

Inge B Mathijssen

List of Publications by Year in descending order

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Version: 2024-02-01

18
papers

487
citations

759233

12
h-index

839539

18
g-index

18
all docs

18
docs citations

18
times ranked

1150
citing authors

#	ARTICLE	IF	CITATIONS
1	How will new genetic technologies, such as gene editing, change reproductive decision-making? Views of high-risk couples. <i>European Journal of Human Genetics</i> , 2021, 29, 39-50.	2.8	4
2	TMEM218 dysfunction causes ciliopathies, including Joubert and Meckel syndromes. <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100016.	1.7	7
3	Germline variants in HEY2 functional domains lead to congenital heart defects and thoracic aortic aneurysms. <i>Genetics in Medicine</i> , 2021, 23, 103-110.	2.4	7
4	Couples' experiences with expanded carrier screening: evaluation of a university hospital screening offer. <i>European Journal of Human Genetics</i> , 2021, 29, 1252-1258.	2.8	13
5	Homozygous <i>DMRT2</i> variant associates with severe rib malformations in a newborn. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1216-1221.	1.2	16
6	Preconception carrier screening for multiple disorders: evaluation of a screening offer in a Dutch founder population. <i>European Journal of Human Genetics</i> , 2018, 26, 166-175.	2.8	18
7	Biallelic loss of function variants in <i>COASY</i> cause prenatal onset pontocerebellar hypoplasia, microcephaly, and arthrogryposis. <i>European Journal of Human Genetics</i> , 2018, 26, 1752-1758.	2.8	32
8	Further delineation of Malan syndrome. <i>Human Mutation</i> , 2018, 39, 1226-1237.	2.5	42
9	The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families. <i>Genetics in Medicine</i> , 2017, 19, 45-52.	2.4	94
10	With expanded carrier screening, founder populations run the risk of being overlooked. <i>Journal of Community Genetics</i> , 2017, 8, 327-333.	1.2	6
11	LONG-TERM FOLLOW-UP OF PATIENTS WITH RETINITIS PIGMENTOSA TYPE 12 CAUSED BY <i>CRB1</i> MUTATIONS. <i>Retina</i> , 2017, 37, 161-172.	1.7	30
12	Factors for successful implementation of population-based expanded carrier screening: learning from existing initiatives: Table 1. <i>European Journal of Public Health</i> , 2016, 27, ckw110.	0.3	31
13	Phenotype and genotype in 103 patients with tricho-rhino-phalangeal syndrome. <i>European Journal of Medical Genetics</i> , 2015, 58, 279-292.	1.3	62
14	Targeted carrier screening for four recessive disorders: High detection rate within a founder population. <i>European Journal of Medical Genetics</i> , 2015, 58, 123-128.	1.3	31
15	Noninvasive prenatal diagnosis of Huntington disease: detection of the paternally inherited expanded CAG repeat in maternal plasma. <i>Prenatal Diagnosis</i> , 2015, 35, 945-949.	2.3	23
16	Identification of a Dutch founder mutation in <i>MUSK</i> causing fetal akinesia deformation sequence. <i>European Journal of Human Genetics</i> , 2015, 23, 1151-1157.	2.8	42
17	Testicular cancer in a patient with Primrose syndrome. <i>European Journal of Medical Genetics</i> , 2006, 49, 127-133.	1.3	19
18	Array comparative genomic hybridization analysis of a familial duplication of chromosome 13q: A recognizable syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2005, 136A, 76-80.	1.2	10