MariÃ«tte Hoffer

List of Publications by Year
in descending order

34 papers	1,299 citations	394421 19 h-index	377865 34 g-index
36 all docs	36 docs citations	36 times ranked	2719 citing authors

#	Article	IF	Citations
1	Prenatal exome sequencing: A useful tool for the fetal neurologist. Clinical Genetics, 2022, 101, 65-77.	2.0	14
2	Hearing loss, cleft palate, and congenital hip dysplasia in female carriers of an intragenic deletion of AMMECR1. American Journal of Medical Genetics, Part A, 2022, , .	1.2	2
3	Noninvasive Prenatal Test Results Indicative of Maternal Malignancies: A Nationwide Genetic and Clinical Follow-Up Study. Journal of Clinical Oncology, 2022, 40, 2426-2435.	1.6	23
4	Biallelic variants in <scp> CENPF </scp> causing a phenotype distinct from StrÃ¸mme syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2022, , .	1.6	3
5	Clinical impact of additional findings detected by genome-wide non-invasive prenatal testing: Follow-up results of the TRIDENT-2 study. American Journal of Human Genetics, 2022, 109, 1140-1152.	6.2	39
6	ANK3 related neurodevelopmental disorders: expanding the spectrum of heterozygous loss-of-function variants. Neurogenetics, 2021, 22, 263-269.	1.4	8
7	Does nonâ€invasive prenatal testing affect the livebirth prevalence of Down syndrome in the Netherlands? A populationâ€based register study. Prenatal Diagnosis, 2021, 41, 1351-1359.	2.3	14
8	Nonâ€invasive prenatal diagnosis for translocation carriersâ€"YES please or NO go?. Acta Obstetricia Et Gynecologica Scandinavica, 2021, 100, 2036-2043.	2.8	1
9	Widening of the genetic and clinical spectrum of Lambâ€"Shaffer syndrome, a neurodevelopmental disorder due to SOX5 haploinsufficiency. Genetics in Medicine, 2020, 22, 524-537.	2.4	21
10	Repurposing of Diagnostic Whole Exome Sequencing Data of 1,583 Individuals for Clinical Pharmacogenetics. Clinical Pharmacology and Therapeutics, 2020, 107, 617-627.	4.7	24
11	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain, 2020, 143, 55-68.	7.6	38
12	Distinct effects on mRNA export factor GANP underlie neurological disease phenotypes and alter gene expression depending on intron content. Human Molecular Genetics, 2020, 29, 1426-1439.	2.9	4
13	The prevalence of genetic diagnoses in fetuses with severe congenital heart defects. Genetics in Medicine, 2020, 22, 1206-1214.	2.4	48
14	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	12.8	105
15	Adultâ€onset betaâ€thalassaemia intermedia caused by a 5â€Mb somatic clonal segmental deletion in haemopoietic stem cells involving the Î²â€globin locus. British Journal of Haematology, 2019, 186, e165-e170.	2.5	3
16	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	12.8	43
17	TRIDENT-2: National Implementation of Genome-wide Non-invasive Prenatal Testing as a First-Tier Screening Test in the Netherlands. American Journal of Human Genetics, 2019, 105, 1091-1101.	6.2	222
18	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

#	Article	IF	Citations
19	From diagnostic yield to clinical impact: a pilot study on the implementation of prenatal exome sequencing in routine care. Genetics in Medicine, 2019, 21, 2303-2310.	2.4	41
20	Putting genome-wide sequencing in neonates into perspective. Genetics in Medicine, 2019, 21, 1074-1082.	2.4	15
21	Origin and clinical relevance of chromosomal aberrations other than the common trisomies detected by genome-wide NIPS: results of the TRIDENT study. Genetics in Medicine, 2018, 20, 480-485.	2.4	85
22	NBEA: Developmental disease gene with early generalized epilepsy phenotypes. Annals of Neurology, 2018, 84, 788-795.	5.3	44
23	Estimates of live birth prevalence of children with <scp>D</scp>own syndrome in the period 1991â€“2015 in the <scp>N</scp>etherlands. Journal of Intellectual Disability Research, 2017, 61, 461-470.	2.0	25
24	Equivalent missense variant in the FOXP2 and FOXP1 transcription factors causes distinct neurodevelopmental disorders. Human Mutation, 2017, 38, 1542-1554.	2.5	28
25	Genotype-phenotype correlation in ATAD3A deletions: not just of scientific relevance. Brain, 2017, 140, e66-e66.	7.6	24
26	CREBBP mutations in individuals without Rubinsteinâ€“Taybi syndrome phenotype. American Journal of Medical Genetics, Part A, 2016, 170, 2681-2693.	1.2	43
27	Mutations in CDC45 , Encoding an Essential Component of the Pre-initiation Complex, Cause Meier-Gorlin Syndrome and Craniosynostosis. American Journal of Human Genetics, 2016, 99, 125-138.	6.2	92
28	Copy number variation analysis in adults with catatonia confirms haploinsufficiency of SHANK3 as a predisposing factor. European Journal of Medical Genetics, 2016, 59, 436-443.	1.3	20
29	Chromosomal abnormalities and copy number variations in fetal leftâ€sided congenital heart defects. Prenatal Diagnosis, 2016, 36, 177-185.	2.3	12
30	Clinical and molecular characterization of an infant with a tandem duplication and deletion of 19p13. American Journal of Medical Genetics, Part A, 2015, 167, 1884-1889.	1.2	4
31	Next-Generation Diagnostics: Gene Panel, Exome, or Whole Genome?. Human Mutation, 2015, 36, 648-655.	2.5	124
32	Copy number variants in patients with short stature. European Journal of Human Genetics, 2014, 22, 602-609.	2.8	60
33	Successful Noninvasive Trisomy 18 Detection Using Single Molecule Sequencing. Clinical Chemistry, 2013, 59, 705-709.	3.2	11
34	A 6Mb deletion in band 2q22 due to a complex chromosome rearrangement associated with severe psychomotor retardation, microcephaly and distinctive dysmorphic facial features. European Journal of Medical Genetics, 2007, 50, 149-154.	1.3	19