Nicole Corsten-Janssen

List of Publications by Year
in descending order

13	617	1040056 9	1199594 12
papers	citations	h-index	g-index

13	13	13	1703
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	De novo DHDDS variants cause a neurodevelopmental and neurodegenerative disorder with myoclonus. Brain, 2022, 145, 208-223.	7.6	15
2	Parental experiences of rapid exome sequencing in cases with major ultrasound anomalies during pregnancy. Prenatal Diagnosis, 2022, 42, 762-774.	2.3	17
3	A prospective study on rapid exome sequencing as a diagnostic test for multiple congenital anomalies on fetal ultrasound. Prenatal Diagnosis, 2020, 40, 1300-1309.	2.3	36
4	Rapid whole exome sequencing in pregnancies to identify the underlying genetic cause in fetuses with congenital anomalies detected by ultrasound imaging. Prenatal Diagnosis, 2020, 40, 972-983.	2.3	49
5	Maternal occupational exposure and congenital heart defects in offspring. Scandinavian Journal of Work, Environment and Health, 2020, 46, 599-608.	3.4	4
6	Cohesin complex-associated holoprosencephaly. Brain, 2019, 142, 2631-2643.	7.6	43
7	Variable cardiovascular phenotypes associated with SMAD2 pathogenic variants. Human Mutation, 2018, 39, 1875-1884.	2.5	23
8	Clinical and molecular effects of CHD7 in the heart. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 487-495.	1.6	16
9	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
10	Congenital arch vessel anomalies in CHARGE syndrome: A frequent feature with risk for co-morbidity. IJC Heart and Vasculature, 2016, 12, 21-25.	1.1	14
11	CHD7 mutations are not a major cause of atrioventricular septal and conotruncal heart defects. American Journal of Medical Genetics, Part A, 2014, 164, 3003-3009.	1.2	10
12	The Cardiac Phenotype in Patients With a CHD7 Mutation. Circulation: Cardiovascular Genetics, 2013, 6, 248-254.	5.1	53
13	Molecular studies of theCHD7gene: an obligatory diagnostic step in an expanding range of clinical phenotypes. Expert Review of Molecular Diagnostics, 2012, 12, 795-797.	3.1	0