

Seungmin Lee

List of Publications by Year in descending order

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14
papers

251
citations

1040056

9
h-index

1058476

14
g-index

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all docs

14
docs citations

14
times ranked

359
citing authors

#	ARTICLE	IF	CITATIONS
1	A nonsense <i>TMEM43</i> variant leads to disruption of connexin-linked function and autosomal dominant auditory neuropathy spectrum disorder. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021, 118, .	7.1	17
2	Powerful use of automated prioritization of candidate variants in genetic hearing loss with extreme etiologic heterogeneity. <i>Scientific Reports</i> , 2021, 11, 19476.	3.3	2
3	Significant Mendelian genetic contribution to pediatric mild-to-moderate hearing loss and its comprehensive diagnostic approach. <i>Genetics in Medicine</i> , 2020, 22, 1119-1128.	2.4	34
4	<i>POLD1</i> variants leading to reduced polymerase activity can cause hearing loss without syndromic features. <i>Human Mutation</i> , 2020, 41, 913-920.	2.5	7
5	The molecular etiology of deafness and auditory performance in the postlingually deafened cochlear implantees. <i>Scientific Reports</i> , 2020, 10, 5768.	3.3	17
6	Severe or Profound Sensorineural Hearing Loss Caused by Novel <i>USH2A</i> Variants in Korea: Potential Genotype-Phenotype Correlation. <i>Clinical and Experimental Otorhinolaryngology</i> , 2020, 13, 113-122.	2.1	32
7	Identification of a Potential Founder Effect of a Novel <i>PDZD7</i> Variant Involved in Moderate-to-Severe Sensorineural Hearing Loss in Koreans. <i>International Journal of Molecular Sciences</i> , 2019, 20, 4174.	4.1	16
8	Identification of a Novel Frameshift Variant of <i>POU3F4</i> and Genetic Counseling of Korean Incomplete Partition Type III Subjects Based on Detailed Genotypes. <i>Genetic Testing and Molecular Biomarkers</i> , 2019, 23, 423-427.	0.7	6
9	Clarification of glycosylphosphatidylinositol anchorage of <i>OTOANCORIN</i> and human <i>OTOA</i> variants associated with deafness. <i>Human Mutation</i> , 2019, 40, 525-531.	2.5	8
10	Differential disruption of autoinhibition and defect in assembly of cytoskeleton during cell division decide the fate of human <i>DIAPH1</i> -related cytoskeletopathy. <i>Journal of Medical Genetics</i> , 2019, 56, 818-827.	3.2	11
11	A clinical guidance to <i>DFNA22</i> drawn from a Korean cohort study with an autosomal dominant deaf population: A retrospective cohort study. <i>Journal of Gene Medicine</i> , 2018, 20, e3019.	2.8	7
12	One-step noninvasive prenatal testing (NIPT) for autosomal recessive homozygous point mutations using digital PCR. <i>Scientific Reports</i> , 2018, 8, 2877.	3.3	25
13	Mutational and phenotypic spectrum of <i>OTOF</i> -related auditory neuropathy in Koreans: eliciting reciprocal interaction between bench and clinics. <i>Journal of Translational Medicine</i> , 2018, 16, 330.	4.4	19
14	<i>ATP1A3</i> mutations can cause progressive auditory neuropathy: a new gene of auditory synaptopathy. <i>Scientific Reports</i> , 2017, 7, 16504.	3.3	50