Pietro Farinelli

List of Publications by Year
in descending order

15	712	759233 12	996975 15
papers	citations	h-index	g-index

18	18	18	1151
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	CEP78 functions downstream of CEP350 to control biogenesis of primary cilia by negatively regulating CP110 levels. ELife, 2021, 10, .	6.0	29
2	Functional characterization of the first missense variant in CEP78 , a founder allele associated with coneâ€rod dystrophy, hearing loss, and reduced male fertility. Human Mutation, 2020, 41, 998-1011.	2.5	15
3	A frequent variant in the Japanese population determines quasi-Mendelian inheritance of rare retinal ciliopathy. Nature Communications, 2019, 10, 2884.	12.8	21
4	A novel missense variant in IDH3A causes autosomal recessive retinitis pigmentosa. Ophthalmic Genetics, 2019, 40, 177-181.	1.2	10
5	IFT20 modulates ciliary PDGFRÎ± signaling by regulating the stability of Cbl E3 ubiquitin ligases. Journal of Cell Biology, 2018, 217, 151-161.	5.2	54
6	A large multiexonic genomic deletion within the <scp>ALMS1</scp> gene causes AlstrÃ¶m syndrome in a consanguineous Pakistani family. Clinical Genetics, 2016, 89, 510-511.	2.0	5
7	Isolated and Syndromic Retinal Dystrophy Caused by Biallelic Mutations in RCBTB1 , a Gene Implicated in Ubiquitination. American Journal of Human Genetics, 2016, 99, 470-480.	6.2	39
8	Mutations in the polyglutamylase geneTTLL5, expressed in photoreceptor cells and spermatozoa, are associated with cone-rod degeneration and reduced male fertility. Human Molecular Genetics, 2016, 25, ddw282.	2.9	27
9	Mutations in CEP78 Cause Cone-Rod Dystrophy and Hearing Loss Associated with Primary-Cilia Defects. American Journal of Human Genetics, 2016, 99, 770-776.	6.2	44
10	Interactome analysis reveals that FAM161A, deficient in recessive retinitis pigmentosa, is a component of the Golgi-centrosomal network. Human Molecular Genetics, 2015, 24, 3359-3371.	2.9	19
11	Identification of a Common Non-Apoptotic Cell Death Mechanism in Hereditary Retinal Degeneration. PLoS ONE, 2014, 9, e112142.	2.5	191
12	DNA methylation and differential gene regulation in photoreceptor cell death. Cell Death and Disease, 2014, 5, e1558-e1558.	6.3	47
13	Retinitis Pigmentosa: overâ€expression of antiâ€ageing protein Klotho in degenerating photoreceptors. Journal of Neurochemistry, 2013, 127, 868-879.	3.9	14
14	Calpain and PARP Activation during Photoreceptor Cell Death in P23H and S334ter Rhodopsin Mutant Rats. PLoS ONE, 2011, 6, e22181.	2.5	94
15	Excessive HDAC activation is critical for neurodegeneration in the rd1 mouse. Cell Death and Disease, 2010, 1, e24-e24.	6.3	100