Tiziana Bachetti

List of Publications by Year
in descending order

52	6,065	304743 22	197818 49
papers	citations	h-index	g-index

54	54	54	15356
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	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. Stem Cell Research, 2022, 61, 102781.	0.7	0
2	Amphioxus neuroglia: Molecular characterization and evidence for early compartmentalization of the developing nerve cord. Glia, 2021, 69, 1654-1678.	4.9	12
3	A Common 3â€²UTR Variant of the PHOX2B Gene Is Associated With Infant Life-Threatening and Sudden Death Events in the Italian Population. Frontiers in Neurology, 2021, 12, 642735.	2.4	10
4	The OSMR Gene Is Involved in Hirschsprung Associated Enterocolitis Susceptibility through an Altered Downstream Signaling. International Journal of Molecular Sciences, 2021, 22, 3831.	4.1	6
5	Paired-like homeobox gene (PHOX2B) nonpolyalanine repeat expansion mutations (NPARMs): genotypeâ€“phenotype correlation in congenital central hypoventilation syndrome (CCHS). Genetics in Medicine, 2021, 23, 1656-1663.	2.4	16
6	A Focus on Regulatory Networks Linking MicroRNAs, Transcription Factors and Target Genes in Neuroblastoma. Cancers, 2021, 13, 5528.	3.7	16
7	Functional Conservation and Genetic Divergence of Chordate Glycinergic Neurotransmission: Insights from Amphioxus Glycine Transporters. Cells, 2021, 10, 3392.	4.1	3
8	Parental Somatic Mosaicism Uncovers Inheritance of an Apparently De Novo GFAP Mutation. Frontiers in Genetics, 2021, 12, 744068.	2.3	0
9	Beneficial Effect of Phenytoin and Carbamazepine on GFAP Gene Expression and Mutant GFAP Folding in a Cellular Model of Alexanderâ€™s Disease. Frontiers in Pharmacology, 2021, 12, 723218.	3.5	2
10	Causative and common PHOX2B variants define a broad phenotypic spectrum. Clinical Genetics, 2020, 97, 103-113.	2.0	39
11	Alexander Disease Modeling in Zebrafish: An In Vivo System Suitable to Perform Drug Screening. Genes, 2020, 11, 1490.	2.4	2
12	Medico-legal investigation in an explicable case of congenital central hypoventilation syndrome due to a rare variant of the PHOX2B gene. Journal of Clinical Forensic and Legal Medicine, 2018, 58, 1-5.	1.0	10
13	Structural and functional differences inPHOX2Bframeshift mutations underlie isolated or syndromic congenital central hypoventilation syndrome. Human Mutation, 2018, 39, 219-236.	2.5	28
14	Curcumin induces a fatal energetic impairment in tumor cells in vitro and in vivo by inhibiting ATP-synthase activity. Carcinogenesis, 2018, 39, 1141-1150.	2.8	37
15	Common PHOX2B poly-alanine contractions impair RET gene transcription, predisposing to Hirschsprung disease. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2017, 1863, 1770-1777.	3.8	25
16	Targeting ofPHOX2Bexpression allows the identification of drugs effective in counteracting neuroblastoma cell growth. Oncotarget, 2017, 8, 72133-72146.	1.8	8
17	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). Autophagy, 2016, 12, 1-222.	9.1	4,701
18	miR-204 mediates post-transcriptional down-regulation of PHOX2B gene expression in neuroblastoma cells. Biochimica Et Biophysica Acta - Gene Regulatory Mechanisms, 2015, 1849, 1057-1065.	1.9	25

