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	A pathogenic deletion in Forkhead Box L1 (FOXL1) identifies the first otosclerosis (OTSC) gene. Human Genetics, 2022, 141, 965-979.	3.8	7
2	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
3	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
4	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
5	Association of SLC26A4 mutations, morphology, and hearing in pendred syndrome and NSEVA. Laryngoscope, 2019, 129, 2574-2579.	2.0	24
6	Oral therapy for riboflavin transporter deficiency - What is the regimen of choice?. Parkinsonism and Related Disorders, 2019, 61, 245-247.	2.2	12
7	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
8	Putative digenic inheritance of heterozygousRP1L1andC2orf71null mutations in syndromic retinal dystrophy. Ophthalmic Genetics, 2017, 38, 127-132.	1.2	22
9	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
10	Functional validation of ABHD12 mutations in the neurodegenerative disease PHARC. Neurobiology of Disease, 2017, 98, 36-51.	4.4	31
11	An Expanded Multi-Organ Disease Phenotype Associated with Mutations in YARS. Genes, 2017, 8, 381.	2.4	19
12	Phenotypic subregions within the split-hand/foot malformation 1 locus. Human Genetics, 2016, 135, 345-357.	3.8	15
13	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
14	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
15	Partial USH2A deletions contribute to Usher syndrome in Denmark. European Journal of Human Genetics, 2015, 23, 1646-1651.	2.8	8
16	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