

Julien FaurÃ©

List of Publications by Year in descending order

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Version: 2024-02-01

69
papers

3,532
citations

201674

27
h-index

138484

58
g-index

72
all docs

72
docs citations

72
times ranked

5556
citing authors

#	ARTICLE	IF	CITATIONS
1	Clinical phenotype and loss of the slow skeletal muscle troponin T in three new patients with recessive TNNT1 nemaline myopathy. <i>Journal of Medical Genetics</i> , 2021, 58, 602-608.	3.2	11
2	Role of oculocerebrorenal syndrome of Lowe (OCRL) protein in megakaryocyte maturation, platelet production and functions: a study in patients with Lowe syndrome. <i>British Journal of Haematology</i> , 2021, 192, 909-921.	2.5	6
3	Second Report of Chronic Granulomatous Disease in Jordan: Clinical and Genetic Description of 31 Patients From 21 Different Families, Including Families From Lybia and Iraq. <i>Frontiers in Immunology</i> , 2021, 12, 639226.	4.8	12
4	A recurrent RYR1 mutation associated with early-onset hypotonia and benign disease course. <i>Acta Neuropathologica Communications</i> , 2021, 9, 155.	5.2	1
5	Characterization of Loss-Of-Function KCNJ2 Mutations in Atypical Andersen Tawil Syndrome. <i>Frontiers in Genetics</i> , 2021, 12, 773177.	2.3	1
6	Diagnostic workup in children with arthrogryposis: description of practices from a single reference centre, comparison with literature and suggestion of recommendations. <i>Journal of Medical Genetics</i> , 2021, , jmedgenet-2021-107823.	3.2	1
7	Interplay between Triadin and Calsequestrin in the Pathogenesis of CPVT in the Mouse. <i>Molecular Therapy</i> , 2020, 28, 171-179.	8.2	17
8	Association of fingerprint bodies with rods in a case with mutations in the LMOD3 gene. <i>Neuromuscular Disorders</i> , 2020, 30, 207-212.	0.6	6
9	Pathogenic Variants in the Myosin Chaperone UNC-45B Cause Progressive Myopathy with Eccentric Cores. <i>American Journal of Human Genetics</i> , 2020, 107, 1078-1095.	6.2	24
10	Response to Hall et Al.. <i>American Journal of Human Genetics</i> , 2020, 107, 1188-1189.	6.2	0
11	In vivo RyR1 reduction in muscle triggers a core-like myopathy. <i>Acta Neuropathologica Communications</i> , 2020, 8, 192.	5.2	9
12	Mutations in MYLPF Cause a Novel Segmental Amyoplasia that Manifests as Distal Arthrogryposis. <i>American Journal of Human Genetics</i> , 2020, 107, 293-310.	6.2	21
13	Variations in the TRPV1 gene are associated to exertional heat stroke. <i>Journal of Science and Medicine in Sport</i> , 2020, 23, 1021-1027.	1.3	7
14	Dynamics of triadin, a muscle-specific triad protein, within sarcoplasmic reticulum subdomains. <i>Molecular Biology of the Cell</i> , 2020, 31, 261-272.	2.1	1
15	New recessive mutations in <i>SYT2</i> causing severe presynaptic congenital myasthenic syndromes. <i>Neurology: Genetics</i> , 2020, 6, e534.	1.9	6
16	TRPV1 variants impair intracellular Ca ²⁺ signaling and may confer susceptibility to malignant hyperthermia. <i>Genetics in Medicine</i> , 2019, 21, 441-450.	2.4	17
17	Familial deep cavitating state with a glutathione metabolism defect. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 2573-2578.	3.7	1
18	A National French consensus on gene lists for the diagnosis of myopathies using next-generation sequencing. <i>European Journal of Human Genetics</i> , 2019, 27, 349-352.	2.8	27

