

Ingvild Aukrust

List of Publications by Year in descending order

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Version: 2024-02-01

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	Association of a Low-Frequency Variant in <i>HNF1A</i> With Type 2 Diabetes in a Latino Population. <i>JAMA - Journal of the American Medical Association</i> , 2014, 311, 2305.	7.4	230
2	HUWE1 variants cause dominant X-linked intellectual disability: a clinical study of 21 patients. <i>European Journal of Human Genetics</i> , 2018, 26, 64-74.	2.8	72
3	Functional Investigations of <i>HNF1A</i> Identify Rare Variants as Risk Factors for Type 2 Diabetes in the General Population. <i>Diabetes</i> , 2017, 66, 335-346.	0.6	54
4	Phenotypic extremes of BICD2-opathies: from lethal, congenital muscular atrophy with arthrogyriposis to asymptomatic with subclinical features. <i>European Journal of Human Genetics</i> , 2017, 25, 1040-1048.	2.8	35
5	The intronic <i>ABCA4</i> c.5461T>C variant, frequently seen in patients with Stargardt disease, causes splice defects and reduced <i>ABCA4</i> protein level. <i>Acta Ophthalmologica</i> , 2017, 95, 240-246.	1.1	35
6	High Incidence of Heterozygous <i>ABCC8</i> and <i>HNF1A</i> Mutations in Czech Patients With Congenital Hyperinsulinism. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2015, 100, E1540-E1549.	3.6	32
7	SUMOylation of Pancreatic Glucokinase Regulates Its Cellular Stability and Activity*. <i>Journal of Biological Chemistry</i> , 2013, 288, 5951-5962.	3.4	30
8	<i>In vitro</i> characterization of six <i>STUB1</i> variants in spinocerebellar ataxia 16 reveals altered structural properties for the encoded CHIP proteins. <i>Bioscience Reports</i> , 2017, 37, .	2.4	27
9	Unsupervised Clustering of Missense Variants in <i>HNF1A</i> Using Multidimensional Functional Data Aids Clinical Interpretation. <i>American Journal of Human Genetics</i> , 2020, 107, 670-682.	6.2	25
10	<i>NAA10</i> dysfunction with normal NatA-complex activity in a girl with non-syndromic ID and a de novo <i>NAA10</i> p.(V111G) variant – a case report. <i>BMC Medical Genetics</i> , 2018, 19, 47.	2.1	24
11	Phenotypic spectrum and transcriptomic profile associated with germline variants in <i>TRAF7</i> . <i>Genetics in Medicine</i> , 2020, 22, 1215-1226.	2.4	22
12	Post-translational modifications of Annexin A2 are linked to its association with perinuclear nonpolysomal mRNP complexes. <i>FEBS Open Bio</i> , 2017, 7, 160-173.	2.3	19
13	Dominant <i>ARL3</i> -related retinitis pigmentosa. <i>Ophthalmic Genetics</i> , 2019, 40, 124-128.	1.2	16
14	GCK-MODY diabetes as a protein misfolding disease: The mutation R275C promotes protein misfolding, self-association and cellular degradation. <i>Molecular and Cellular Endocrinology</i> , 2014, 382, 55-65.	3.2	15
15	The E3 SUMO ligase PIAS ³ is a novel interaction partner regulating the activity of diabetes associated hepatocyte nuclear factor-1 α . <i>Scientific Reports</i> , 2018, 8, 12780.	3.3	14
16	De novo truncating variants in <i>PHF21A</i> cause intellectual disability and craniofacial anomalies. <i>European Journal of Human Genetics</i> , 2019, 27, 378-383.	2.8	14
17	Functional Analyses of <i>HNF1A</i> -MODY Variants Refine the Interpretation of Identified Sequence Variants. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, e1377-e1386.	3.6	14
18	Genetic Dominant Variants in <i>STUB1</i> , Segregating in Families with SCA48, Display In Vitro Functional Impairments Indistinctive from Recessive Variants Associated with SCAR16. <i>International Journal of Molecular Sciences</i> , 2021, 22, 5870.	4.1	10

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19	De novo variants in <i>SUPT16H</i> cause neurodevelopmental disorders associated with corpus callosum abnormalities. <i>Journal of Medical Genetics</i> , 2020, 57, 461-465.	3.2	7
20	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. <i>Journal of Physical Education and Sports Management</i> , 2018, 4, a002303.	1.2	6
21	Clinical features and molecular genetics of patients with ABCA4 retinal dystrophies. <i>Acta Ophthalmologica</i> , 2021, 99, e733-e746.	1.1	6
22	Nuclear import of glucokinase in pancreatic beta-cells is mediated by a nuclear localization signal and modulated by SUMOylation. <i>Molecular and Cellular Endocrinology</i> , 2017, 454, 146-157.	3.2	5
23	Structural and biophysical characterization of transcription factor HNF-1A as a tool to study MODY3 diabetes variants. <i>Journal of Biological Chemistry</i> , 2022, 298, 101803.	3.4	4
24	Pellino ² in nonimmune cells: novel interaction partners and intracellular localization. <i>FEBS Letters</i> , 2021, 595, 2909-2921.	2.8	3
25	BRCA1 Norway: comparison of classification for BRCA1 germline variants detected in families with suspected hereditary breast and ovarian cancer between different laboratories. <i>Familial Cancer</i> , 2022, 21, 389-398.	1.9	2