Anne-Marie Bisgaard Pedersen

List of Publications by Year
in descending order

18 papers	628 citations	759233 12 h-index	839539 18 g-index
18 all docs	18 docs citations	18 times ranked	1162 citing authors

#	Article	IF	Citations
1	Diagnosis and management of Cornelia de Lange syndrome: first international consensus statement. Nature Reviews Genetics, 2018, 19, 649-666.	16.3	223
2	Phenotypes and genotypes in individuals with SMC1A variants. American Journal of Medical Genetics, Part A, 2017, 173, 2108-2125.	1.2	69
3	Additional chromosomal abnormalities in patients with a previously detected abnormal karyotype, mental retardation, and dysmorphic features. American Journal of Medical Genetics, Part A, 2006, 140A, 2180-2187.	1.2	54
4	Validating the Rett Syndrome Gross Motor Scale. PLoS ONE, 2016, 11, e0147555.	2.5	51
5	Transmitted cytogenetic abnormalities in patients with mental retardation: Pathogenic or normal variants?. European Journal of Medical Genetics, 2007, 50, 243-255.	1.3	40
6	Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. Human Genetics, 2020, 139, 575-592.	3.8	24
7	Analysis of the Phenotypes in the Rett Networked Database. International Journal of Genomics, 2019, 2019, 1-9.	1.6	23
8	Building the repertoire of measures of walking in Rett syndrome. Disability and Rehabilitation, 2017, 39, 1926-1931.	1.8	20
9	Functional abilities in aging women with Rett syndrome â€“ the Danish cohort. Disability and Rehabilitation, 2017, 39, 911-918.	1.8	19
10	Twins with mental retardation and an interstitial deletion 7q34q36.2 leading to the diagnosis of long QT syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 644-648.	1.2	16
11	Deletion of 11q12.3â€“11q13.1 in a patient with intellectual disability and childhood facial features resembling Cornelia de Lange syndrome. Gene, 2015, 572, 130-134.	2.2	16
12	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	2.4	16
13	Interstitial deletion of the short arm of chromosome 1 (1p13.1p21.1) in a girl with mental retardation, short stature and colobomata. Clinical Dysmorphology, 2007, 16, 109-112.	0.3	13
14	Is it possible to diagnose Rett syndrome before classical symptoms become obvious? Review of 24 Danish cases born between 2003 and 2012. European Journal of Paediatric Neurology, 2015, 19, 679-687.	1.6	11
15	Patterns of sedentary time and ambulatory physical activity in a Danish population of girls and women with Rett syndrome. Disability and Rehabilitation, 2019, 41, 133-141.	1.8	11
16	Mosaic MECP2 variants in males with classical Rett syndrome features, including stereotypical hand movements. Clinical Genetics, 2019, 95, 403-408.	2.0	10
17	Decline in gross motor skills in adult Rett syndrome; results from a Danish longitudinal study. American Journal of Medical Genetics, Part A, 2021, 185, 3683-3693.	1.2	7
18	Oliver McFarlane syndrome: two new cases and a review of the literature. Ophthalmic Genetics, 2021, 42, 464-473.	1.2	5