

# Matthew R Avenarius

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/2163015/publications.pdf>

Version: 2024-02-01

25  
papers

1,489  
citations

516710

16  
h-index

580821

25  
g-index

28  
all docs

28  
docs citations

28  
times ranked

2029  
citing authors

#	ARTICLE	IF	CITATIONS
1	A novel deletion involving the connexin-30 gene, del(GJB6-d13s1854), found in trans with mutations in the GJB2 gene (connexin-26) in subjects with DFNB1 non-syndromic hearing impairment. <i>Journal of Medical Genetics</i> , 2005, 42, 588-594.	3.2	282
2	Human Male Infertility Caused by Mutations in the CATSPER1 Channel Protein. <i>American Journal of Human Genetics</i> , 2009, 84, 505-510.	6.2	206
3	A Forward Genetics Screen in Mice Identifies Recessive Deafness Traits and Reveals That Pejvakin Is Essential for Outer Hair Cell Function. <i>Journal of Neuroscience</i> , 2007, 27, 2163-2175.	3.6	159
4	OTOF mutations revealed by genetic analysis of hearing loss families including a potential temperature sensitive auditory neuropathy allele. <i>Journal of Medical Genetics</i> , 2005, 43, 576-581.	3.2	139
5	Genetic male infertility and mutation of CATSPER ion channels. <i>European Journal of Human Genetics</i> , 2010, 18, 1178-1184.	2.8	139
6	GJB2 mutations: Passage through Iran. <i>American Journal of Medical Genetics, Part A</i> , 2005, 133A, 132-137.	1.2	77
7	Stereocilia-staircase spacing is influenced by myosin III motors and their cargos espin-1 and espin-like. <i>Nature Communications</i> , 2016, 7, 10833.	12.8	72
8	Complete sequencing of the SMN2 gene in SMA patients detects SMN gene deletion junctions and variants in SMN2 that modify the SMA phenotype. <i>Human Genetics</i> , 2019, 138, 241-256.	3.8	57
9	Mutations in Grxcr1 Are The Basis for Inner Ear Dysfunction in the Pirouette Mouse. <i>American Journal of Human Genetics</i> , 2010, 86, 148-160.	6.2	49
10	Heterodimeric capping protein is required for stereocilia length and width regulation. <i>Journal of Cell Biology</i> , 2017, 216, 3861-3881.	5.2	48
11	Evidence for a direct role of the disease modifier SCN11 in splicing. <i>Human Molecular Genetics</i> , 2007, 16, 2506-2516.	2.9	41
12	The Coxsackievirus and Adenovirus Receptor: A new adhesion protein in cochlear development. <i>Hearing Research</i> , 2006, 215, 1-9.	2.0	28
13	Loss of <i>Baiap2l2</i> destabilizes the transducing stereocilia of cochlear hair cells and leads to deafness. <i>Journal of Physiology</i> , 2021, 599, 1173-1198.	2.9	28
14	Correlation of Actin Crosslinker and Capper Expression Levels with Stereocilia Growth Phases. <i>Molecular and Cellular Proteomics</i> , 2014, 13, 606-620.	3.8	26
15	Somatic PIK3R1 variation as a cause of vascular malformations and overgrowth. <i>Genetics in Medicine</i> , 2021, 23, 1882-1888.	2.4	26
16	Activation of the RAS pathway through uncommon BRAF mutations in mucinous pancreatic cysts without KRAS mutation. <i>Modern Pathology</i> , 2021, 34, 438-444.	5.5	19
17	Improved Biolistic Transfection of Hair Cells. <i>PLoS ONE</i> , 2012, 7, e46765.	2.5	14
18	Grxcr2 is required for stereocilia morphogenesis in the cochlea. <i>PLoS ONE</i> , 2018, 13, e0201713.	2.5	11

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19	Two Iranian families with a novel mutation in <i>GJB2</i> causing autosomal dominant nonsyndromic hearing loss. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1202-1211.	1.2	9
20	Genetic Characterization of Pediatric Sarcomas by Targeted RNA Sequencing. <i>Journal of Molecular Diagnostics</i> , 2020, 22, 1238-1245.	2.8	9
21	Genome sequencing identifies somatic BRAF duplication c.1794_1796dupTAC;p.Thr599dup in pediatric patient with low-grade ganglioglioma. <i>Journal of Physical Education and Sports Management</i> , 2018, 4, a002618.	1.2	7
22	TRPV6, TRPM6 and TRPM7 Do Not Contribute to Hair-Cell Mechanotransduction. <i>Frontiers in Cellular Neuroscience</i> , 2018, 12, 41.	3.7	6
23	<i>EGFR</i> internal tandem duplications in fusion-negative congenital and neonatal spindle cell tumors. <i>Genes Chromosomes and Cancer</i> , 2023, 62, 17-26.	2.8	3
24	The clinical utility of a risk-modifying SNP to detect carriers for spinal muscular atrophy with increased sensitivity. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2022, 10, e1897.	1.2	2
25	Normal FISH CLL Represents a Heterogeneous Subgroup Where Prognosis Can be Refined with IGHV Mutational Status. <i>Blood</i> , 2021, 138, 1563-1563.	1.4	0