gene cause intellectual disability with pigmentary mosaicism and storage disorder-like features. <i>Journal of Medical Genetics</i> , 2020, 57, 808-819.	1.5	11
45	<i>TBL1XR1</i> mutations in Pierpont syndrome are not restricted to the recurrent p.Tyr446Cys mutation. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2813-2818.	0.7	10
46	Genome sequencing in cytogenetics: Comparison of short Δ read and linked Δ read approaches for germline structural variant detection and characterization. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1114.	0.6	10
47	Consolidation of the clinical and genetic definition of a <i>SOX4</i>-related neurodevelopmental syndrome. <i>Journal of Medical Genetics</i> , 2022, 59, 1058-1068.	1.5	10
48	Clinical delineation of a subtype of frontonasal dysplasia with creased nasal ridge and upper limb anomalies: Report of six unrelated patients. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 3136-3142.	0.7	9
49	Extending the <i>ALDH18A1</i> clinical spectrum to severe autosomal recessive fetal cutis laxa with corpus callosum agenesis. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2509-2512.	0.7	9
50	Whole Exome Sequencing Reveals a Large Genetic Heterogeneity and Revisits the Causes of Hypertrophic Cardiomyopathy. <i>Circulation Genomic and Precision Medicine</i> , 2019, 12, e002500.	1.6	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. <i>Human Genetics</i> , 2020, 139, 1381-1390.	1.8	8
52	Compassionate use of everolimus for refractory epilepsy in a patient with MTOR mosaic mutation. <i>European Journal of Medical Genetics</i> , 2020, 63, 104036.	0.7	8
53	Growth charts in Kabuki syndrome 1. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 446-453.	0.7	7
54	Excess of de novo variants in genes involved in chromatin remodelling in patients with marfanoid habitus and intellectual disability. <i>Journal of Medical Genetics</i> , 2020, 57, 466-474.	1.5	7

#	ARTICLE	IF	CITATIONS
55	The oculoauriculofrontonasal syndrome: Further clinical characterization and additional evidence suggesting a nontraditional mode of inheritance. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2740-2750.	0.7	6
56	High efficiency and clinical relevance of exome sequencing in the daily practice of neurogenetics. <i>Journal of Medical Genetics</i> , 2022, 59, 445-452.	1.5	6
57	Clinical and molecular data in cases of prenatal localized overgrowth disorder: major implication of genetic variants in $\text{PI3K}\text{-AKT}\text{-mTOR}$ signaling pathway. <i>Ultrasound in Obstetrics and Gynecology</i> , 2022, 59, 532-542.	0.9	6
58	Copy number variants calling from WES data through eXome hidden Markov model (XHMM) identifies additional 2.5% pathogenic genomic imbalances smaller than 30kb undetected by array-CGH. <i>Annals of Human Genetics</i> , 2022, 86, 171-180.	0.3	6
59	Malignant transformation of presacral mass in Currarino syndrome. <i>Pediatric Blood and Cancer</i> , 2019, 66, e27659.	0.8	5
60	Associations between cognitive performance and the rehabilitation, medical care and social support provided to French children with Prader-Willi syndrome. <i>European Journal of Medical Genetics</i> , 2020, 63, 104064.	0.7	5
61	Neutralization of HSF1 in cells from PIK3CA-related overgrowth spectrum patients blocks abnormal proliferation. <i>Biochemical and Biophysical Research Communications</i> , 2020, 530, 520-526.	1.0	5
62	Broadening the phenotypic spectrum and physiological insights related to <i>EIF2S3</i> variants. <i>Human Mutation</i> , 2021, 42, 827-834.	1.1	5
63	Interest of exome sequencing trio-like strategy based on pooled parental DNA for diagnosis and translational research in rare diseases. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1836.	0.6	5
64	Time-based prospective memory in children and adolescents with 22q11.2 deletion syndrome. <i>Clinical Neuropsychologist</i> , 2018, 32, 981-992.	1.5	3
65	Severe gynaecological involvement in Proteus Syndrome. <i>European Journal of Medical Genetics</i> , 2019, 62, 270-272.	0.7	3
66	Mandibular-pelvic-patellar syndrome is a novel PITX1-related disorder due to alteration of PITX1 transactivation ability. <i>Human Mutation</i> , 2020, 41, 1499-1506.	1.1	2
67	The diagnostic rate of inherited metabolic disorders by exome sequencing in a cohort of 547 individuals with developmental disorders. <i>Molecular Genetics and Metabolism Reports</i> , 2021, 29, 100812.	0.4	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. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2036-2047.	0.7	1
69	Same performance of exome sequencing before and after fetal autopsy for congenital abnormalities: toward a paradigm shift in prenatal diagnosis?. <i>European Journal of Human Genetics</i> , 2022, , .	1.4	1
70	Refining the clinical phenotype associated with missense variants in exons 38 and 39 of KMT2D. <i>American Journal of Medical Genetics, Part A</i> , 2022, , .	0.7	0