Nanna Dahl Rendtorff

List of Publications by Year
in descending order

16	339	933447 10	888059 17
papers	citations	h-index	g-index

17	17	17	856
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Identification of p.A684V missense mutation in the WFS1 gene as a frequent cause of autosomal dominant optic atrophy and hearing impairment. American Journal of Medical Genetics, Part A, 2011, 155, 1298-1313.	1.2	95
2	Functional validation of ABHD12 mutations in the neurodegenerative disease PHARC. Neurobiology of Disease, 2017, 98, 36-51.	4.4	31
3	A commonSLC26A4-linked haplotype underlying non-syndromic hearing loss with enlargement of the vestibular aqueduct. Journal of Medical Genetics, 2017, 54, 665-673.	3.2	29
4	The CAPOS mutation in ATP1A3 alters Na/K-ATPase function and results in auditory neuropathy which has implications for management. Human Genetics, 2018, 137, 111-127.	3.8	24
5	Association of SLC26A4 mutations, morphology, and hearing in pendred syndrome and NSEVA. Laryngoscope, 2019, 129, 2574-2579.	2.0	24
6	Putative digenic inheritance of heterozygousRP1L1andC2orf71null mutations in syndromic retinal dystrophy. Ophthalmic Genetics, 2017, 38, 127-132.	1.2	22
7	An Expanded Multi-Organ Disease Phenotype Associated with Mutations in YARS. Genes, 2017, 8, 381.	2.4	19
8	A Novel Locus Harbouring a Functional CD164 Nonsense Mutation Identified in a Large Danish Family with Nonsyndromic Hearing Impairment. PLoS Genetics, 2015, 11, e1005386.	3.5	18
9	Phenotypic subregions within the split-hand/foot malformation 1 locus. Human Genetics, 2016, 135, 345-357.	3.8	15
10	Oral therapy for riboflavin transporter deficiency - What is the regimen of choice?. Parkinsonism and Related Disorders, 2019, 61, 245-247.	2.2	12
11	Phenotypic variability in a seven-generation Swedish family segregating autosomal dominant hearing impairment due to a novel EYA4 frameshift mutation. Gene, 2015, 563, 10-16.	2.2	11
12	Long-Read Sequencing to Unravel Complex Structural Variants of CEP78 Leading to Cone-Rod Dystrophy and Hearing Loss. Frontiers in Cell and Developmental Biology, 2021, 9, 664317.	3.7	11
13	Novel HARS2 missense variants identified in individuals with sensorineural hearing impairment and Perrault syndrome. European Journal of Medical Genetics, 2020, 63, 103733.	1.3	9
14	Partial USH2A deletions contribute to Usher syndrome in Denmark. European Journal of Human Genetics, 2015, 23, 1646-1651.	2.8	8
15	A pathogenic deletion in Forkhead Box L1 (FOXL1) identifies the first otosclerosis (OTSC) gene. Human Genetics, 2022, 141, 965-979.	3.8	7
16	Clinical manifestations and novel pathogenic variants in SOX10 in eight Danish probands with Waardenburg syndrome. European Journal of Medical Genetics, 2021, 64, 104265.	1.3	2