

# Anne-Francoise Roux

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/9665786/publications.pdf>

Version: 2024-02-01

15  
papers

1,690  
citations

933447

10  
h-index

1058476

14  
g-index

16  
all docs

16  
docs citations

16  
times ranked

4496  
citing authors

#	ARTICLE	IF	CITATIONS
1	Identification of the First Single GSDME Exon 8 Structural Variants Associated with Autosomal Dominant Hearing Loss. <i>Diagnostics</i> , 2022, 12, 207.	2.6	4
2	Natural history of Usher type 2 with the c.2299delG mutation of <i>USH2A</i> in a large cohort. <i>Ophthalmic Genetics</i> , 2022, 43, 470-475.	1.2	3
3	MobiDetails: online DNA variants interpretation. <i>European Journal of Human Genetics</i> , 2021, 29, 356-360.	2.8	34
4	CRB1-Related Retinal Dystrophies in a Cohort of 50 Patients: A Reappraisal in the Light of Specific Müller Cell and Photoreceptor CRB1 Isoforms. <i>International Journal of Molecular Sciences</i> , 2021, 22, 12642.	4.1	11
5	The Study of a 231 French Patient Cohort Significantly Extends the Mutational Spectrum of the Two Major Usher Genes MYO7A and USH2A. <i>International Journal of Molecular Sciences</i> , 2021, 22, 13294.	4.1	5
6	SLC12A2 variants cause a neurodevelopmental disorder or cochleovestibular defect. <i>Brain</i> , 2020, 143, 2380-2387.	7.6	34
7	Mutations in <i>PLS1</i> , encoding fimbrin, cause autosomal dominant nonsyndromic hearing loss. <i>Human Mutation</i> , 2019, 40, 2286-2295.	2.5	23
8	Whole <i>USH2A</i> Gene Sequencing Identifies Several New Deep Intronic Mutations. <i>Human Mutation</i> , 2016, 37, 184-193.	2.5	80
9	HGVS Recommendations for the Description of Sequence Variants: 2016 Update. <i>Human Mutation</i> , 2016, 37, 564-569.	2.5	1,194
10	Relative Frequencies of Inherited Retinal Dystrophies and Optic Neuropathies in Southern France: Assessment of 21-year Data Management. <i>Ophthalmic Epidemiology</i> , 2013, 20, 13-25.	1.7	44
11	The contribution of GPR98 and DFNB31 genes to a Spanish Usher syndrome type 2 cohort. <i>Molecular Vision</i> , 2013, 19, 367-73.	1.1	13
12	Usher syndrome type 2 caused by activation of an USH2A pseudoexon: Implications for diagnosis and therapy. <i>Human Mutation</i> , 2012, 33, 104-108.	2.5	102
13	Non-USH2A mutations in USH2 patients. <i>Human Mutation</i> , 2012, 33, 504-510.	2.5	57
14	Four-Year Follow-up of Diagnostic Service in USH1 Patients. , 2011, 52, 4063.		47
15	Ex vivo splicing assays of mutations at noncanonical positions of splice sites in USHER genes. <i>Human Mutation</i> , 2010, 31, 347-355.	2.5	39