Fabiana L Motta

List of Publications by Year
in descending order

28	457	686830 13	713013 21
papers	citations	h-index	g-index

28	28	28	932
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A rare canonical splice-site variant in VPS13B causes attenuated Cohen syndrome. Ophthalmic Genetics, 2022, 43, 110-115.	0.5	2
2	Expanding the Phenotypic and Genotypic Spectrum of Bietti Crystalline Dystrophy. Genes, 2021, 12, 713.	1.0	7
3	Analysis of an NGS retinopathy panel detects chromosome 1 uniparental isodisomy in a patient with RPE65-related leber congenital amaurosis. Ophthalmic Genetics, 2021, 42, 553-560.	0.5	2
4	Novel Mutation in CRYBB3 Causing Pediatric Cataract and Microphthalmia. Genes, 2021, 12, 1069.	1.0	2
5	Pathogenicity Reclassification of RPE65 Missense Variants Related to Leber Congenital Amaurosis and Early-Onset Retinal Dystrophy. Genes, 2020, 11, 24.	1.0	14
6	Clinical and molecular findings in a cohort of 152 Brazilian severe early onset inherited retinal dystrophy patients. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 728-752.	0.7	20
7	Synonymous Variant in the CHM Gene Causes Aberrant Splicing in Choroideremia. , 2020, 61, 38.		4
8	Retinitis Pigmentosa Due to Rp1 Biallelic Variants. Scientific Reports, 2020, 10, 1603.	1.6	8
9	TUBGCP4â€“ associated microcephaly and chorioretinopathy. Ophthalmic Genetics, 2020, 41, 189-193.	0.5	11
10	Correlation between GLA variants and alpha-Galactosidase A profile in dried blood spot: an observational study in Brazilian patients. Orphanet Journal of Rare Diseases, 2020, 15, 30.	1.2	15
11	A New Mutation in IDS Gene Causing Hunter Syndrome: A Case Report. Frontiers in Genetics, 2019, 10, 1383.	1.1	2
12	Relative frequency of inherited retinal dystrophies in Brazil. Scientific Reports, 2018, 8, 15939.	1.6	35
13	Exfoliation syndrome associated with LOXL1 gene polymorphisms in a Black patient from Latin America: a case report. Arquivos Brasileiros De Oftalmologia, 2018, 81, 437-439.	0.2	2
14	Variants in the gene in a Brazilian population with Stargardt disease. Molecular Vision, 2018, 24, 546-559.	1.1	12
15	PROM1 gene variations in Brazilian patients with macular dystrophy. Ophthalmic Genetics, 2017, 38, 39-42.	0.5	8
16	The correlation between CRB1 variants and the clinical severity of Brazilian patients with different inherited retinal dystrophy phenotypes. Scientific Reports, 2017, 7, 8654.	1.6	17
17	Leveraging spliceâ€affecting variant predictors and a minigene validation system to identify Mendelian diseaseâ€causing variants among exonâ€captured variants of uncertain significance. Human Mutation, 2017, 38, 1521-1533.	1.1	27
18	Novel Complex ABCA4 Alleles in Brazilian Patients With Stargardt Disease: Genotypeâ€“Phenotype Correlation. , 2017, 58, 5723.		12

#	Article	IF	Citations
19	Novel GAA mutations in patients with Pompe disease. Gene, 2015, 561, 124-131.	1.0	20
20	Kinin B1 receptor gene ablation affects hypothalamic CART production^b. Biological Chemistry, 2013, 394, 901-908.	1.2	2
21	Kinin B1 Receptor in Adipocytes Regulates Glucose Tolerance and Predisposition to Obesity. PLoS ONE, 2012, 7, e44782.	1.1	28
22	New mutations in the GLA gene in Brazilian families with Fabry disease. Journal of Human Genetics, 2012, 57, 347-351.	1.1	22
23	Short-Term Withdrawal of Mitogens Prior to Plating Increases Neuronal Differentiation of Human Neural Precursor Cells. PLoS ONE, 2009, 4, e4642.	1.1	12
24	Effects of FGF-2 and EGF removal on the differentiationof mouse neural precursor cells. Anais Da Academia Brasileira De Ciencias, 2009, 81, 443-452.	0.3	33
25	Multiple RNAs from the mouse carboxypeptidase M locus: functional RNAs or transcription noise?. BMC Molecular Biology, 2009, 10, 7.	3.0	3
26	GCN2 activation and eIF2Î± phosphorylation in the maturation of mouse oocytes. Biochemical and Biophysical Research Communications, 2009, 378, 41-44.	1.0	15
27	Chemically-Induced RAT Mesenchymal Stem Cells Adopt Molecular Properties of Neuronal-Like Cells but Do Not Have Basic Neuronal Functional Properties. PLoS ONE, 2009, 4, e5222.	1.1	76
28	Neuropathic Pain-Like Behavior after Brachial Plexus Avulsion in Mice: The Relevance of Kinin B₁and B₂Receptors. Journal of Neuroscience, 2008, 28, 2856-2863.	1.7	46