Jaap Oostrik

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/9786918/publications.pdf

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	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
2	Genetic Spectrum of Autosomal Recessive Non-Syndromic Hearing Loss in Pakistani Families. PLoS ONE, 2014, 9, e100146.	2.5	52
3	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
4	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
5	Heterozygous missense variants of LMX1A lead to nonsyndromic hearing impairment and vestibular dysfunction. Human Genetics, 2018, 137, 389-400.	3.8	32
6	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
7	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
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	Cochlear supporting cells require GAS2 for cytoskeletal architecture and hearing. Developmental Cell, 2021, 56, 1526-1540.e7.	7.0	18
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	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
12	Usher syndrome type IV: clinically and molecularly confirmed by novel ARSG variants. Human Genetics, 2022, 141, 1723-1738.	3.8	16
13	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
14	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
15	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
16	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