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19	Identification of novel pathways and molecules able to down-regulate PHOX2B gene expression by in vitro drug screening approaches in neuroblastoma cells. Experimental Cell Research, 2015, 336, 43-57.	2.6	9
20	Recurrence of CCHS associated PHOX2B polyâ€alanine expansion mutation due to maternal mosaicism. Pediatric Pulmonology, 2014, 49, E45-7.	2.0	14
21	Tumor necrosis factor receptor-associated periodic syndrome as a model linking autophagy and inflammation in protein aggregation diseases. Journal of Molecular Medicine, 2014, 92, 583-594.	3.9	23
22	Transcriptional dysregulation and impairment of PHOX2B auto-regulatory mechanism induced by polyalanine expansion mutations associated with congenital central hypoventilation syndrome. Neurobiology of Disease, 2013, 50, 187-200.	4.4	29
23	Autophagy contributes to inflammation in patients with TNFR-associated periodic syndrome (TRAPS). Annals of the Rheumatic Diseases, 2013, 72, 1044-1052.	0.9	69
24	Ceftriaxone for Alexanderâ€™s Disease: A Four-Year Follow-Up. JIMD Reports, 2012, 9, 67-71.	1.5	11
25	The E3 ubiquitin ligase TRIM11 mediates the degradation of congenital central hypoventilation syndrome-associated polyalanine-expanded PHOX2B. Journal of Molecular Medicine, 2012, 90, 1025-1035.	3.9	17
26	In vitro drug treatments reduce the deleterious effects of aggregates containing polyAla expanded PHOX2B proteins. Neurobiology of Disease, 2012, 45, 508-518.	4.4	32
27	Beneficial effects of curcumin on GFAP filament organization and down-regulation of GFAP expression in an in vitro model of Alexander disease. Experimental Cell Research, 2012, 318, 1844-1854.	2.6	26
28	Safe drugs to fight mutant protein overload and alpha-1-antitrypsin deficiency. Journal of Hepatology, 2011, 55, 949-950.	3.7	4
29	Toward a therapeutic strategy for polyalanine expansions disorders: In vivo and in vitro models for drugs analysis. European Journal of Paediatric Neurology, 2011, 15, 449-452.	1.6	6
30	Low amounts of PHOX2B expanded alleles in asymptomatic parents suggest unsuspected recurrence risk in congenital central hypoventilation syndrome. Journal of Molecular Medicine, 2011, 89, 505-513.	3.9	37
31	Correspondence Regarding: Alexander Disease Mutant Glial Fibrillary Acidic Protein Compromises Glutamate Transport in Astrocytes J Neuropathol Exp Neurol 2010;69:335â€“45. Journal of Neuropathology and Experimental Neurology, 2010, 69, 1270.1-1270.	1.7	2
32	In vitro treatments with ceftriaxone promote elimination of mutant glial fibrillary acidic protein and transcription down-regulation. Experimental Cell Research, 2010, 316, 2152-2165.	2.6	36
33	A Novel Polymorphic APâ€1 Binding Element of the GFAP Promoter is Associated with Different Allelic Transcriptional Activities. Annals of Human Genetics, 2010, 74, 506-515.	0.8	14
34	PHOX2B-Mediated Regulation of ALK Expression: In Vitro Identification of a Functional Relationship between Two Genes Involved in Neuroblastoma. PLoS ONE, 2010, 5, e13108.	2.5	40
35	Ceftriaxone has a therapeutic role in Alexander disease. Progress in Neuro-Psychopharmacology and Biological Psychiatry, 2010, 34, 416-417.	4.8	16
36	Functional characterization of a minimal sequence essential for the expression of human TLX2 gene. BMB Reports, 2009, 42, 788-793.	2.4	0

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37	Parental origin and somatic mosaicism of PHOX2B mutations in Congenital Central Hypoventilation Syndrome. Human Mutation, 2008, 29, 206-206.	2.5	52
38	Mild functional effects of a novel GFAP mutant allele identified in a familial case of adult-onset Alexander disease. European Journal of Human Genetics, 2008, 16, 462-470.	2.8	20
39	In vitro studies of PHOX2B gene mutations in congenital central hypoventilation syndrome. , 2008, , 71-83.		0
40	Geldanamycin promotes nuclear localisation and clearance of PHOX2B misfolded proteins containing polyalanine expansions. International Journal of Biochemistry and Cell Biology, 2007, 39, 327-339.	2.8	34
41	A common variant located in the 3â€²UTR of theRET gene is associated with protection from Hirschsprung disease. Human Mutation, 2007, 28, 168-176.	2.5	35
42	Transcriptional regulation of TLX2 and impaired intestinal innervation: possible role of the PHOX2A and PHOX2B genes. European Journal of Human Genetics, 2007, 15, 848-855.	2.8	22
43	The TLX2 homeobox gene is a transcriptional target of PHOX2B in neural-crest-derived cells. Biochemical Journal, 2006, 395, 355-361.	3.7	41
44	Brainstem Anomalies in Two Patients Affected by Congenital Central Hypoventilation Syndrome. American Journal of Respiratory and Critical Care Medicine, 2006, 174, 706-709.	5.6	21
45	An In Vitro Approach to Test the Possible Role of Candidate Factors in the Transcriptional Regulation of the <I>RET</I> Proto-Oncogene. Gene Expression, 2005, 12, 137-149.	1.2	24
46	PHOX2B mutations and genetic predisposition to neuroblastoma. Oncogene, 2005, 24, 3050-3053.	5.9	45
47	A common haplotype at the 5â€² end of the RET protoâ€oncogene, overrepresented in Hirschsprung patients, is associated with reduced gene expression. Human Mutation, 2005, 25, 189-195.	2.5	46
48	Comparative genomic sequence analysis coupled to chromatin immunoprecipitation: a screening procedure applied to search for regulatory elements at the RET locus. Physiological Genomics, 2005, 23, 269-274.	2.3	7
49	Distinct pathogenetic mechanisms for PHOX2B associated polyalanine expansions and frameshift mutations in congenital central hypoventilation syndrome. Human Molecular Genetics, 2005, 14, 1815-1824.	2.9	106
50	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. Journal of Medical Genetics, 2004, 41, 373-380.	3.2	248
51	Mutational analysis of theRNX gene in congenital central hypoventilation syndrome. American Journal of Medical Genetics Part A, 2002, 113, 178-182.	2.4	17
52	PHOX2A and PHOX2B genes are highly co-expressed in human neuroblastoma. International Journal of Oncology, 1992, 33, 985.	3.3	8