

Next-generation sequencing reveals the mutational landscape of
Usher syndrome: copy number variations, phenocopies,
translational read-through, and *PEX26* mutated in

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Citation Report

#	ARTICLE	IF	CITATIONS
1	Next-generation sequencing reveals the mutational landscape of clinically diagnosed Usher syndrome: copy number variations, phenocopies, a predominant target for translational read-through, and <i>PEX26</i> mutated in Heimler syndrome. <i>Molecular Genetics & Genomic Medicine</i> , 2017, 5, 531-552.	0.6	55
3	High-throughput sequencing for the molecular diagnosis of Usher syndrome reveals 42 novel mutations and consolidates CEP250 as Usher-like disease causative. <i>Scientific Reports</i> , 2018, 8, 17113.	1.6	30
4	A Natural Occurring Mouse Model with <i>Adgrv1</i> Mutation of Usher Syndrome 2C and Characterization of its Recombinant Inbred Strains. <i>Cellular Physiology and Biochemistry</i> , 2018, 47, 1883-1897.	1.1	22
6	A longitudinal study of retinopathy in the <i>PEX1-Gly844Asp</i> mouse model for mild Zellweger Spectrum Disorder. <i>Experimental Eye Research</i> , 2019, 186, 107713.	1.2	19
7	Prospects and modalities for the treatment of genetic ocular anomalies. <i>Human Genetics</i> , 2019, 138, 1019-1026.	1.8	7
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9	Ataluren for the Treatment of Usher Syndrome 2A Caused by Nonsense Mutations. <i>International Journal of Molecular Sciences</i> , 2019, 20, 6274.	1.8	30
10	Expanding the clinical and genetic spectrum of Heimler syndrome. <i>Orphanet Journal of Rare Diseases</i> , 2019, 14, 290.	1.2	19
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18	Atypical and ultra-rare Usher syndrome: a review. <i>Ophthalmic Genetics</i> , 2020, 41, 401-412.	0.5	20
19	Translational readthrough inducing drugs for the treatment of inherited retinal dystrophies. <i>Expert Review of Ophthalmology</i> , 2020, 15, 169-182.	0.3	3
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22	Clinical and preclinical therapeutic outcome metrics for USH2A-related disease. <i>Human Molecular Genetics</i> , 2020, 29, 1882-1899.	1.4	24
23	Application of targeted panel sequencing and whole exome sequencing for 76 Chinese families with retinitis pigmentosa. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1131.	0.6	27
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31	AAV-mediated PEX1 gene augmentation improves visual function in the PEX1-Gly844Asp mouse model for mild Zellweger spectrum disorder. <i>Molecular Therapy - Methods and Clinical Development</i> , 2021, 23, 225-240.	1.8	9
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43	Caracterizaci3n fenot3pica de la retinitis pigmentaria asociada a sordera. <i>Biomedica</i> , 2022, 42, 130-143.	0.3	0
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