Egbert J W Redeker

List of Publications by Year
in descending order

7 papers	562 citations	1478505 6 h-index	1720034 7 g-index
7 all docs	7 docs citations	7 times ranked	1261 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	Genetic heterogeneity in Cornelia de Lange syndrome (CdLS) and CdLS-like phenotypes with observed and predicted levels of mosaicism. Journal of Medical Genetics, 2014, 51, 659-668.	3.2	141
3	Phenotypes and genotypes in individuals with SMC1A variants. American Journal of Medical Genetics, Part A, 2017, 173, 2108-2125.	1.2	69
4	Whole-exome sequencing is a powerful approach for establishing the etiological diagnosis in patients with intellectual disability and microcephaly. BMC Medical Genomics, 2015, 9, 7.	1.5	65
5	Multiplex ligation-dependent probe amplification (MLPA) enhances the molecular diagnosis of aniridia and related disorders. Molecular Vision, 2008, 14, 836-40.	1.1	43
6	Uptake of prenatal diagnostic testing for retinoblastoma compared to other hereditary cancer syndromes in the Netherlands. Familial Cancer, 2017, 16, 271-277.	1.9	16
7	Framing the potential of public frameshift peptides as immunotherapy targets in colon cancer. PLoS ONE, 2021, 16, e0251630.	2.5	5