

Tiziana Bachetti

List of Publications by Year in descending order

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Version: 2024-02-01

52
papers

6,065
citations

304743

22
h-index

197818

49
g-index

54
all docs

54
docs citations

54
times ranked

15356
citing authors

#	ARTICLE	IF	CITATIONS
1	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). <i>Autophagy</i> , 2016, 12, 1-222.	9.1	4,701
2	PHOX2B mutations and polyalanine expansions correlate with the severity of the respiratory phenotype and associated symptoms in both congenital and late onset Central Hypoventilation syndrome. <i>Journal of Medical Genetics</i> , 2004, 41, 373-380.	3.2	248
3	Distinct pathogenetic mechanisms for PHOX2B associated polyalanine expansions and frameshift mutations in congenital central hypoventilation syndrome. <i>Human Molecular Genetics</i> , 2005, 14, 1815-1824.	2.9	106
4	Autophagy contributes to inflammation in patients with TNFR-associated periodic syndrome (TRAPS). <i>Annals of the Rheumatic Diseases</i> , 2013, 72, 1044-1052.	0.9	69
5	Parental origin and somatic mosaicism of PHOX2B mutations in Congenital Central Hypoventilation Syndrome. <i>Human Mutation</i> , 2008, 29, 206-206.	2.5	52
6	A common haplotype at the 5' end of the RET proto-oncogene, overrepresented in Hirschsprung patients, is associated with reduced gene expression. <i>Human Mutation</i> , 2005, 25, 189-195.	2.5	46
7	PHOX2B mutations and genetic predisposition to neuroblastoma. <i>Oncogene</i> , 2005, 24, 3050-3053.	5.9	45
8	The TLX2 homeobox gene is a transcriptional target of PHOX2B in neural-crest-derived cells. <i>Biochemical Journal</i> , 2006, 395, 355-361.	3.7	41
9	PHOX2B-Mediated Regulation of ALK Expression: In Vitro Identification of a Functional Relationship between Two Genes Involved in Neuroblastoma. <i>PLoS ONE</i> , 2010, 5, e13108.	2.5	40
10	Causative and common PHOX2B variants define a broad phenotypic spectrum. <i>Clinical Genetics</i> , 2020, 97, 103-113.	2.0	39
11	Low amounts of PHOX2B expanded alleles in asymptomatic parents suggest unsuspected recurrence risk in congenital central hypoventilation syndrome. <i>Journal of Molecular Medicine</i> , 2011, 89, 505-513.	3.9	37
12	Curcumin induces a fatal energetic impairment in tumor cells in vitro and in vivo by inhibiting ATP-synthase activity. <i>Carcinogenesis</i> , 2018, 39, 1141-1150.	2.8	37
13	In vitro treatments with ceftriaxone promote elimination of mutant glial fibrillary acidic protein and transcription down-regulation. <i>Experimental Cell Research</i> , 2010, 316, 2152-2165.	2.6	36
14	A common variant located in the 3'UTR of the RET gene is associated with protection from Hirschsprung disease. <i>Human Mutation</i> , 2007, 28, 168-176.	2.5	35
15	Geldanamycin promotes nuclear localisation and clearance of PHOX2B misfolded proteins containing polyalanine expansions. <i>International Journal of Biochemistry and Cell Biology</i> , 2007, 39, 327-339.	2.8	34
16	In vitro drug treatments reduce the deleterious effects of aggregates containing polyAla expanded PHOX2B proteins. <i>Neurobiology of Disease</i> , 2012, 45, 508-518.	4.4	32
17	Transcriptional dysregulation and impairment of PHOX2B auto-regulatory mechanism induced by polyalanine expansion mutations associated with congenital central hypoventilation syndrome. <i>Neurobiology of Disease</i> , 2013, 50, 187-200.	4.4	29
18	Structural and functional differences in PHOX2B frameshift mutations underlie isolated or syndromic congenital central hypoventilation syndrome. <i>Human Mutation</i> , 2018, 39, 219-236.	2.5	28

