

Xiaohua Jin

List of Publications by Year in descending order

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7
papers

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1478505

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1720034

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#	ARTICLE	IF	CITATIONS
1	Identification of novel deep intronic <i>PAH</i> gene variants in patients diagnosed with phenylketonuria. <i>Human Mutation</i> , 2022, 43, 56-66.	2.5	10
2	Novel CRISPR/Cas12a-based genetic diagnostic approach for SLC26A4 mutation-related hereditary hearing loss. <i>European Journal of Medical Genetics</i> , 2022, 65, 104406.	1.3	3
3	Variant analysis of 92 Chinese Han families with hearing loss. <i>BMC Medical Genomics</i> , 2022, 15, 12.	1.5	6
4	Engineering of near-PAMless adenine base editor with enhanced editing activity and reduced off-target. <i>Molecular Therapy - Nucleic Acids</i> , 2022, 28, 732-742.	5.1	8
5	Mutation analysis of <i>TCOF1</i> gene in Chinese Treacher Collins syndrome patients. <i>Journal of Clinical Laboratory Analysis</i> , 2021, 35, e23567.	2.1	10
6	Compound heterozygous variants of the <i>FBXO7</i> gene resulting in infantile-onset Parkinsonian-pyramidal syndrome in siblings of a Chinese family. <i>Journal of Clinical Laboratory Analysis</i> , 2020, 34, e23324.	2.1	8
7	Mutation spectrum of PAH gene in phenylketonuria patients in Northwest China: identification of twenty novel variants. <i>Metabolic Brain Disease</i> , 2019, 34, 733-745.	2.9	14