Julien FaurÃ©

List of Publications by Year
in descending order

69	3,532	201575	138417
papers	citations	27 h-index	58 g-index

72	72	72	5556
all docs	docs citations	times ranked	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. Journal of Medical Genetics, 2021, 58, 602-608.	1.5	11
2	Role of oculocerebrorenal syndrome of Lowe (OCRL) protein in megakaryocyte maturation, platelet production and functions: a study in patients with Lowe syndrome. British Journal of Haematology, 2021, 192, 909-921.	1.2	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. Frontiers in Immunology, 2021, 12, 639226.	2.2	12
4	A recurrent RYR1 mutation associated with early-onset hypotonia and benign disease course. Acta Neuropathologica Communications, 2021, 9, 155.	2.4	1
5	Characterization of Loss-Of-Function KCNJ2 Mutations in Atypical Andersen Tawil Syndrome. Frontiers in Genetics, 2021, 12, 773177.	1.1	1
6	Diagnostic workup in children with arthrogryposis: 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
7	Interplay between Triadin and Calsequestrin in the Pathogenesis of CPVT in the Mouse. Molecular Therapy, 2020, 28, 171-179.	3.7	17
8	Association of fingerprint bodies with rods in a case with mutations in the LMOD3 gene. Neuromuscular Disorders, 2020, 30, 207-212.	0.3	6
9	Pathogenic Variants in the Myosin Chaperone UNC-45B Cause Progressive Myopathy with Eccentric Cores. American Journal of Human Genetics, 2020, 107, 1078-1095.	2.6	24
10	Response to Hall etÂal.. American Journal of Human Genetics, 2020, 107, 1188-1189.	2.6	0
11	In vivo RyR1 reduction in muscle triggers a core-like myopathy. Acta Neuropathologica Communications, 2020, 8, 192.	2.4	9
12	Mutations in MYLPF Cause a Novel Segmental Amyoplasia that Manifests as Distal Arthrogryposis. American Journal of Human Genetics, 2020, 107, 293-310.	2.6	21
13	Variations in the TRPV1 gene are associated to exertional heat stroke. Journal of Science and Medicine in Sport, 2020, 23, 1021-1027.	0.6	7
14	Dynamics of triadin, a muscle-specific triad protein, within sarcoplasmic reticulum subdomains. Molecular Biology of the Cell, 2020, 31, 261-272.	0.9	1
15	New recessive mutations in SYT2 causing severe presynaptic congenital myasthenic syndromes. Neurology: Genetics, 2020, 6, e534.	0.9	6
16	TRPV1 variants impair intracellular Ca2+ signaling and may confer susceptibility to malignant hyperthermia. Genetics in Medicine, 2019, 21, 441-450.	1.1	17
17	Familial deep cavitating state with a glutathione metabolism defect. Annals of Clinical and Translational Neurology, 2019, 6, 2573-2578.	1.7	1
18	A National French consensus on gene lists for the diagnosis of myopathies using next-generation sequencing. European Journal of Human Genetics, 2019, 27, 349-352.	1.4	27

