Polona Le Quesne Stabej

List of Publications by Year
in descending order

15	621	840119 11	940134 16
papers	citations	h-index	g-index

16	16	16	1546
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Comprehensive sequence analysis of nine Usher syndrome genes in the UK National Collaborative Usher Study. Journal of Medical Genetics, 2012, 49, 27-36.	1.5	152
2	Rapid Paediatric Sequencing (RaPS): comprehensive real-life workflow for rapid diagnosis of critically ill children. Journal of Medical Genetics, 2018, 55, 721-728.	1.5	98
3	Mutations in TOP3A Cause a Bloom Syndrome-like Disorder. American Journal of Human Genetics, 2018, 103, 221-231.	2.6	65
4	Mutations in EXTL3 Cause Neuro-immuno-skeletal Dysplasia Syndrome. American Journal of Human Genetics, 2017, 100, 281-296.	2.6	59
5	STAG3 truncating variant as the cause of primary ovarian insufficiency. European Journal of Human Genetics, 2016, 24, 135-138.	1.4	53
6	Screening for duplications, deletions and a common intronic mutation detects 35% of second mutations in patients with USH2A monoallelic mutations on Sanger sequencing. Orphanet Journal of Rare Diseases, 2013, 8, 122.	1.2	47
7	Natural History and Retinal Structure in Patients with Usher Syndrome Type 1 Owing to MYO7A Mutation. Ophthalmology, 2014, 121, 580-587.	2.5	27
8	Seizures Due to a KCNQ2 Mutation: Treatment with Vitamin B6. JIMD Reports, 2015, 27, 79-84.	0.7	22
9	MUTATIONS IN THE USH1C GENE ASSOCIATED WITH SECTOR RETINITIS PIGMENTOSA AND HEARING LOSS. Retina, 2011, 31, 1708-1716.	1.0	20
10	Mutations in MAGEL2 and L1CAM Are Associated With Congenital Hypopituitarism and Arthrogryposis. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 5737-5750.	1.8	17
11	A recessive PRDM13 mutation results in congenital hypogonadotropic hypogonadism and cerebellar hypoplasia. Journal of Clinical Investigation, 2021, 131, .	3.9	16
12	ZSWIM7Is Associated With Human Female Meiosis and Familial Primary Ovarian Insufficiency. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e254-e263.	1.8	13
13	An example of the utility of genomic analysis for fast and accurate clinical diagnosis of complex rare phenotypes. Orphanet Journal of Rare Diseases, 2017, 12, 24.	1.2	10
14	Pathogenic variants in the human m6A reader YTHDC2 are associated with primary ovarian insufficiency. JCI Insight, 2022, 7, .	2.3	8
15	Pathogenic variants in RNPC3 are associated with hypopituitarism and primary ovarian insufficiency. Genetics in Medicine, 2022, 24, 384-397.	1.1	4