Jaap Oostrik

List of Publications by Year
in descending order

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Version: 2024-02-01

16	386	840776 11	940533 16
papers	citations	h-index	g-index

17	17	17	886
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Exploring the missing heritability in subjects with hearing loss, enlarged vestibular aqueducts, and a single or no pathogenic SLC26A4 variant. Human Genetics, 2022, 141, 465-484.	3.8	3
2	Usher syndrome type IV: clinically and molecularly confirmed by novel ARSG variants. Human Genetics, 2022, 141, 1723-1738.	3.8	16
3	Cochlear supporting cells require GAS2 for cytoskeletal architecture and hearing. Developmental Cell, 2021, 56, 1526-1540.e7.	7.0	18
4	Molecular Inversion Probe-Based Sequencing of USH2A Exons and Splice Sites as a Cost-Effective Screening Tool in USH2 and arRP Cases. International Journal of Molecular Sciences, 2021, 22, 6419.	4.1	8
5	AON-based degradation of c.151C>T mutant COCH transcripts associated with dominantly inherited hearing impairment DFNA9. Molecular Therapy - Nucleic Acids, 2021, 24, 274-283.	5.1	9
6	De novo and inherited loss-of-function variants of ATP2B2 are associated with rapidly progressive hearing impairment. Human Genetics, 2019, 138, 61-72.	3.8	27
7	Heterozygous missense variants of LMX1A lead to nonsyndromic hearing impairment and vestibular dysfunction. Human Genetics, 2018, 137, 389-400.	3.8	32
8	MPZL2, Encoding the Epithelial Junctional Protein Myelin Protein Zero-like 2, Is Essential for Hearing in Man and Mouse. American Journal of Human Genetics, 2018, 103, 74-88.	6.2	34
9	A homozygousFITM2mutation causes a deafness-dystonia syndrome with motor regression and signs of ichthyosis and sensory neuropathy. DMM Disease Models and Mechanisms, 2017, 10, 105-118.	2.4	16
10	Broadening the phenotype of DFNB28: Mutations in TRIOBP are associated with moderate, stable hereditary hearing impairment. Hearing Research, 2017, 347, 56-62.	2.0	17
11	Novel and recurrent CIB2 variants, associated with nonsyndromic deafness, do not affect calcium buffering and localization in hair cells. European Journal of Human Genetics, 2016, 24, 542-549.	2.8	28
12	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
13	Allelic Mutations of KITLG, Encoding KIT Ligand, Cause Asymmetric and Unilateral Hearing Loss and Waardenburg Syndrome Type 2. American Journal of Human Genetics, 2015, 97, 647-660.	6.2	55
14	Progressive hearing loss and vestibular dysfunction caused by a homozygous nonsense mutation in CLIC5. European Journal of Human Genetics, 2015, 23, 189-194.	2.8	49
15	Genetic Spectrum of Autosomal Recessive Non-Syndromic Hearing Loss in Pakistani Families. PLoS ONE, 2014, 9, e100146.	2.5	52
16	A canonical splice site mutation in GIPC3 causes sensorineural hearing loss in a large Pakistani family. Journal of Human Genetics, 2014, 59, 683-686.	2.3	4