Anne-Francoise Roux

List of Publications by Year
in descending order

15	1,690	933447 10	1058476 14
papers	citations	h-index	g-index

16	16	16	4496
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	HGVS Recommendations for the Description of Sequence Variants: 2016 Update. Human Mutation, 2016, 37, 564-569.	2.5	1,194
2	Usher syndrome type 2 caused by activation of an USH2A pseudoexon: Implications for diagnosis and therapy. Human Mutation, 2012, 33, 104-108.	2.5	102
3	WholeUSH2AGene Sequencing Identifies Several New Deep Intronic Mutations. Human Mutation, 2016, 37, 184-193.	2.5	80
4	Non-USH2A mutations in USH2 patients. Human Mutation, 2012, 33, 504-510.	2.5	57
5	Four-Year Follow-up of Diagnostic Service in USH1 Patients. , 2011, 52, 4063.		47
6	Relative Frequencies of Inherited Retinal Dystrophies and Optic Neuropathies in Southern France: Assessment of 21-year Data Management. Ophthalmic Epidemiology, 2013, 20, 13-25.	1.7	44
7	Ex vivo splicing assays of mutations at noncanonical positions of splice sites in USHER genes. Human Mutation, 2010, 31, 347-355.	2.5	39
8	SLC12A2 variants cause a neurodevelopmental disorder or cochleovestibular defect. Brain, 2020, 143, 2380-2387.	7.6	34
9	MobiDetails: online DNA variants interpretation. European Journal of Human Genetics, 2021, 29, 356-360.	2.8	34
10	Mutations in PLS1 , encoding fimbrin, cause autosomal dominant nonsyndromic hearing loss. Human Mutation, 2019, 40, 2286-2295.	2.5	23
11	The contribution of GPR98 and DFNB31 genes to a Spanish Usher syndrome type 2 cohort. Molecular Vision, 2013, 19, 367-73.	1.1	13
12	CRB1-Related Retinal Dystrophies in a Cohort of 50 Patients: A Reappraisal in the Light of Specific MÃ¼ller Cell and Photoreceptor CRB1 Isoforms. International Journal of Molecular Sciences, 2021, 22, 12642.	4.1	11
13	The Study of a 231 French Patient Cohort Significantly Extends the Mutational Spectrum of the Two Major Usher Genes MYO7A and USH2A. International Journal of Molecular Sciences, 2021, 22, 13294.	4.1	5
14	Identification of the First Single GSDME Exon 8 Structural Variants Associated with Autosomal Dominant Hearing Loss. Diagnostics, 2022, 12, 207.	2.6	4
15	Natural history of Usher type 2 with the c.2299delG mutation of USH2A in a large cohort. Ophthalmic Genetics, 2022, 43, 470-475.	1.2	3