Melissa Lees

List of Publications by Year
in descending order

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37 papers	1,590 citations	331670 21 h-index	345221 36 g-index
42 all docs	42 docs citations	42 times ranked	3985 citing authors

#	Article	IF	Citations
1	Delineating the <scp>Smithâ€Kingsmore</scp> syndrome phenotype: Investigation of 16 patients with the <scp>MTOR</scp> c.<scp>5395G</scp>Â>â€‰A p.(<scp>Glu1799Lys</scp>) missense variant. American Journal of Medical Genetics, Part A, 2021, 185, 2445-2454.	1.2	4
2	Expanding the phenotype of <scp>ASXL3</scp>â€related syndrome: A comprehensive description of 45 unpublished individuals with inherited and de novo pathogenic variants in <scp>ASXL3</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 3446-3458.	1.2	12
3	A clinical scoring system for congenital contractural arachnodactyly. Genetics in Medicine, 2020, 22, 124-131.	2.4	17
4	A restricted spectrum of missense KMT2D variants cause a multiple malformations disorder distinct fromKabuki syndrome. Genetics in Medicine, 2020, 22, 867-877.	2.4	41
5	DNA Methylation Signature for EZH2 Functionally Classifies Sequence Variants in Three PRC2 Complex Genes. American Journal of Human Genetics, 2020, 106, 596-610.	6.2	59
6	De Novo VPS4A Mutations Cause Multisystem Disease with Abnormal Neurodevelopment. American Journal of Human Genetics, 2020, 107, 1129-1148.	6.2	38
7	The CHD8 overgrowth syndrome: A detailed evaluation of an emerging overgrowth phenotype in 27 patients. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 557-564.	1.6	33
8	The phenotype of Sotos syndrome in adulthood: A review of 44 individuals. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 502-508.	1.6	31
9	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	2.3	14
10	A novel NAA10 p.(R83H) variant with impaired acetyltransferase activity identified in two boys with ID and microcephaly. BMC Medical Genetics, 2019, 20, 101.	2.1	21
11	DLK1 Is a Novel Link Between Reproduction and Metabolism. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 2112-2120.	3.6	75
12	Mutation update for the SATB2 gene. Human Mutation, 2019, 40, 1013-1029.	2.5	38
13	The clinical presentation caused by truncating CHD8 variants. Clinical Genetics, 2019, 96, 72-84.	2.0	32
14	De novo and inherited TCF20 pathogenic variants are associated with intellectual disability, dysmorphic features, hypotonia, and neurological impairments with similarities to Smithâ€“Magenis syndrome. Genome Medicine, 2019, 11, 12.	8.2	23
15	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	6.2	27
16	P493â€...Late and atypical presentation of mecp2 mutation. , 2019, , .		0
17	Pathogenicity and selective constraint on variation near splice sites. Genome Research, 2019, 29, 159-170.	5.5	70
18	The contribution of 7q33 copy number variations for intellectual disability. Neurogenetics, 2018, 19, 27-40.	1.4	3

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19	Quantifying the contribution of recessive coding variation to developmental disorders. Science, 2018, 362, 1161-1164.	12.6	158
20	Refining the phenotype associated with GNB1 mutations: Clinical data on 18 newly identified patients and review of the literature. American Journal of Medical Genetics, Part A, 2018, 176, 2259-2275.	1.2	47
21	Further delineation of Malan syndrome. Human Mutation, 2018, 39, 1226-1237.	2.5	42
22	The Tatton-Brown-Rahman Syndrome: A clinical study of 55 individuals with de novo constitutive DNMT3A variants. Wellcome Open Research, 2018, 3, 46.	1.8	75
23	Clinical and molecular consequences of disease-associated de novo mutations in SATB2. Genetics in Medicine, 2017, 19, 900-908.	2.4	46
24	A novel gain-of-function mutation in the ITPR1 suppressor domain causes spinocerebellar ataxia with altered Ca2+ signal patterns. Journal of Neurology, 2017, 264, 1444-1453.	3.6	22
25	Mutations in HYAL2, Encoding Hyaluronidase 2, Cause a Syndrome of Orofacial Clefting and Cor Triatriatum Sinister in Humans and Mice. PLoS Genetics, 2017, 13, e1006470.	3.5	20
26	COLEC10 is mutated in 3MC patients and regulates early craniofacial development. PLoS Genetics, 2017, 13, e1006679.	3.5	62
27	Genetic Analysis of â€˜PAX6-Negativeâ€™ Individuals with Aniridia or Gillespie Syndrome. PLoS ONE, 2016, 11, e0153757.	2.5	54
28	Cover Image, Volume 170A, Number 5, May 2016. , 2016, 170, i-i.		0
29	Cerebroâ€“costoâ€“mandibular syndrome: Clinical, radiological, and genetic findings. American Journal of Medical Genetics, Part A, 2016, 170, 1115-1126.	1.2	21
30	Intra-familial variability associated with recessive RYR1 mutation diagnosed prenatally by exome sequencing. Prenatal Diagnosis, 2016, 36, 1020-1026.	2.3	13
31	NAA10 mutation causing a novel intellectual disability syndrome with Long QT due to N-terminal acetyltransferase impairment. Scientific Reports, 2015, 5, 16022.	3.3	61
32	Clinical and genetic characterisation of infantile liver failure syndrome type 1, due to recessive mutations in LARS. Journal of Inherited Metabolic Disease, 2015, 38, 1085-1092.	3.6	43
33	Discovery of four recessive developmental disorders using probabilistic genotype and phenotype matching among 4,125 families. Nature Genetics, 2015, 47, 1363-1369.	21.4	133
34	Periventricular Calcification, Abnormal Pterins and Dry Thickened Skin: Expanding the Clinical Spectrum of RMND1?. JIMD Reports, 2015, 26, 13-19.	1.5	6
35	A case report of primary ciliary dyskinesia, laterality defects and developmental delay caused by the co-existence of a single gene and chromosome disorder. BMC Medical Genetics, 2015, 16, 45.	2.1	15
36	Friedreich Ataxia in Classical Galactosaemia. JIMD Reports, 2015, 26, 1-5.	1.5	4

#	Article	IF	Citations
37	Mutations in lectin complement pathway genes COLEC11 and MASP1 cause 3MC syndrome. Nature Genetics, 2011, 43, 197-203.	21.4	229