Torunn Fiskerstrand

List of Publications by Year
in descending order

16	864	1040056 9	940533 16
papers	citations	h-index	g-index

17	17	17	1742
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Dominant Mutations in the Autoimmune Regulator AIRE Are Associated with Common Organ-Specific Autoimmune Diseases. Immunity, 2015, 42, 1185-1196.	14.3	246
2	Mutations in ABHD12 Cause the Neurodegenerative Disease PHARC: An Inborn Error of Endocannabinoid Metabolism. American Journal of Human Genetics, 2010, 87, 410-417.	6.2	188
3	Familial Diarrhea Syndrome Caused by an ActivatingGUCY2CMutation. New England Journal of Medicine, 2012, 366, 1586-1595.	27.0	175
4	A Cross-Sectional Study of the Prevalence of Gastrointestinal Symptoms and Pathology in Patients With Common Variable Immunodeficiency. American Journal of Gastroenterology, 2016, 111, 1467-1475.	0.4	85
5	The Discovery of a LEMD2-Associated Nuclear Envelopathy with Early Progeroid Appearance Suggests Advanced Applications for AI-Driven Facial Phenotyping. American Journal of Human Genetics, 2019, 104, 749-757.	6.2	41
6	Duplicated Enhancer Region Increases Expression of CTSB and Segregates with Keratolytic Winter Erythema in South African and Norwegian Families. American Journal of Human Genetics, 2017, 100, 737-750.	6.2	35
7	Identification of a Gene for Renal-Hepatic-Pancreatic Dysplasia by Microarray-Based Homozygosity Mapping. Journal of Molecular Diagnostics, 2010, 12, 125-131.	2.8	22
8	Prolonged intestinal transit and diarrhea in patients with an activating GUCY2C mutation. PLoS ONE, 2017, 12, e0185496.	2.5	19
9	Guanylate Cyclase C Activation Shapes the Intestinal Microbiota in Patients with Familial Diarrhea and Increased Susceptibility for CrohnÊ¼s Disease. Inflammatory Bowel Diseases, 2017, 23, 1752-1761.	1.9	13
10	Genetic and transcriptional analysis of inflammatory bowel disease-associated pathways in patients with GUCY2C-linked familial diarrhea. Scandinavian Journal of Gastroenterology, 2018, 53, 1264-1273.	1.5	9
11	The presence of anaemia negatively influences survival in patients with POLG disease. Journal of Inherited Metabolic Disease, 2017, 40, 861-866.	3.6	8
12	RareVariantVis: new tool for visualization of causative variants in rare monogenic disorders using whole genome sequencing data. Bioinformatics, 2016, 32, 3018-3020.	4.1	7
13	Elevated cerebrospinal fluid protein in <scp>POLG</scp>â€related epilepsy: Diagnostic and prognostic implications. Epilepsia, 2018, 59, 1595-1602.	5.1	6
14	The intronic BRCA1 c.5407-25T>A variant causing partly skipping of exon 23â€”a likely pathogenic variant with reduced penetrance?. European Journal of Human Genetics, 2020, 28, 1078-1086.	2.8	6
15	Frontometaphyseal dysplasia: Mutations inFLNA and phenotypic diversity (Am J Med Genet 140A:) Tj ETQq1 1 0.784314 rgBT /Overlo	1.2	2
16	Comparison of pre-analytical conditions for quantification of serotonin in platelet-poor plasma. Practical Laboratory Medicine, 2019, 17, e00136.	1.3	2