Vardiella Meiner

List of Publications by Year
in descending order

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	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
2	Delineation of C12orf65-related phenotypes: a genotypeâ€“phenotype relationship. European Journal of Human Genetics, 2014, 22, 1019-1025.	2.8	48
3	Deep IntronicGBE1Mutation in Manifesting Heterozygous Patients With Adult Polyglucosan Body Disease. JAMA Neurology, 2015, 72, 441.	9.0	33
4	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356.	6.2	30
5	Frequent misdiagnosis of adult polyglucosan body disease. Journal of Neurology, 2015, 262, 2346-2351.	3.6	29
6	Homozygous frameshift mutations in FAT1 cause a syndrome characterized by colobomatous-microphthalmia, ptosis, nephropathy and syndactyly. Nature Communications, 2019, 10, 1180.	12.8	27
7	Stepwise CaSR, AP2S1, and GNA11 sequencing in patients with suspected familial hypocalciuric hypercalcemia. Endocrine, 2017, 55, 741-747.	2.3	26
8	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
9	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
10	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
11	Infantile SOD1 deficiency syndrome caused by a homozygous SOD1 variant with absence of enzyme activity. Brain, 2022, 145, 872-878.	7.6	10
12	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
13	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
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	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
16	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
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	Toll-like receptor 3 (TLR3) variant and NLRP12 mutation confer susceptibility to a complex clinical presentation. Clinical Immunology, 2020, 212, 108249.	3.2	6

#	Article	IF	Citations
19	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
20	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
21	Ashkenazi carrier screening for reproductive planning: is this what we planned for?. Genetics in Medicine, 2016, 18, 529-529.	2.4	4
22	Grandparental genotyping enhances exome variant interpretation. American Journal of Medical Genetics, Part A, 2020, 182, 689-696.	1.2	4
23	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
24	Postpartum women's attitudes to disclosure of adultâ€onset conditions in pregnancy. Prenatal Diagnosis, 2022, 42, 1038-1048.	2.3	3
25	Variable phenotype of Knobloch syndrome due to biallelic COL18A1 mutations in children. European Journal of Ophthalmology, 2021, 31, 3349-3354.	1.3	2
26	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
27	Progressive Visual Loss Without Retinal Detachment in Stickler Syndrome: An Uncommon and Novel Presentation. TÃ¼rk Oftalmoloji Dergisi, 2020, 50, 387-389.	0.9	0