

MariÃ«tte Hoffer

List of Publications by Year in descending order

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Version: 2024-02-01

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	TRIDENT-2: National Implementation of Genome-wide Non-invasive Prenatal Testing as a First-Tier Screening Test in the Netherlands. <i>American Journal of Human Genetics</i> , 2019, 105, 1091-1101.	6.2	222
2	Next-Generation Diagnostics: Gene Panel, Exome, or Whole Genome?. <i>Human Mutation</i> , 2015, 36, 648-655.	2.5	124
3	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. <i>Nature Communications</i> , 2020, 11, 4932.	12.8	105
4	Mutations in CDC45 , Encoding an Essential Component of the Pre-initiation Complex, Cause Meier-Gorlin Syndrome and Craniosynostosis. <i>American Journal of Human Genetics</i> , 2016, 99, 125-138.	6.2	92
5	Origin and clinical relevance of chromosomal aberrations other than the common trisomies detected by genome-wide NIPS: results of the TRIDENT study. <i>Genetics in Medicine</i> , 2018, 20, 480-485.	2.4	85
6	Copy number variants in patients with short stature. <i>European Journal of Human Genetics</i> , 2014, 22, 602-609.	2.8	60
7	The prevalence of genetic diagnoses in fetuses with severe congenital heart defects. <i>Genetics in Medicine</i> , 2020, 22, 1206-1214.	2.4	48
8	<i>NBEA</i>: Developmental disease gene with early generalized epilepsy phenotypes. <i>Annals of Neurology</i> , 2018, 84, 788-795.	5.3	44
9	<i>CREBBP</i> mutations in individuals without Rubinsteinâ€“Taybi syndrome phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2681-2693.	1.2	43
10	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. <i>Nature Communications</i> , 2019, 10, 4679.	12.8	43
11	From diagnostic yield to clinical impact: a pilot study on the implementation of prenatal exome sequencing in routine care. <i>Genetics in Medicine</i> , 2019, 21, 2303-2310.	2.4	41
12	Clinical impact of additional findings detected by genome-wide non-invasive prenatal testing: Follow-up results of the TRIDENT-2 study. <i>American Journal of Human Genetics</i> , 2022, 109, 1140-1152.	6.2	39
13	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. <i>Brain</i> , 2020, 143, 55-68.	7.6	38
14	The <i>CHD8</i> overgrowth syndrome: A detailed evaluation of an emerging overgrowth phenotype in 27 patients. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2019, 181, 557-564.	1.6	33
15	Equivalent missense variant in the <i>FOXP2</i> and <i>FOXP1</i> transcription factors causes distinct neurodevelopmental disorders. <i>Human Mutation</i> , 2017, 38, 1542-1554.	2.5	28
16	Estimates of live birth prevalence of children with <sc>D</sc>own syndrome in the period 1991â€“2015 in the <sc>N</sc>etherlands. <i>Journal of Intellectual Disability Research</i> , 2017, 61, 461-470.	2.0	25
17	Genotype-phenotype correlation in ATAD3A deletions: not just of scientific relevance. <i>Brain</i> , 2017, 140, e66-e66.	7.6	24
18	Repurposing of Diagnostic Whole Exome Sequencing Data of 1,583 Individuals for Clinical Pharmacogenetics. <i>Clinical Pharmacology and Therapeutics</i> , 2020, 107, 617-627.	4.7	24

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19	Noninvasive Prenatal Test Results Indicative of Maternal Malignancies: A Nationwide Genetic and Clinical Follow-Up Study. <i>Journal of Clinical Oncology</i> , 2022, 40, 2426-2435.	1.6	23
20	Widening of the genetic and clinical spectrum of Lambdâ€“Shaffer syndrome, a neurodevelopmental disorder due to SOX5 haploinsufficiency. <i>Genetics in Medicine</i> , 2020, 22, 524-537.	2.4	21
21	Copy number variation analysis in adults with catatonia confirms haploinsufficiency of SHANK3 as a predisposing factor. <i>European Journal of Medical Genetics</i> , 2016, 59, 436-443.	1.3	20
22	A 6Mb deletion in band 2q22 due to a complex chromosome rearrangement associated with severe psychomotor retardation, microcephaly and distinctive dysmorphic facial features. <i>European Journal of Medical Genetics</i> , 2007, 50, 149-154.	1.3	19
23	Putting genome-wide sequencing in neonates into perspective. <i>Genetics in Medicine</i> , 2019, 21, 1074-1082.	2.4	15
24	Does nonâ€“invasive prenatal testing affect the livebirth prevalence of Down syndrome in the Netherlands? A populationâ€“based register study. <i>Prenatal Diagnosis</i> , 2021, 41, 1351-1359.	2.3	14
25	Prenatal exome sequencing: A useful tool for the fetal neurologist. <i>Clinical Genetics</i> , 2022, 101, 65-77.	2.0	14
26	Chromosomal abnormalities and copy number variations in fetal leftâ€“sided congenital heart defects. <i>Prenatal Diagnosis</i> , 2016, 36, 177-185.	2.3	12
27	Successful Noninvasive Trisomy 18 Detection Using Single Molecule Sequencing. <i>Clinical Chemistry</i> , 2013, 59, 705-709.	3.2	11
28	ANK3 related neurodevelopmental disorders: expanding the spectrum of heterozygous loss-of-function variants. <i>Neurogenetics</i> , 2021, 22, 263-269.	1.4	8
29	Clinical and molecular characterization of an infant with a tandem duplication and deletion of 19p13. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1884-1889.	1.2	4
30	Distinct effects on mRNA export factor GANP underlie neurological disease phenotypes and alter gene expression depending on intron content. <i>Human Molecular Genetics</i> , 2020, 29, 1426-1439.	2.9	4
31	Adultâ€“onset betaâ€“thalassaemia intermedia caused by a 5â€“Mb somatic clonal segmental deletion in haemopoietic stem cells involving the Î²â€“globin locus. <i>British Journal of Haematology</i> , 2019, 186, e165-e170.	2.5	3
32	Biallelic variants in <sc> <i>CENPF</i> </sc> causing a phenotype distinct from StrÃ„mme syndrome. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2022, , .	1.6	3
33	Hearing loss, cleft palate, and congenital hip dysplasia in female carriers of an intragenic deletion of AMMECR1. <i>American Journal of Medical Genetics, Part A</i> , 2022, , .	1.2	2
34	Nonâ€“invasive prenatal diagnosis for translocation carriersâ€“YES please or NO go?. <i>Acta Obstetrica Et Gynecologica Scandinavica</i> , 2021, 100, 2036-2043.	2.8	1