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19	A novel nonsense PIEZO2 mutation in a family with scoliosis and proprioceptive defect. <i>Neuromuscular Disorders</i> , 2019, 29, 75-79.	0.6	13
20	“Dusty core disease” (DuCD): expanding morphological spectrum of RYR1 recessive myopathies. <i>Acta Neuropathologica Communications</i> , 2019, 7, 3.	5.2	31
21	International Triadin Knockout Syndrome Registry. <i>Circulation Genomic and Precision Medicine</i> , 2019, 12, e002419.	3.6	32
22	Mutations in CFAP43 and CFAP44 cause male infertility and flagellum defects in <i>Trypanosoma</i> and human. <i>Nature Communications</i> , 2018, 9, 686.	12.8	173
23	“Lowe syndrome: A particularly severe phenotype without clinical kidney involvement” <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 460-464.	1.2	4
24	ALK fusion variants detection by targeted RNA-next generation sequencing and clinical responses to crizotinib in ALK-positive non-small cell lung cancer. <i>Lung Cancer</i> , 2018, 116, 15-24.	2.0	44
25	Deletion of the microtubule-associated protein 6 (MAP6) results in skeletal muscle dysfunction. <i>Skeletal Muscle</i> , 2018, 8, 30.	4.2	21
26	Interplay between Triadin and Calsequestrin in the Pathogenesis of CPVT. <i>Biophysical Journal</i> , 2018, 114, 618a.	0.5	0
27	<i>STAC3</i> variants cause a congenital myopathy with distinctive dysmorphic features and malignant hyperthermia susceptibility. <i>Human Mutation</i> , 2018, 39, 1980-1994.	2.5	42
28	Common and variable clinical, histological, and imaging findings of recessive RYR1-related centronuclear myopathy patients. <i>Neuromuscular Disorders</i> , 2017, 27, 975-985.	0.6	34
29	<i>SPINK2</i> deficiency causes infertility by inducing sperm defects in heterozygotes and azoospermia in homozygotes. <i>EMBO Molecular Medicine</i> , 2017, 9, 1132-1149.	6.9	95
30	Dynamics of Triad Organization. <i>Biophysical Journal</i> , 2017, 112, 99a.	0.5	0
31	Functional Characterization and Rescue of a Deep Intronic Mutation in <i>OCRL</i> Gene Responsible for Lowe Syndrome. <i>Human Mutation</i> , 2017, 38, 152-159.	2.5	13
32	Excitation-Contraction Coupling Alterations in Myopathies. <i>Journal of Neuromuscular Diseases</i> , 2016, 3, 443-453.	2.6	22
33	The Microtubule-Associated Protein CLIMP-63 is a New Member of the Calcium Release Complex. <i>Biophysical Journal</i> , 2016, 110, 181a-182a.	0.5	0
34	Phospholipase A2 Receptor-Related Membranous Nephropathy and Mannan-Binding Lectin Deficiency. <i>Journal of the American Society of Nephrology: JASN</i> , 2016, 27, 3539-3544.	6.1	86
35	Triadin and CLIMP-63 form a link between triads and microtubules in muscle cells. <i>Journal of Cell Science</i> , 2016, 129, 3744-3755.	2.0	37
36	Mild clinical presentation in KLHL40-related nemaline myopathy (NEM 8). <i>Neuromuscular Disorders</i> , 2016, 26, 712-716.	0.6	16

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37	The transcription coactivator ASC-1 is a regulator of skeletal myogenesis, and its deficiency causes a novel form of congenital muscle disease. <i>Human Molecular Genetics</i> , 2016, 25, 1559-1573.	2.9	25
38	Functional Characterization of a Central Core Disease RyR1 Mutation (p.Y4864H) Associated with Quantitative Defect in RyR1 Protein. <i>Journal of Neuromuscular Diseases</i> , 2015, 2, 421-432.	2.6	16
39	Exertional Heat Stroke and Susceptibility to Malignant Hyperthermia in an Athlete: Evidence for a Link?. <i>Journal of Athletic Training</i> , 2015, 50, 1212-1214.	1.8	25
40	Immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome and recurrent intrauterine fetal death. <i>Lancet, The</i> , 2015, 385, 2120.	13.7	24
41	OCRL-mutated fibroblasts from patients with Dent-2 disease exhibit INPP5B-independent phenotypic variability relatively to Lowe syndrome cells. <i>Human Molecular Genetics</i> , 2015, 24, 994-1006.	2.9	28
42	Mutations of the aurora kinase C gene causing macrozoospermia are the most frequent genetic cause of male infertility in Algerian men. <i>Asian Journal of Andrology</i> , 2015, 17, 68.	1.6	37
43	Adult-onset autosomal dominant centronuclear myopathy due to BIN1 mutations. <i>Brain</i> , 2014, 137, 3160-3170.	7.6	76
44	Prevalence and significance of rare RYR2 variants in arrhythmogenic right ventricular cardiomyopathy/dysplasia: Results of a systematic screening. <i>Heart Rhythm</i> , 2014, 11, 1999-2009.	0.7	58
45	7p22.3 microdeletion disrupting <i>SNX8</i> in a patient presenting with intellectual disability but no tetralogy of Fallot. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2133-2135.	1.2	7
46	Exon Skipping as a Therapeutic Strategy Applied to a RyR1 Mutation Causing Severe Core Myopathy. <i>Biophysical Journal</i> , 2013, 104, 203a.	0.5	0
47	The neuronal endopeptidase ECEL1 is associated with a distinct form of recessive distal arthrogryposis. <i>Human Molecular Genetics</i> , 2013, 22, 1483-1492.	2.9	66
48	Exon Skipping as a Therapeutic Strategy Applied to an <i>RYR1</i> Mutation with Pseudo-Exon Inclusion Causing a Severe Core Myopathy. <i>Human Gene Therapy</i> , 2013, 24, 702-713.	2.7	27
49	Role of Triadin in the Organization of Reticulum Membrane at the Muscle Triad. <i>Journal of Cell Science</i> , 2012, 125, 3443-53.	2.0	20
50	Role of Triadin in the Organization of Reticulum Membrane at the Muscle Triad. <i>Biophysical Journal</i> , 2012, 102, 363a.	0.5	0
51	Identification of the First Mutations in the Human Triadin Gene, Associated to Catecholaminergic Tachycardia, a Pathology of the Cardiac Calcium Release Complex. <i>Biophysical Journal</i> , 2012, 102, 408a-409a.	0.5	0
52	Absence of triadin, a protein of the calcium release complex, is responsible for cardiac arrhythmia with sudden death in human. <i>Human Molecular Genetics</i> , 2012, 21, 2759-2767.	2.9	227
53	Pathological RyR1 Mutations to Identify RyR1 Functional Domains. <i>Biophysical Journal</i> , 2011, 100, 592a-593a.	0.5	0
54	Functional analysis reveals splicing mutations of the <i>CASQ2</i> gene in patients with CPVT: implication for genetic counselling and clinical management. <i>Human Mutation</i> , 2011, 32, 995-999.	2.5	12