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19	A novel nonsense PIEZO2 mutation in a family with scoliosis and proprioceptive defect. Neuromuscular Disorders, 2019, 29, 75-79.	0.3	13
20	â€˜Dusty core diseaseâ€™ (DuCD): expanding morphological spectrum of RYR1 recessive myopathies. Acta Neuropathologica Communications, 2019, 7, 3.	2.4	31
21	International Triadin Knockout Syndrome Registry. Circulation Genomic and Precision Medicine, 2019, 12, e002419.	1.6	32
22	Mutations in CFAP43 and CFAP44 cause male infertility and flagellum defects in Trypanosoma and human. Nature Communications, 2018, 9, 686.	5.8	173
23	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
24	ALK fusion variants detection by targeted RNA-next generation sequencing and clinical responses to crizotinib in ALK-positive non-small cell lung cancer. Lung Cancer, 2018, 116, 15-24.	0.9	44
25	Deletion of the microtubule-associated protein 6 (MAP6) results in skeletal muscle dysfunction. Skeletal Muscle, 2018, 8, 30.	1.9	21
26	Interplay between Triadin and Calsequestrin in the Pathogenesis of CPVT. Biophysical Journal, 2018, 114, 618a.	0.2	0
27	STAC3variants cause a congenital myopathy with distinctive dysmorphic features and malignant hyperthermia susceptibility. Human Mutation, 2018, 39, 1980-1994.	1.1	42
28	Common and variable clinical, histological, and imaging findings of recessive RYR1-related centronuclear myopathy patients. Neuromuscular Disorders, 2017, 27, 975-985.	0.3	34
29	<scp>SPINK</scp>2 deficiency causes infertility by inducing sperm defects in heterozygotes and azoospermia inÂhomozygotes. EMBO Molecular Medicine, 2017, 9, 1132-1149.	3.3	95
30	Dynamics of Triad Organization. Biophysical Journal, 2017, 112, 99a.	0.2	0
31	Functional Characterization and Rescue of a Deep Intronic Mutation inOCRLGene Responsible for Lowe Syndrome. Human Mutation, 2017, 38, 152-159.	1.1	13
32	Excitation-Contraction Coupling Alterations in Myopathies. Journal of Neuromuscular Diseases, 2016, 3, 443-453.	1.1	22
33	The Microtubule-Associated Protein CLIMP-63 is a New Member of the Calcium Release Complex. Biophysical Journal, 2016, 110, 181a-182a.	0.2	0
34	Phospholipase A2 Receptorâ€“Related Membranous Nephropathy and Mannan-Binding Lectin Deficiency. Journal of the American Society of Nephrology: JASN, 2016, 27, 3539-3544.	3.0	86
35	Triadin and CLIMP-63 form a link between triads and microtubules in muscle cells. Journal of Cell Science, 2016, 129, 3744-3755.	1.2	37
36	Mild clinical presentation in KLHL40-related nemaline myopathy (NEM 8). Neuromuscular Disorders, 2016, 26, 712-716.	0.3	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. Human Molecular Genetics, 2016, 25, 1559-1573.	1.4	25
38	Functional Characterization of a Central Core Disease RyR1 Mutation (p.Y4864H) Associated with Quantitative Defect in RyR1 Protein. Journal of Neuromuscular Diseases, 2015, 2, 421-432.	1.1	16
39	Exertional Heat Stroke and Susceptibility to Malignant Hyperthermia in an Athlete: Evidence for a Link?. Journal of Athletic Training, 2015, 50, 1212-1214.	0.9	25
40	Immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome and recurrent intrauterine fetal death. Lancet, The, 2015, 385, 2120.	6.3	24
41	OCRL-mutated fibroblasts from patients with Dent-2 disease exhibit INPP5B-independent phenotypic variability relatively to Lowe syndrome cells. Human Molecular Genetics, 2015, 24, 994-1006.	1.4	28
42	Mutations of the aurora kinase C gene causing macrozoospermia are the most frequent genetic cause of male infertility in Algerian men. Asian Journal of Andrology, 2015, 17, 68.	0.8	37
43	Adult-onset autosomal dominant centronuclear myopathy due to BIN1 mutations. Brain, 2014, 137, 3160-3170.	3.7	76
44	Prevalence and significance of rare RYR2 variants in arrhythmogenic right ventricular cardiomyopathy/dysplasia: Results of a systematic screening. Heart Rhythm, 2014, 11, 1999-2009.	0.3	58
45	7p22.3 microdeletion disrupting SNX8 in a patient presenting with intellectual disability but no tetralogy of Fallot. American Journal of Medical Genetics, Part A, 2014, 164, 2133-2135.	0.7	7
46	Exon Skipping as a Therapeutic Strategy Applied to a RyR1 Mutation Causing Severe Core Myopathy. Biophysical Journal, 2013, 104, 203a.	0.2	0
47	The neuronal endopeptidase ECEL1 is associated with a distinct form of recessive distal arthrogryposis. Human Molecular Genetics, 2013, 22, 1483-1492.	1.4	66
48	Exon Skipping as a Therapeutic Strategy Applied to anRYR1Mutation with Pseudo-Exon Inclusion Causing a Severe Core Myopathy. Human Gene Therapy, 2013, 24, 702-713.	1.4	27
49	Role of Triadin in the Organization of Reticulum Membrane at the Muscle Triad. Journal of Cell Science, 2012, 125, 3443-53.	1.2	20
50	Role of Triadin in the Organization of Reticulum Membrane at the Muscle Triad. Biophysical Journal, 2012, 102, 363a.	0.2	0
51	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
52	Absence of triadin, a protein of the calcium release complex, is responsible for cardiac arrhythmia with sudden death in human. Human Molecular Genetics, 2012, 21, 2759-2767.	1.4	227
53	Pathological RyR1 Mutations to Identify RyR1 Functional Domains. Biophysical Journal, 2011, 100, 592a-593a.	0.2	0
54	Functional analysis reveals splicing mutations of the CASQ2 gene in patients with CPVT: implication for genetic counselling and clinical management. Human Mutation, 2011, 32, 995-999.	1.1	12

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55	Caveolin 3 Is Associated with the Calcium Release Complex and Is Modified via in Vivo Triadin Modification. Biochemistry, 2010, 49, 6130-6135.	1.2	18
56	Triadin Deletion Induces Impaired Skeletal Muscle Function. Journal of Biological Chemistry, 2009, 284, 34918-34929.	1.6	71
57	Triadin: what possible function 20 years later?. Journal of Physiology, 2009, 587, 3117-3121.	1.3	36
58	Triadin Function In Sarcoplasmic Reticulum Structure?. Biophysical Journal, 2009, 96, 237a.	0.2	1
59	Null mutations causing depletion of the type 1 ryanodine receptor (RYR1) are commonly associated with recessive structural congenital myopathies with cores. Human Mutation, 2008, 29, 670-678.	1.1	89
60	Tubulin tyrosination is a major factor affecting the recruitment of CAP-Gly proteins at microtubule plus ends. Journal of Cell Biology, 2006, 174, 839-849.	2.3	271
61	Endosome-to-cytosol transport of viral nucleocapsids. Nature Cell Biology, 2005, 7, 653-664.	4.6	290
62	ARF1 Regulates Nef-Induced CD4 Degradation. Current Biology, 2004, 14, 1056-1064.	1.8	45
63	Role of LBPA and Alix in Multivesicular Liposome Formation and Endosome Organization. Science, 2004, 303, 531-534.	6.0	608
64	Late endosome motility depends on lipids via the small GTPase Rab7. EMBO Journal, 2002, 21, 1289-1300.	3.5	296
65	Mechanism of NADPH Oxidase Activation by the Rac/Rho-GDI Complexâ€. Biochemistry, 2001, 40, 10014-10022.	1.2	82
66	Interactions between Rho GTPases and Rho GDP dissociation inhibitor (Rho-GDI). Biochimie, 2001, 83, 409-414.	1.3	59
67	Characterization of membrane-localized and cytosolic Rac-GTPase-activating proteins in human neutrophil granulocytes: contribution to the regulation of NADPH oxidase. Biochemical Journal, 2001, 355, 851-858.	1.7	28
68	Phosphoinositide-dependent activation of Rho A involves partial opening of the RhoA/Rho-GDI complex. FEBS Journal, 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. Biochimica Et Biophysica Acta - Molecular Cell Research, 1996, 1312, 39-47.	1.9	49