Polona Le Quesne Stabej

List of Publications by Year
in descending order

15 papers	621 citations	840585 11 h-index	940416 16 g-index
16 all docs	16 docs citations	16 times ranked	1546 citing authors

#	Article	IF	Citations
1	ZSWIM7Is Associated With Human Female Meiosis and Familial Primary Ovarian Insufficiency. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e254-e263.	1.8	13
2	Pathogenic variants in RNPC3 are associated with hypopituitarism and primary ovarian insufficiency. Genetics in Medicine, 2022, 24, 384-397.	1.1	4
3	Pathogenic variants in the human m6A reader YTHDC2 are associated with primary ovarian insufficiency. JCI Insight, 2022, 7, .	2.3	8
4	A recessive PRDM13 mutation results in congenital hypogonadotropic hypogonadism and cerebellar hypoplasia. Journal of Clinical Investigation, 2021, 131, .	3.9	16
5	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
6	Mutations in TOP3A Cause a Bloom Syndrome-like Disorder. American Journal of Human Genetics, 2018, 103, 221-231.	2.6	65
7	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
8	Mutations in EXTL3 Cause Neuro-immuno-skeletal Dysplasia Syndrome. American Journal of Human Genetics, 2017, 100, 281-296.	2.6	59
9	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
10	STAG3 truncating variant as the cause of primary ovarian insufficiency. European Journal of Human Genetics, 2016, 24, 135-138.	1.4	53
11	Seizures Due to a KCNQ2 Mutation: Treatment with Vitamin B6. JIMD Reports, 2015, 27, 79-84.	0.7	22
12	Natural History and Retinal Structure in Patients with Usher Syndrome Type 1 Owing to MYO7A Mutation. Ophthalmology, 2014, 121, 580-587.	2.5	27
13	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
14	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
15	MUTATIONS IN THE USH1C GENE ASSOCIATED WITH SECTOR RETINITIS PIGMENTOSA AND HEARING LOSS. Retina, 2011, 31, 1708-1716.	1.0	20