Veronique Pingault

List of Publications by Year
in descending order

24 papers	2,388 citations	430874 18 h-index	610901 24 g-index
24 all docs	24 docs citations	24 times ranked	2697 citing authors

#	Article	IF	Citations
1	SOX10 mutations in patients with Waardenburg-Hirschsprung disease. Nature Genetics, 1998, 18, 171-173.	21.4	733
2	Review and update of mutations causing Waardenburg syndrome. Human Mutation, 2010, 31, 391-406.	2.5	481
3	Deletions at the SOX10 Gene Locus Cause Waardenburg Syndrome Types 2 and 4. American Journal of Human Genetics, 2007, 81, 1169-1185.	6.2	216
4	Loss-of-Function Mutations in SOX10 Cause Kallmann Syndrome with Deafness. American Journal of Human Genetics, 2013, 92, 707-724.	6.2	177
5	SOX10 mutations in chronic intestinal pseudo-obstruction suggest a complex physiopathological mechanism. Human Genetics, 2002, 111, 198-206.	3.8	123
6	Interactions between Sox10, Edn3 and Ednrb during enteric nervous system and melanocyte development. Developmental Biology, 2006, 295, 232-249.	2.0	115
7	Peripheral neuropathy with hypomyelination, chronic intestinal pseudo-obstruction and deafness: A developmental ?neural crest syndrome? related to a SOX10 mutation. Annals of Neurology, 2000, 48, 671-676.	5.3	98
8	Identification and functional analysis ofSOX10missense mutations in different subtypes of waardenburg syndrome. Human Mutation, 2011, 32, 1436-1449.	2.5	71
9	News from the endothelin-3/EDNRB signaling pathway: Role during enteric nervous system development and involvement in neural crest-associated disorders. Developmental Biology, 2018, 444, S156-S169.	2.0	47
10	SOX10: 20 years of phenotypic plurality and current understanding of its developmental function. Journal of Medical Genetics, 2022, 59, 105-114.	3.2	38
11	Alu-mediated deletion of SOX10 regulatory elements in Waardenburg syndrome type 4. European Journal of Human Genetics, 2012, 20, 990-994.	2.8	37
12	An Impairment of Long Distance SOX10 Regulatory Elements Underlies Isolated Hirschsprung Disease. Human Mutation, 2014, 35, 303-307.	2.5	33
13	EDNRBmutations cause Waardenburg syndrome type II in the heterozygous state. Human Mutation, 2017, 38, 581-593.	2.5	33
14	ADAR1 mediated regulation of neural crest derived melanocytes and Schwann cell development. Nature Communications, 2020, 11, 198.	12.8	30
15	Sox10 and Itgb1 interaction in enteric neural crest cell migration. Developmental Biology, 2013, 379, 92-106.	2.0	28
16	Differentiation of Mouse Enteric Nervous System Progenitor Cells Is Controlled by Endothelin 3 and Requires Regulation of Ednrb by SOX10 and ZEB2. Gastroenterology, 2017, 152, 1139-1150.e4.	1.3	28
17	Phenotypic similarities and differences in patients with a p.Met112Ile mutation in SOX10. American Journal of Medical Genetics, Part A, 2014, 164, 2344-2350.	1.2	21
18	A Novel Mutation in the Endothelin B Receptor Gene in a Moroccan Family with Shah-Waardenburg Syndrome. Molecular Syndromology, 2015, 6, 44-49.	0.8	21

#	Article	IF	Citations
19	EFTUD2 missense variants disrupt protein function and splicing in mandibulofacial dysostosis Guionâ€Almeida type. Human Mutation, 2020, 41, 1372-1382.	2.5	15
20	LKB1 specifies neural crest cell fates through pyruvate-alanine cycling. Science Advances, 2019, 5, eaau5106.	10.3	12
21	Screening of MITF and SOX10 Regulatory Regions in Waardenburg Syndrome Type 2. PLoS ONE, 2012, 7, e41927.	2.5	12
22	Subnuclear re-localization of SOX10 and p54NRB correlates with a unique neurological phenotype associated with SOX10 missense mutations. Human Molecular Genetics, 2015, 24, 4933-4947.	2.9	11
23	Further delineation of auriculocondylar syndrome based on 14 novel cases and reassessment of 25 published cases. Human Mutation, 2022, 43, 582-594.	2.5	6
24	Generation of an iPSC line (IMAGINi022-A) from a patient carrying a SOX10 missense mutation and presenting with deafness, depigmentation and progressive neurological impairment. Stem Cell Research, 2020, 48, 101936.	0.7	2