Inge B Mathijssen

List of Publications by Year
in descending order

18	487	759233 12	839539 18
papers	citations	h-index	g-index

18	18	18	1150
all docs	docs citations	times ranked	citing authors

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