Ingvild Aukrust

List of Publications by Year
in descending order

25 papers	721 citations	687363 13 h-index	610901 24 g-index
25 all docs	25 docs citations	25 times ranked	2117 citing authors

#	Article	IF	Citations
1	BRCA1 Norway: comparison of classification for BRCA1 germline variants detected in families with suspected hereditary breast and ovarian cancer between different laboratories. Familial Cancer, 2022, 21, 389-398.	1.9	2
2	Structural and biophysical characterization of transcription factor HNF-1A as a tool to study MODY3 diabetes variants. Journal of Biological Chemistry, 2022, 298, 101803.	3.4	4
3	Clinical features and molecular genetics of patients with ABCA4â€retinal dystrophies. Acta Ophthalmologica, 2021, 99, e733-e746.	1.1	6
4	Genetic Dominant Variants in STUB1, Segregating in Families with SCA48, Display In Vitro Functional Impairments Indistinctive from Recessive Variants Associated with SCAR16. International Journal of Molecular Sciences, 2021, 22, 5870.	4.1	10
5	Pellinoâ€2 in nonimmune cells: novel interaction partners and intracellular localization. FEBS Letters, 2021, 595, 2909-2921.	2.8	3
6	Unsupervised Clustering of Missense Variants in HNF1A Using Multidimensional Functional Data Aids Clinical Interpretation. American Journal of Human Genetics, 2020, 107, 670-682.	6.2	25
7	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	2.4	22
8	De novo variants in SUPT16H cause neurodevelopmental disorders associated with corpus callosum abnormalities. Journal of Medical Genetics, 2020, 57, 461-465.	3.2	7
9	Functional Analyses of HNF1A-MODY Variants Refine the Interpretation of Identified Sequence Variants. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e1377-e1386.	3.6	14
10	DominantARL3-related retinitis pigmentosa. Ophthalmic Genetics, 2019, 40, 124-128.	1.2	16
11	De novo truncating variants in PHF21A cause intellectual disability and craniofacial anomalies. European Journal of Human Genetics, 2019, 27, 378-383.	2.8	14
12	NAA10 dysfunction with normal NatA-complex activity in a girl with non-syndromic ID and a de novo NAA10 p.(V111G) variant â€“ a case report. BMC Medical Genetics, 2018, 19, 47.	2.1	24
13	A de novo Ser111Thr variant in aquaporin-4 in a patient with intellectual disability, transient signs of brain ischemia, transient cardiac hypertrophy, and progressive gait disturbance. Journal of Physical Education and Sports Management, 2018, 4, a002303.	1.2	6
14	The E3 SUMO ligase PIASÎ³ is a novel interaction partner regulating the activity of diabetes associated hepatocyte nuclear factor-1Î±. Scientific Reports, 2018, 8, 12780.	3.3	14
15	HUWE1 variants cause dominant X-linked intellectual disability: a clinical study of 21 patients. European Journal of Human Genetics, 2018, 26, 64-74.	2.8	72
16	Postâ€translational modifications of Annexin A2 are linked to its association with perinuclear nonpolysomal mRNP complexes. FEBS Open Bio, 2017, 7, 160-173.	2.3	19
17	In vitro characterization of six STUB1 variants in spinocerebellar ataxia 16 reveals altered structural properties for the encoded CHIP proteins. Bioscience Reports, 2017, 37, .	2.4	27
18	Functional Investigations of HNF1A Identify Rare Variants as Risk Factors for Type 2 Diabetes in the General Population. Diabetes, 2017, 66, 335-346.	0.6	54

#	Article	IF	Citations
19	Phenotypic extremes of BICD2-opathies: from lethal, congenital muscular atrophy with arthrogryposis to asymptomatic with subclinical features. European Journal of Human Genetics, 2017, 25, 1040-1048.	2.8	35
20	Nuclear import of glucokinase in pancreatic beta-cells is mediated by a nuclear localization signal and modulated by SUMOylation. Molecular and Cellular Endocrinology, 2017, 454, 146-157.	3.2	5
21	The intronic <scp>ABCA</scp>4 c.5461â€10T>C variant, frequently seen in patients with Stargardt disease, causes splice defects and reduced <scp>ABCA</scp>4 protein level. Acta Ophthalmologica, 2017, 95, 240-246.	1.1	35
22	High Incidence of HeterozygousABCC8andHNF1AMutations in Czech Patients With Congenital Hyperinsulinism. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E1540-E1549.	3.6	32
23	Association of a Low-Frequency Variant inHNF1AWith Type 2 Diabetes in a Latino Population. JAMA - Journal of the American Medical Association, 2014, 311, 2305.	7.4	230
24	GCK-MODY diabetes as a protein misfolding disease: The mutation R275C promotes protein misfolding, self-association and cellular degradation. Molecular and Cellular Endocrinology, 2014, 382, 55-65.	3.2	15
25	SUMOylation of Pancreatic Glucokinase Regulates Its Cellular Stability and Activity*. Journal of Biological Chemistry, 2013, 288, 5951-5962.	3.4	30