

Next-generation sequencing reveals the mutational landscape of
Usher syndrome: copy number variations, phenocopies, and
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. Molecular Genetics & Genomic Medicine, 2017, 5, 531-552.	1.2	55
3	High-throughput sequencing for the molecular diagnosis of Usher syndrome reveals 42 novel mutations and consolidates CEP250 as Usher-like disease causative. Scientific Reports, 2018, 8, 17113.	3.3	30
4	A Natural Occurring Mouse Model with Adgrv1 Mutation of Usher Syndrome 2C and Characterization of its Recombinant Inbred Strains. Cellular Physiology and Biochemistry, 2018, 47, 1883-1897.	1.6	22
6	A longitudinal study of retinopathy in the PEX1-Gly844Asp mouse model for mild Zellweger Spectrum Disorder. Experimental Eye Research, 2019, 186, 107713.	2.6	19
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9	Ataluren for the Treatment of Usher Syndrome 2A Caused by Nonsense Mutations. International Journal of Molecular Sciences, 2019, 20, 6274.	4.1	30
10	Expanding the clinical and genetic spectrum of Heimler syndrome. Orphanet Journal of Rare Diseases, 2019, 14, 290.	2.7	19
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21	A Review of Gene, Drug and Cell-Based Therapies for Usher Syndrome. <i>Frontiers in Cellular Neuroscience</i> , 2020, 14, 183.	3.7	18
22	Clinical and preclinical therapeutic outcome metrics for USH2A-related disease. <i>Human Molecular Genetics</i> , 2020, 29, 1882-1899.	2.9	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.	1.2	27
24	The Peroxisomal PTS1-Import Defect of PEX1- Deficient Cells Is Independent of Pexophagy in <i>Saccharomyces cerevisiae</i> . <i>International Journal of Molecular Sciences</i> , 2020, 21, 867.	4.1	6
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37	Heimler Syndrome. <i>Advances in Experimental Medicine and Biology</i> , 2020, 1299, 81-87.	1.6	4
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44	Molecular insights into peroxisome homeostasis and peroxisome biogenesis disorders. Biochimica Et Biophysica Acta - Molecular Cell Research, 2022, 1869, 119330.	4.1	15
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