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19	Beneficial effects of curcumin on GFAP filament organization and down-regulation of GFAP expression in an in vitro model of Alexander disease. <i>Experimental Cell Research</i> , 2012, 318, 1844-1854.	2.6	26
20	miR-204 mediates post-transcriptional down-regulation of PHOX2B gene expression in neuroblastoma cells. <i>Biochimica Et Biophysica Acta - Gene Regulatory Mechanisms</i> , 2015, 1849, 1057-1065.	1.9	25
21	Common PHOX2B poly-alanine contractions impair RET gene transcription, predisposing to Hirschsprung disease. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2017, 1863, 1770-1777.	3.8	25
22	An In Vitro Approach to Test the Possible Role of Candidate Factors in the Transcriptional Regulation of the RET Proto-Oncogene. <i>Gene Expression</i> , 2005, 12, 137-149.	1.2	24
23	Tumor necrosis factor receptor-associated periodic syndrome as a model linking autophagy and inflammation in protein aggregation diseases. <i>Journal of Molecular Medicine</i> , 2014, 92, 583-594.	3.9	23
24	Transcriptional regulation of TLX2 and impaired intestinal innervation: possible role of the PHOX2A and PHOX2B genes. <i>European Journal of Human Genetics</i> , 2007, 15, 848-855.	2.8	22
25	Brainstem Anomalies in Two Patients Affected by Congenital Central Hypoventilation Syndrome. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2006, 174, 706-709.	5.6	21
26	Mild functional effects of a novel GFAP mutant allele identified in a familial case of adult-onset Alexander disease. <i>European Journal of Human Genetics</i> , 2008, 16, 462-470.	2.8	20
27	Mutational analysis of the RNX gene in congenital central hypoventilation syndrome. <i>American Journal of Medical Genetics Part A</i> , 2002, 113, 178-182.	2.4	17
28	The E3 ubiquitin ligase TRIM11 mediates the degradation of congenital central hypoventilation syndrome-associated polyalanine-expanded PHOX2B. <i>Journal of Molecular Medicine</i> , 2012, 90, 1025-1035.	3.9	17
29	Ceftriaxone has a therapeutic role in Alexander disease. <i>Progress in Neuro-Psychopharmacology and Biological Psychiatry</i> , 2010, 34, 416-417.	4.8	16
30	Paired-like homeobox gene (PHOX2B) nonpolyalanine repeat expansion mutations (NPARMs): genotype-phenotype correlation in congenital central hypoventilation syndrome (CCHS). <i>Genetics in Medicine</i> , 2021, 23, 1656-1663.	2.4	16
31	A Focus on Regulatory Networks Linking MicroRNAs, Transcription Factors and Target Genes in Neuroblastoma. <i>Cancers</i> , 2021, 13, 5528.	3.7	16
32	A Novel Polymorphic AP-1 Binding Element of the $GFAP$ Promoter is Associated with Different Allelic Transcriptional Activities. <i>Annals of Human Genetics</i> , 2010, 74, 506-515.	0.8	14
33	Recurrence of CCHS associated PHOX2B poly-alanine expansion mutation due to maternal mosaicism. <i>Pediatric Pulmonology</i> , 2014, 49, E45-7.	2.0	14
34	Amphioxus neuroglia: Molecular characterization and evidence for early compartmentalization of the developing nerve cord. <i>Glia</i> , 2021, 69, 1654-1678.	4.9	12
35	Ceftriaxone for Alexander's Disease: A Four-Year Follow-Up. <i>JIMD Reports</i> , 2012, 9, 67-71.	1.5	11
36	Medico-legal investigation in an explicable case of congenital central hypoventilation syndrome due to a rare variant of the PHOX2B gene. <i>Journal of Clinical Forensic and Legal Medicine</i> , 2018, 58, 1-5.	1.0	10

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37	A Common 3'UTR Variant of the PHOX2B Gene Is Associated With Infant Life-Threatening and Sudden Death Events in the Italian Population. <i>Frontiers in Neurology</i> , 2021, 12, 642735.	2.4	10
38	Identification of novel pathways and molecules able to down-regulate PHOX2B gene expression by in vitro drug screening approaches in neuroblastoma cells. <i>Experimental Cell Research</i> , 2015, 336, 43-57.	2.6	9
39	PHOX2A and PHOX2B genes are highly co-expressed in human neuroblastoma. <i>International Journal of Oncology</i> , 1992, 33, 985.	3.3	8
40	Targeting of PHOX2B expression allows the identification of drugs effective in counteracting neuroblastoma cell growth. <i>Oncotarget</i> , 2017, 8, 72133-72146.	1.8	8
41	Comparative genomic sequence analysis coupled to chromatin immunoprecipitation: a screening procedure applied to search for regulatory elements at the RET locus. <i>Physiological Genomics</i> , 2005, 23, 269-274.	2.3	7
42	Toward a therapeutic strategy for polyalanine expansions disorders: In vivo and in vitro models for drugs analysis. <i>European Journal of Paediatric Neurology</i> , 2011, 15, 449-452.	1.6	6
43	The OSMR Gene Is Involved in Hirschsprung Associated Enterocolitis Susceptibility through an Altered Downstream Signaling. <i>International Journal of Molecular Sciences</i> , 2021, 22, 3831.	4.1	6
44	Safe drugs to fight mutant protein overload and alpha-1-antitrypsin deficiency. <i>Journal of Hepatology</i> , 2011, 55, 949-950.	3.7	4
45	Functional Conservation and Genetic Divergence of Chordate Glycinergic Neurotransmission: Insights from Amphioxus Glycine Transporters. <i>Cells</i> , 2021, 10, 3392.	4.1	3
46	Correspondence Regarding: Alexander Disease Mutant Glial Fibrillary Acidic Protein Compromises Glutamate Transport in Astrocytes <i>J Neuropathol Exp Neurol</i> 2010;69:335-45. <i>Journal of Neuropathology and Experimental Neurology</i> , 2010, 69, 1270.1-1270.	1.7	2
47	Alexander Disease Modeling in Zebrafish: An In Vivo System Suitable to Perform Drug Screening. <i>Genes</i> , 2020, 11, 1490.	2.4	2
48	Beneficial Effect of Phenytoin and Carbamazepine on GFAP Gene Expression and Mutant GFAP Folding in a Cellular Model of Alexander's Disease. <i>Frontiers in Pharmacology</i> , 2021, 12, 723218.	3.5	2
49	Functional characterization of a minimal sequence essential for the expression of human TLX2 gene. <i>BMB Reports</i> , 2009, 42, 788-793.	2.4	0
50	In vitro studies of PHOX2B gene mutations in congenital central hypoventilation syndrome. , 2008, , 71-83.		0
51	Generation of two hiPSC lines (UMILi027-A and UMILi028-A) from early and late-onset Congenital Central hypoventilation Syndrome (CCHS) patients carrying a polyalanine expansion mutation in the PHOX2B gene. <i>Stem Cell Research</i> , 2022, 61, 102781.	0.7	0
52	Parental Somatic Mosaicism Uncovers Inheritance of an Apparently De Novo GFAP Mutation. <i>Frontiers in Genetics</i> , 2021, 12, 744068.	2.3	0