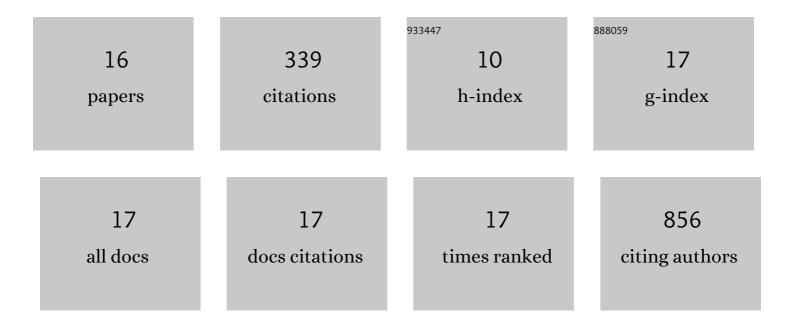
## Nanna Dahl Rendtorff

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/1221193/publications.pdf Version: 2024-02-01



#	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 <i>SLC26A4</i> 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 heterozygous <i>RP1L1</i> and <i>C2orf71</i> null mutations in syndromic retinal dystrophy. Ophthalmic Genetics, 2017, 38, 127-132.	1.2	22
9	A common <i>SLC26A4</i> -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 <i>WFS1</i> 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