Vardiella Meiner

List of Publications by Year
in descending order

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Version: 2024-02-01

27 papers	420 citations	933447 10 h-index	794594 19 g-index
28 all docs	28 docs citations	28 times ranked	874 citing authors

#	Article	IF	Citations
1	Infantile SOD1 deficiency syndrome caused by a homozygous SOD1 variant with absence of enzyme activity. Brain, 2022, 145, 872-878.	7.6	10
2	Bi-allelic variants in neuronal cell adhesion molecule cause a neurodevelopmental disorder characterized by developmental delay, hypotonia, neuropathy/spasticity. American Journal of Human Genetics, 2022, 109, 518-532.	6.2	8
3	Detection of copy number variants associated with late-onset conditions in ~16â€‰200 pregnancies: parameters for disclosure and pregnancy outcome. Journal of Medical Genetics, 2022, , jmedgenet-2021-107890.	3.2	1
4	Postpartum women's attitudes to disclosure of adultâ€onset conditions in pregnancy. Prenatal Diagnosis, 2022, 42, 1038-1048.	2.3	3
5	Variable phenotype of Knobloch syndrome due to biallelic COL18A1 mutations in children. European Journal of Ophthalmology, 2021, 31, 3349-3354.	1.3	2
6	De novo variants in SIAH1, encoding an E3 ubiquitin ligase, are associated with developmental delay, hypotonia and dysmorphic features. Journal of Medical Genetics, 2021, 58, 205-212.	3.2	6
7	Parental exome analysis identifies shared carrier status for a second recessive disorder in couples with an affected child. European Journal of Human Genetics, 2021, 29, 455-462.	2.8	8
8	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356.	6.2	30
9	Homozygous variant in MADD, encoding a Rab guanine nucleotide exchange factor, results in pleiotropic effects and a multisystemic disorder. European Journal of Human Genetics, 2021, 29, 977-987.	2.8	6
10	Biallelic deletion in a minimal <scp>CAPN15</scp> intron in siblings with a recognizable syndrome of congenital malformations and developmental delay. Clinical Genetics, 2021, 99, 577-582.	2.0	9
11	Toll-like receptor 3 (TLR3) variant and NLRP12 mutation confer susceptibility to a complex clinical presentation. Clinical Immunology, 2020, 212, 108249.	3.2	6
12	Nonâ€confined longâ€standing blood chimerism in a spontaneous monochorionic dizygotic twin pregnancy. International Journal of Gynecology and Obstetrics, 2020, 148, 399-400.	2.3	3
13	Grandparental genotyping enhances exome variant interpretation. American Journal of Medical Genetics, Part A, 2020, 182, 689-696.	1.2	4
14	Smithâ€“Lemliâ€“Opitz syndrome: what is the actual risk for couples carriers of the DHCR7:c.964-1G>C variant?. European Journal of Human Genetics, 2020, 28, 938-942.	2.8	8
15	Progressive Visual Loss Without Retinal Detachment in Stickler Syndrome: An Uncommon and Novel Presentation. TÃ¼rk Oftalmoloji Dergisi, 2020, 50, 387-389.	0.9	0
16	An Ashkenazi Jewish founder mutation in CACNA1F causes retinal phenotype in both hemizygous males and heterozygous female carriers. Ophthalmic Genetics, 2019, 40, 443-448.	1.2	8
17	Homozygous stop-gain variant in LRRC32, encoding a TGFÎ² receptor, associated with cleft palate, proliferative retinopathy, and developmental delay. European Journal of Human Genetics, 2019, 27, 1315-1319.	2.8	7
18	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

#	Article	IF	Citations
19	Homozygous frameshift mutations in FAT1 cause a syndrome characterized by colobomatous-microphthalmia, ptosis, nephropathy and syndactyly. Nature Communications, 2019, 10, 1180.	12.8	27
20	Role of a conserved glutamine in the function of voltage-gated Ca2+ channels revealed by a mutation in human CACNA1D. Journal of Biological Chemistry, 2018, 293, 14444-14454.	3.4	13
21	Stepwise CaSR, AP2S1, and GNA11 sequencing in patients with suspected familial hypocalciuric hypercalcemia. Endocrine, 2017, 55, 741-747.	2.3	26
22	Homozygous null variant in CRADD, encoding an adaptor protein that mediates apoptosis, is associated with lissencephaly. American Journal of Medical Genetics, Part A, 2017, 173, 2539-2544.	1.2	18
23	Ashkenazi carrier screening for reproductive planning: is this what we planned for?. Genetics in Medicine, 2016, 18, 529-529.	2.4	4
24	Postnatal microcephaly and pain insensitivity due to a de novo heterozygous DNM1L mutation causing impaired mitochondrial fission and function. American Journal of Medical Genetics, Part A, 2016, 170, 1603-1607.	1.2	80
25	Deep IntronicGBE1Mutation in Manifesting Heterozygous Patients With Adult Polyglucosan Body Disease. JAMA Neurology, 2015, 72, 441.	9.0	33
26	Frequent misdiagnosis of adult polyglucosan body disease. Journal of Neurology, 2015, 262, 2346-2351.	3.6	29
27	Delineation of C12orf65-related phenotypes: a genotypeâ€“phenotype relationship. European Journal of Human Genetics, 2014, 22, 1019-1025.	2.8	48