Pauline Gaignard

List of Publications by Year
in descending order

28 papers	730 citations	643344 15 h-index	620720 26 g-index
29 all docs	29 docs citations	29 times ranked	1702 citing authors

#	Article	IF	Citations
1	Heterogeneity of PNPT1 neuroimaging: mitochondriopathy, interferonopathy or both?. Journal of Medical Genetics, 2022, 59, 204-208.	1.5	6
2	FDX2 and ISCU Gene Variations Lead to Rhabdomyolysis With Distinct Severity and Iron Regulation. Neurology: Genetics, 2022, 8, e648.	0.9	4
3	Adenosine kinase deficiency: Three new cases and diagnostic value of hypermethioninemia. Molecular Genetics and Metabolism, 2021, 132, 38-43.	0.5	8
4	Enteral tube feeding in patients receiving dietary treatment for metabolic diseases: A retrospective analysis in a large French cohort. Molecular Genetics and Metabolism Reports, 2021, 26, 100655.	0.4	7
5	Response to systemic therapy in fumarate hydrataseâ€“deficient renal cell carcinoma. European Journal of Cancer, 2021, 151, 106-114.	1.3	18
6	Adult Cerebellar Ataxia, Axonal Neuropathy, and Sensory Impairments Caused by Biallelic SCO2 Variants. Neurology: Genetics, 2021, 7, e630.	0.9	1
7	Mitochondrial dysfunction caused by novel ATAD3A mutations. Molecular Genetics and Metabolism, 2020, 131, 107-113.	0.5	17
8	Brain White Matter Lesions and Presumed Crohnâ€™s Disease: Did You Consider MNGIE?. Canadian Journal of Neurological Sciences, 2020, 47, 572-575.	0.3	0
9	Intranasal administration of progesterone: A potential efficient route of delivery for cerebroprotection after acute brain injuries. Neuropharmacology, 2019, 145, 283-291.	2.0	28
10	Expanding and Underscoring the Hepatoâ€Encephalopathic Phenotype of QIL1/MIC13. Hepatology, 2019, 70, 1066-1070.	3.6	17
11	MyoNeuroGastroIntestinal Encephalopathy: Natural History and Means for Early Diagnosis. Gastroenterology, 2019, 156, 1525-1527.e4.	0.6	7
12	Steroids in Stroke with Special Reference to Progesterone. Cellular and Molecular Neurobiology, 2019, 39, 551-568.	1.7	29
13	Long-term liver disease in methylmalonic and propionic acidemias. Molecular Genetics and Metabolism, 2018, 123, 433-440.	0.5	23
14	Les dÃ©ficits de la chaÃ®ne respiratoire : dÃ©marche diagnostique des cytopathies mitochondriales. Revue Francophone Des Laboratoires, 2018, 2018, 26-35.	0.0	1
15	Primary carnitine deficiency in a 57-year-old patient with recurrent exertional rhabdomyolysis. BMJ Case Reports, 2018, 2018, bcr-2018-224272.	0.2	0
16	UQCRC2 mutation in a patient with mitochondrial complex III deficiency causing recurrent liver failure, lactic acidosis and hypoglycemia. Journal of Human Genetics, 2017, 62, 729-731.	1.1	30
17	Role of Sex Hormones on Brain Mitochondrial Function, with Special Reference to Aging and Neurodegenerative Diseases. Frontiers in Aging Neuroscience, 2017, 9, 406.	1.7	82
18	Bovine and murine models highlight novel roles for SLC25A46 in mitochondrial dynamics and metabolism, with implications for human and animal health. PLoS Genetics, 2017, 13, e1006597.	1.5	18

#	Article	IF	Citations
19	Abnormal Glycosylation Profile and High Alpha-Fetoprotein in a Patient with Twinkle Variants. JIMD Reports, 2016, 29, 109-113.	0.7	5
20	Role of lipid phosphate phosphatase 3 in human aortic endothelial cell function. Cardiovascular Research, 2016, 112, 702-713.	1.8	25
21	Progesterone reduces brain mitochondrial dysfunction after transient focal ischemia in male and female mice. Journal of Cerebral Blood Flow and Metabolism, 2016, 36, 562-568.	2.4	29
22	QIL1 mutation causes MICOS disassembly and early onset fatal mitochondrial encephalopathy with liver disease. ELife, 2016, 5, .	2.8	46
23	Effect of Sex Differences on Brain Mitochondrial Function and Its Suppression by Ovariectomy and in Aged Mice. Endocrinology, 2015, 156, 2893-2904.	1.4	104
24	Liver transcript analysis reveals aberrant splicing due to silent and intronic variations in the ABCB11 gene. Molecular Genetics and Metabolism, 2014, 113, 225-229.	0.5	12
25	Mutations in CYC1, Encoding Cytochrome c1 Subunit of Respiratory Chain Complex III, Cause Insulin-Responsive Hyperglycemia. American Journal of Human Genetics, 2013, 93, 384-389.	2.6	61
26	Characterization of seven novel mutations on the HEXB gene in French Sandhoff patients. Gene, 2013, 512, 521-526.	1.0	20
27	Mitochondrial Infantile Liver Disease due to TRMU Gene Mutations: Three New Cases. JIMD Reports, 2013, 11, 117-123.	0.7	37
28	Prevalence of rare mitochondrial DNA mutations in mitochondrial disorders. Journal of Medical Genetics, 2013, 50, 704-714.	1.5	95