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55	Caveolin 3 Is Associated with the Calcium Release Complex and Is Modified via in Vivo Triadin Modification. <i>Biochemistry</i> , 2010, 49, 6130-6135.	2.5	18
56	Triadin Deletion Induces Impaired Skeletal Muscle Function. <i>Journal of Biological Chemistry</i> , 2009, 284, 34918-34929.	3.4	71
57	Triadin: what possible function 20 years later?. <i>Journal of Physiology</i> , 2009, 587, 3117-3121.	2.9	36
58	Triadin Function In Sarcoplasmic Reticulum Structure?. <i>Biophysical Journal</i> , 2009, 96, 237a.	0.5	1
59	Null mutations causing depletion of the type 1 ryanodine receptor (RYR1) are commonly associated with recessive structural congenital myopathies with cores. <i>Human Mutation</i> , 2008, 29, 670-678.	2.5	89
60	Tubulin tyrosination is a major factor affecting the recruitment of CAP-Gly proteins at microtubule plus ends. <i>Journal of Cell Biology</i> , 2006, 174, 839-849.	5.2	271
61	Endosome-to-cytosol transport of viral nucleocapsids. <i>Nature Cell Biology</i> , 2005, 7, 653-664.	10.3	290
62	ARF1 Regulates Nef-Induced CD4 Degradation. <i>Current Biology</i> , 2004, 14, 1056-1064.	3.9	45
63	Role of LBPA and Alix in Multivesicular Liposome Formation and Endosome Organization. <i>Science</i> , 2004, 303, 531-534.	12.6	608
64	Late endosome motility depends on lipids via the small GTPase Rab7. <i>EMBO Journal</i> , 2002, 21, 1289-1300.	7.8	296
65	Mechanism of NADPH Oxidase Activation by the Rac/Rho-GDI Complex. <i>Biochemistry</i> , 2001, 40, 10014-10022.	2.5	82
66	Interactions between Rho GTPases and Rho GDP dissociation inhibitor (Rho-GDI). <i>Biochimie</i> , 2001, 83, 409-414.	2.6	59
67	Characterization of membrane-localized and cytosolic Rac-GTPase-activating proteins in human neutrophil granulocytes: contribution to the regulation of NADPH oxidase. <i>Biochemical Journal</i> , 2001, 355, 851-858.	3.7	28
68	Phosphoinositide-dependent activation of Rho A involves partial opening of the RhoA/Rho-GDI complex. <i>FEBS Journal</i> , 1999, 262, 879-889.	0.2	61
69	Topological organization of the cytosolic activating complex of the superoxide-generating NADPH-oxidase. Pinpointing the sites of interaction between p47phox, p67phox and p40phox using the two-hybrid system. <i>Biochimica Et Biophysica Acta - Molecular Cell Research</i> , 1996, 1312, 39-47.	4.1	49