Tuula Rinne

List of Publications by Year
in descending order

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29	2,045	430874	477307
papers	citations	18	29
		h-index	g-index

29	29	29	5268
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Variants of SOS2 are a rare cause of Noonan syndrome with particular predisposition for lymphatic complications. European Journal of Human Genetics, 2021, 29, 51-60.	2.8	17
2	Truncating SRCAP variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature. American Journal of Human Genetics, 2021, 108, 1053-1068.	6.2	31
3	Molecular Inversion Probe-Based Sequencing of USH2A Exons and Splice Sites as a Cost-Effective Screening Tool in USH2 and arRP Cases. International Journal of Molecular Sciences, 2021, 22, 6419.	4.1	8
4	TAB2 deletions and variants cause a highly recognisable syndrome with mitral valve disease, cardiomyopathy, short stature and hypermobility. European Journal of Human Genetics, 2021, 29, 1669-1676.	2.8	19
5	KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. Brain, 2020, 143, 3242-3261.	7.6	57
6	ZMYND11 â€related syndromic intellectual disability: 16 patients delineating and expanding the phenotypic spectrum. Human Mutation, 2020, 41, 1042-1050.	2.5	20
7	Rapid whole exome sequencing in pregnancies to identify the underlying genetic cause in fetuses with congenital anomalies detected by ultrasound imaging. Prenatal Diagnosis, 2020, 40, 972-983.	2.3	49
8	Under-reported aspects of diagnosis and treatment addressed in the Dutch-Flemish guideline for comprehensive diagnostics in disorders/differences of sex development. Journal of Medical Genetics, 2020, 57, 581-589.	3.2	8
9	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	12.8	43
10	De novo variants in FBXO11 cause a syndromic form of intellectual disability with behavioral problems and dysmorphisms. European Journal of Human Genetics, 2019, 27, 738-746.	2.8	32
11	Exome Sequencing of a Primary Ovarian Insufficiency Cohort Reveals Common Molecular Etiologies for a Spectrum of Disease. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 3049-3067.	3.6	53
12	Functional characterisation of a novel class of in-frame insertion variants of KRAS and HRAS. Scientific Reports, 2019, 9, 8239.	3.3	12
13	Prenatal ultrasound findings of rasopathies in a cohort of 424 fetuses: update on genetic testing in the NGS era. Journal of Medical Genetics, 2019, 56, 654-661.	3.2	38
14	Deleterious Variation in BRSK2 Associates with a Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 701-708.	6.2	19
15	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	6.2	30
16	Further delineation of an entity caused by CREBBP and EP300 mutations but not resembling Rubinsteinâ€“Taybi syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 862-876.	1.2	52
17	Loss-of-function mutations in ADCY3 cause monogenic severe obesity. Nature Genetics, 2018, 50, 175-179.	21.4	122
18	De novo mutations in the SET nuclear proto-oncogene, encoding a component of the inhibitor of histone acetyltransferases (INHAT) complex in patients with nonsyndromic intellectual disability. Human Mutation, 2018, 39, 1014-1023.	2.5	18

#	Article	IF	Citations
19	Recessive mutations in ATP8A2 cause severe hypotonia, cognitive impairment, hyperkinetic movement disorders and progressive optic atrophy. Orphanet Journal of Rare Diseases, 2018, 13, 86.	2.7	29
20	Identification of a de novo variant in CHUK in a patient with an EEC/AEC syndromeâ€like phenotype and hypogammaglobulinemia. American Journal of Medical Genetics, Part A, 2017, 173, 1813-1820.	1.2	8
21	Variable phenotypic expression in a large Noonan syndrome family segregating a novel SOS1 mutation. American Journal of Medical Genetics, Part A, 2017, 173, 2968-2972.	1.2	8
22	Detection of clinically relevant copy-number variants by exome sequencing in a large cohort of genetic disorders. Genetics in Medicine, 2017, 19, 667-675.	2.4	143
23	Two cases of RIT1 associated Noonan syndrome: Further delineation of the clinical phenotype and review of the literature. American Journal of Medical Genetics, Part A, 2016, 170, 1874-1880.	1.2	21
24	Meta-analysis of 2,104 trios provides support for 10 new genes for intellectual disability. Nature Neuroscience, 2016, 19, 1194-1196.	14.8	407
25	A Post-Hoc Comparison of the Utility of Sanger Sequencing and Exome Sequencing for the Diagnosis of Heterogeneous Diseases. Human Mutation, 2013, 34, 1721-1726.	2.5	303
26	A novel translation re-initiation mechanism for the p63 gene revealed by amino-terminal truncating mutations in Rapp-Hodgkin/Hay-Wells-like syndromes. Human Molecular Genetics, 2008, 17, 1968-1977.	2.9	53
27	p63-Associated Disorders. Cell Cycle, 2007, 6, 262-268.	2.6	267
28	Delineation of the ADULT syndrome phenotype due to arginine 298 mutations of the p63 gene. European Journal of Human Genetics, 2006, 14, 904-910.	2.8	41
29	Pattern ofp63 mutations and their phenotypesâ€”update. American Journal of Medical Genetics, Part A, 2006, 140A, 1396-1406.	1.2	137