

Julien FaurÃ©

List of Publications by Year in descending order

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Version: 2024-02-01

69
papers

3,532
citations

201575

27
h-index

138417

58
g-index

72
all docs

72
docs citations

72
times ranked

5556
citing authors

#	ARTICLE	IF	CITATIONS
1	Role of LBPA and Alix in Multivesicular Liposome Formation and Endosome Organization. <i>Science</i> , 2004, 303, 531-534.	6.0	608
2	Late endosome motility depends on lipids via the small GTPase Rab7. <i>EMBO Journal</i> , 2002, 21, 1289-1300.	3.5	296
3	Endosome-to-cytosol transport of viral nucleocapsids. <i>Nature Cell Biology</i> , 2005, 7, 653-664.	4.6	290
4	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.	2.3	271
5	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.	1.4	227
6	Mutations in CFAP43 and CFAP44 cause male infertility and flagellum defects in <i>Trypanosoma</i> and human. <i>Nature Communications</i> , 2018, 9, 686.	5.8	173
7	<sc>SPINK</sc>2 deficiency causes infertility by inducing sperm defects in heterozygotes and azoospermia in homozygotes. <i>EMBO Molecular Medicine</i> , 2017, 9, 1132-1149.	3.3	95
8	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.	1.1	89
9	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.	3.0	86
10	Mechanism of NADPH Oxidase Activation by the Rac/Rho-GDI Complex. <i>Biochemistry</i> , 2001, 40, 10014-10022.	1.2	82
11	Adult-onset autosomal dominant centronuclear myopathy due to BIN1 mutations. <i>Brain</i> , 2014, 137, 3160-3170.	3.7	76
12	Triadin Deletion Induces Impaired Skeletal Muscle Function. <i>Journal of Biological Chemistry</i> , 2009, 284, 34918-34929.	1.6	71
13	The neuronal endopeptidase ECEL1 is associated with a distinct form of recessive distal arthrogryposis. <i>Human Molecular Genetics</i> , 2013, 22, 1483-1492.	1.4	66
14	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
15	Interactions between Rho GTPases and Rho GDP dissociation inhibitor (Rho-GDI). <i>Biochimie</i> , 2001, 83, 409-414.	1.3	59
16	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.	0.3	58
17	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.	1.9	49
18	ARF1 Regulates Nef-Induced CD4 Degradation. <i>Current Biology</i> , 2004, 14, 1056-1064.	1.8	45

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19	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.	0.9	44
20	<i>STAC3</i> variants cause a congenital myopathy with distinctive dysmorphic features and malignant hyperthermia susceptibility. <i>Human Mutation</i> , 2018, 39, 1980-1994.	1.1	42
21	Triadin and CLIMP-63 form a link between triads and microtubules in muscle cells. <i>Journal of Cell Science</i> , 2016, 129, 3744-3755.	1.2	37
22	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.	0.8	37
23	Triadin: what possible function 20 years later?. <i>Journal of Physiology</i> , 2009, 587, 3117-3121.	1.3	36
24	Common and variable clinical, histological, and imaging findings of recessive RYR1-related centronuclear myopathy patients. <i>Neuromuscular Disorders</i> , 2017, 27, 975-985.	0.3	34
25	International Triadin Knockout Syndrome Registry. <i>Circulation Genomic and Precision Medicine</i> , 2019, 12, e002419.	1.6	32
26	â€Dusty core diseaseâ€™ (DuCD): expanding morphological spectrum of RYR1 recessive myopathies. <i>Acta Neuropathologica Communications</i> , 2019, 7, 3.	2.4	31
27	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.	1.7	28
28	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.	1.4	28
29	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.	1.4	27
30	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.	1.4	27
31	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.	0.9	25
32	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.	1.4	25
33	Immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome and recurrent intrauterine fetal death. <i>Lancet, The</i> , 2015, 385, 2120.	6.3	24
34	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.	2.6	24
35	Excitation-Contraction Coupling Alterations in Myopathies. <i>Journal of Neuromuscular Diseases</i> , 2016, 3, 443-453.	1.1	22
36	Deletion of the microtubule-associated protein 6 (MAP6) results in skeletal muscle dysfunction. <i>Skeletal Muscle</i> , 2018, 8, 30.	1.9	21

