Erik de Vrieze

List of Publications by Year
in descending order

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40 papers	1,361 citations	361045 20 h-index	360668 35 g-index
45 all docs	45 docs citations	45 times ranked	2256 citing authors

#	Article	IF	Citations
1	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.	1.8	3
2	Genotype-Phenotype Correlations of Pathogenic COCH Variants in DFNA9: A HuGE Systematic Review and Audiometric Meta-Analysis. Biomolecules, 2022, 12, 220.	1.8	5
3	Generation of Humanized Zebrafish Models for the In Vivo Assessment of Antisense Oligonucleotide-Based Splice Modulation Therapies. Methods in Molecular Biology, 2022, 2434, 281-299.	0.4	2
4	Altering gene expression using antisense oligonucleotide therapy for hearing loss. Hearing Research, 2022, 426, 108523.	0.9	5
5	Scrutinizing pathogenicity of the USH2A c.2276â€‰Gâ€‰>â€‰T; p.(Cys759Phe) variant. Npj Genomic Medicine, 2022, 7, .	1.7	5
6	A RIPOR2 in-frame deletion is a frequent and highly penetrant cause of adult-onset hearing loss. Journal of Medical Genetics, 2021, 58, 96-104.	1.5	14
7	Loss of Gap Junction Delta-2 (GJD2) gene orthologs leads to refractive error in zebrafish. Communications Biology, 2021, 4, 676.	2.0	19
8	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.	2.3	9
9	Antisense oligonucleotide-based treatment of retinitis pigmentosa caused by USH2A exon 13 mutations. Molecular Therapy, 2021, 29, 2441-2455.	3.7	75
10	Zebrafish as a Model to Evaluate a CRISPR/Cas9-Based Exon Excision Approach as a Future Treatment Option for EYS-Associated Retinitis Pigmentosa. International Journal of Molecular Sciences, 2021, 22, 9154.	1.8	6
11	Untargeted metabolomics and infrared ion spectroscopy identify biomarkers for pyridoxine-dependent epilepsy. Journal of Clinical Investigation, 2021, 131, .	3.9	33
12	Efficient Generation of Knock-In Zebrafish Models for Inherited Disorders Using CRISPR-Cas9 Ribonucleoprotein Complexes. International Journal of Molecular Sciences, 2021, 22, 9429.	1.8	10
13	A Novel COCH Mutation Affects the vWFA2 Domain and Leads to a Relatively Mild DFNA9 Phenotype. Otology and Neurotology, 2021, 42, e399-e407.	0.7	2
14	Attitudes of Potential Participants Towards Potential Gene Therapy Trials in Autosomal Dominant Progressive Sensorineural Hearing Loss. Otology and Neurotology, 2021, 42, 384-389.	0.7	3
15	Clinical and preclinical therapeutic outcome metrics for USH2A-related disease. Human Molecular Genetics, 2020, 29, 1882-1899.	1.4	24
16	CiliaCarta: An integrated and validated compendium of ciliary genes. PLoS ONE, 2019, 14, e0216705.	1.1	104
17	Whole-exome sequencing reveals POC5 as a novel gene associated with autosomal recessive retinitis pigmentosa. Human Molecular Genetics, 2018, 27, 614-624.	1.4	26
18	Poor Splice-Site Recognition in a Humanized Zebrafish Knockin Model for the Recurrent Deep-Intronic c.7595-2144A>G Mutation inUSH2A. Zebrafish, 2018, 15, 597-609.	0.5	21

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19	Usherin defects lead to early-onset retinal dysfunction in zebrafish. Experimental Eye Research, 2018, 173, 148-159.	1.2	53
20	Homozygous variants in KIAA1549, encoding a ciliary protein, are associated with autosomal recessive retinitis pigmentosa. Journal of Medical Genetics, 2018, 55, 705-712.	1.5	13
21	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.	1.2	16
22	Missense mutations in the WD40 domain ofAHI1cause non-syndromic retinitis pigmentosa. Journal of Medical Genetics, 2017, 54, 624-632.	1.5	21
23	An organelle-specific protein landscape identifies novel diseases and molecular mechanisms. Nature Communications, 2016, 7, 11491.	5.8	207
24	Identification of a homozygous nonsense mutation in KIAA0556 in a consanguineous family displaying Joubert syndrome. Human Genetics, 2016, 135, 919-921.	1.8	18
25	PDZD7 connects the Usher protein complex to the intraflagellar transport machinery. Cilia, 2015, 4, .	1.8	0
26	The Ciliopathy Protein CC2D2A Associates with NINL and Functions in RAB8-MICAL3-Regulated Vesicle Trafficking. PLoS Genetics, 2015, 11, e1005575.	1.5	64
27	KIAA0556 is a novel ciliary basal body component mutated in Joubert syndrome. Genome Biology, 2015, 16, 293.	3.8	56
28	Identification of novel osteogenic compounds by an ex-vivo sp7:luciferase zebrafish scale assay. Bone, 2015, 74, 106-113.	1.4	33
29	NINL and DZANK1 Co-function in Vesicle Transport and Are Essential for Photoreceptor Development in Zebrafish. PLoS Genetics, 2015, 11, e1005574.	1.5	23
30	Prednisolone induces osteoporosis-like phenotype in regenerating zebrafish scales. Osteoporosis International, 2014, 25, 567-578.	1.3	68
31	Knockdown of Monocarboxylate Transporter 8 (mct8) Disturbs Brain Development and Locomotion in Zebrafish. Endocrinology, 2014, 155, 2320-2330.	1.4	39
32	Disruption of the Basal Body Protein POC1B Results in Autosomal-Recessive Cone-Rod Dystrophy. American Journal of Human Genetics, 2014, 95, 131-142.	2.6	65
33	Arachidonic Acid Enhances Turnover of the Dermal Skeleton: Studies on Zebrafish Scales. PLoS ONE, 2014, 9, e89347.	1.1	30
34	Subfunctionalization of POMC paralogues in Senegalese sole (Solea senegalensis). General and Comparative Endocrinology, 2012, 175, 407-415.	0.8	22
35	Evidence for a hydroxyapatite precursor in regenerating cyprinid scales. Journal of Applied Ichthyology, 2012, 28, 388-392.	0.3	11
36	Elasmoid scales of fishes as model in biomedical bone research. Journal of Applied Ichthyology, 2012, 28, 382-387.	0.3	42

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37	Matrix metalloproteinases in osteoclasts of ontogenetic and regenerating zebrafish scales. Bone, 2011, 48, 704-712.	1.4	78
38	Identification and Functional Characterization of Zebrafish Solute Carrier Slc16a2 (Mct8) as a Thyroid Hormone Membrane Transporter. Endocrinology, 2011, 152, 5065-5073.	1.4	65
39	STAT genes display differential evolutionary rates that correlate with their roles in the endocrine and immune system. Journal of Endocrinology, 2011, 209, 175-184.	1.2	37
40	ALP, TRAcP and cathepsin K in elasmoid scales: a role in mineral metabolism?. Journal of Applied Ichthyology, 2010, 26, 210-213.	0.3	25