

Torunn Fiskerstrand

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/3297602/publications.pdf>

Version: 2024-02-01

16
papers

864
citations

1040056

9
h-index

940533

16
g-index

17
all docs

17
docs citations

17
times ranked

1742
citing authors

#	ARTICLE	IF	CITATIONS
1	The intronic BRCA1 c.5407-25T>A variant causing partly skipping of exon 23—a likely pathogenic variant with reduced penetrance?. <i>European Journal of Human Genetics</i> , 2020, 28, 1078-1086.	2.8	6
2	Comparison of pre-analytical conditions for quantification of serotonin in platelet-poor plasma. <i>Practical Laboratory Medicine</i> , 2019, 17, e00136.	1.3	2
3	The Discovery of a LEMD2-Associated Nuclear Envelopathy with Early Progeroid Appearance Suggests Advanced Applications for AI-Driven Facial Phenotyping. <i>American Journal of Human Genetics</i> , 2019, 104, 749-757.	6.2	41
4	Genetic and transcriptional analysis of inflammatory bowel disease-associated pathways in patients with <i>GUCY2C</i> -linked familial diarrhea. <i>Scandinavian Journal of Gastroenterology</i> , 2018, 53, 1264-1273.	1.5	9
5	Elevated cerebrospinal fluid protein in <i>POLG</i> -related epilepsy: Diagnostic and prognostic implications. <i>Epilepsia</i> , 2018, 59, 1595-1602.	5.1	6
6	Duplicated Enhancer Region Increases Expression of <i>CTSB</i> and Segregates with Keratolytic Winter Erythema in South African and Norwegian Families. <i>American Journal of Human Genetics</i> , 2017, 100, 737-750.	6.2	35
7	The presence of anaemia negatively influences survival in patients with <i>POLG</i> disease. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 861-866.	3.6	8
8	Guanylate Cyclase C Activation Shapes the Intestinal Microbiota in Patients with Familial Diarrhea and Increased Susceptibility for Crohn's Disease. <i>Inflammatory Bowel Diseases</i> , 2017, 23, 1752-1761.	1.9	13
9	Prolonged intestinal transit and diarrhea in patients with an activating <i>GUCY2C</i> mutation. <i>PLoS ONE</i> , 2017, 12, e0185496.	2.5	19
10	A Cross-Sectional Study of the Prevalence of Gastrointestinal Symptoms and Pathology in Patients With Common Variable Immunodeficiency. <i>American Journal of Gastroenterology</i> , 2016, 111, 1467-1475.	0.4	85
11	RareVariantVis: new tool for visualization of causative variants in rare monogenic disorders using whole genome sequencing data. <i>Bioinformatics</i> , 2016, 32, 3018-3020.	4.1	7
12	Dominant Mutations in the Autoimmune Regulator <i>AIRE</i> Are Associated with Common Organ-Specific Autoimmune Diseases. <i>Immunity</i> , 2015, 42, 1185-1196.	14.3	246
13	Familial Diarrhea Syndrome Caused by an Activating <i>GUCY2C</i> Mutation. <i>New England Journal of Medicine</i> , 2012, 366, 1586-1595.	27.0	175
14	Mutations in <i>ABHD12</i> Cause the Neurodegenerative Disease PHARC: An Inborn Error of Endocannabinoid Metabolism. <i>American Journal of Human Genetics</i> , 2010, 87, 410-417.	6.2	188
15	Identification of a Gene for Renal-Hepatic-Pancreatic Dysplasia by Microarray-Based Homozygosity Mapping. <i>Journal of Molecular Diagnostics</i> , 2010, 12, 125-131.	2.8	22
16	Frontometaphyseal dysplasia: Mutations in <i>FLNA</i> and phenotypic diversity (<i>Am J Med Genet</i> 140A:) Tj ETQq0 0 0 rgBT, /Overlock 10 Tf 50		