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37	Mutations in MYLPH Cause a Novel Segmental Amyoplasia that Manifests as Distal Arthrogyposis. <i>American Journal of Human Genetics</i> , 2020, 107, 293-310.	2.6	21
38	Role of Triadin in the Organization of Reticulum Membrane at the Muscle Triad. <i>Journal of Cell Science</i> , 2012, 125, 3443-53.	1.2	20
39	Caveolin 3 Is Associated with the Calcium Release Complex and Is Modified via in Vivo Triadin Modification. <i>Biochemistry</i> , 2010, 49, 6130-6135.	1.2	18
40	TRPV1 variants impair intracellular Ca ²⁺ signaling and may confer susceptibility to malignant hyperthermia. <i>Genetics in Medicine</i> , 2019, 21, 441-450.	1.1	17
41	Interplay between Triadin and Calsequestrin in the Pathogenesis of CPVT in the Mouse. <i>Molecular Therapy</i> , 2020, 28, 171-179.	3.7	17
42	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.	1.1	16
43	Mild clinical presentation in KLHL40-related nemaline myopathy (NEM 8). <i>Neuromuscular Disorders</i> , 2016, 26, 712-716.	0.3	16
44	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.	1.1	13
45	A novel nonsense PIEZO2 mutation in a family with scoliosis and proprioceptive defect. <i>Neuromuscular Disorders</i> , 2019, 29, 75-79.	0.3	13
46	Functional analysis reveals splicing mutations of the CASQ2 gene in patients with CPVT: implication for genetic counselling and clinical management. <i>Human Mutation</i> , 2011, 32, 995-999.	1.1	12
47	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.	2.2	12
48	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.	1.5	11
49	In vivo RyR1 reduction in muscle triggers a core-like myopathy. <i>Acta Neuropathologica Communications</i> , 2020, 8, 192.	2.4	9
50	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.	0.7	7
51	Variations in the TRPV1 gene are associated to exertional heat stroke. <i>Journal of Science and Medicine in Sport</i> , 2020, 23, 1021-1027.	0.6	7
52	Association of fingerprint bodies with rods in a case with mutations in the LMOD3 gene. <i>Neuromuscular Disorders</i> , 2020, 30, 207-212.	0.3	6
53	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.	1.2	6
54	New recessive mutations in <i>SYT2</i> causing severe presynaptic congenital myasthenic syndromes. <i>Neurology: Genetics</i> , 2020, 6, e534.	0.9	6

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55	â€œLowe syndrome: A particularly severe phenotype without clinical kidney involvementâ€ American Journal of Medical Genetics, Part A, 2018, 176, 460-464.	0.7	4
56	Triadin Function In Sarcoplasmic Reticulum Structure?. Biophysical Journal, 2009, 96, 237a.	0.2	1
57	Familial deep cavitating state with a glutathione metabolism defect. Annals of Clinical and Translational Neurology, 2019, 6, 2573-2578.	1.7	1
58	Dynamics of triadin, a muscle-specific triad protein, within sarcoplasmic reticulum subdomains. Molecular Biology of the Cell, 2020, 31, 261-272.	0.9	1
59	A recurrent RYR1 mutation associated with early-onset hypotonia and benign disease course. Acta Neuropathologica Communications, 2021, 9, 155.	2.4	1
60	Characterization of Loss-Of-Function KCNJ2 Mutations in Atypical Andersen Tawil Syndrome. Frontiers in Genetics, 2021, 12, 773177.	1.1	1
61	Diagnostic workup in children with arthrogyrosis: description of practices from a single reference centre, comparison with literature and suggestion of recommendations. Journal of Medical Genetics, 2021, , jmedgenet-2021-107823.	1.5	1
62	Pathological RyR1 Mutations to Identify RyR1 Functional Domains. Biophysical Journal, 2011, 100, 592a-593a.	0.2	0
63	Role of Triadin in the Organization of Reticulum Membrane at the Muscle Triad. Biophysical Journal, 2012, 102, 363a.	0.2	0
64	Identification of the First Mutations in the Human Triadin Gene, Associated to Catecholaminergic Tachycardia, a Pathology of the Cardiac Calcium Release Complex. Biophysical Journal, 2012, 102, 408a-409a.	0.2	0
65	Exon Skipping as a Therapeutic Strategy Applied to a RyR1 Mutation Causing Severe Core Myopathy. Biophysical Journal, 2013, 104, 203a.	0.2	0
66	The Microtubule-Associated Protein CLIMP-63 is a New Member of the Calcium Release Complex. Biophysical Journal, 2016, 110, 181a-182a.	0.2	0
67	Dynamics of Triad Organization. Biophysical Journal, 2017, 112, 99a.	0.2	0
68	Interplay between Triadin and Calsequestrin in the Pathogenesis of CPVT. Biophysical Journal, 2018, 114, 618a.	0.2	0
69	Response to Hall etÂal.. American Journal of Human Genetics, 2020, 107, 1188-1189.	2.6	0