Sara Benedetti

List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	Gene therapy of experimental brain tumors using neural progenitor cells. Nature Medicine, 2000, 6, 447-450.	15.2	450
2	The "Bystander Effectâ€: Association of U-87 Cell Death with Ganciclovir-Mediated Apoptosis of Nearby Cells and Lack of Effect in Athymic Mice. Human Gene Therapy, 1995, 6, 763-772.	1.4	135
3	Limited Efficacy of the HSV-TK/GCV System for Gene Therapy of Malignant Gliomas and Perspectives for the Combined Transduction of the Interleukin-4 Gene. Human Gene Therapy, 1997, 8, 1345-1353.	1.4	69
4	Brugada syndrome genetics is associated with phenotype severity. European Heart Journal, 2021, 42, 1082-1090.	1.0	59
5	Genetics can contribute to the prognosis of Brugada syndrome: a pilot model for risk stratification. European Journal of Human Genetics, 2013, 21, 911-917.	1.4	58
6	<i>LMNA</i> -associated myopathies. Neurology, 2014, 83, 1634-1644.	1.5	57
7	A Brugada syndrome mutation (p.S216L) and its modulation by p.H558R polymorphism: standard and dynamic characterization. Cardiovascular Research, 2011, 91, 606-616.	1.8	50
8	Analyzing Histopathological Features of Rare Charcot-Marie-Tooth Neuropathies to Unravel Their Pathogenesis. Archives of Neurology, 2010, 67, 1498-505.	4.9	48
9	High-throughput genetic characterization of a cohort of Brugada syndrome patients. Human Molecular Genetics, 2015, 24, 5828-5835.	1.4	35
10	Heat shock protein 27 R127W mutation: evidence of a continuum between axonal Charcot-Marie-Tooth and distal hereditary motor neuropathy. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, 958-962.	0.9	33
11	Cardiac and Neuromuscular Features of Patients With <i>LMNA</i> -Related Cardiomyopathy. Annals of Internal Medicine, 2019, 171, 458.	2.0	33
12	Hereditary Spastic Paraplegia: Beyond Clinical Phenotypes toward a Unified Pattern of Central Nervous System Damage. Radiology, 2015, 276, 207-218.	3.6	32
13	Impaired turnover of hyperfused mitochondria in severe axonal neuropathy due to a novel DRP1 mutation. Human Molecular Genetics, 2020, 29, 177-188.	1.4	30
14	Laminopathies: from the heart of the cell to the clinics. Current Opinion in Neurology, 2004, 17, 553-560.	1.8	25
15	Late gadolinium enhancement role in arrhythmic risk stratification of patients with LMNA cardiomyopathy: results from a long-term follow-up multicentre study. Europace, 2020, 22, 1864-1872.	0.7	21
16	Evaluation of human gene variant detection in amplicon pools by the GS-FLX parallel Pyrosequencer. BMC Genomics, 2008, 9, 464.	1.2	18
17	Skewed X-chromosome inactivation is not associated with premature ovarian failure in a large cohort of Italian patients. American Journal of Medical Genetics, Part A, 2006, 140A, 1349-1351.	0.7	16
18	Protein profiling reveals energy metabolism and cytoskeletal protein alterations in LMNA mutation carriers. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 970-979.	1.8	16

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19	Comparable clinical characteristics in Brugada syndrome patients harboring SCN5A or novel SCN10A variants. Europace, 2019, 21, 1550-1558.	0.7	15
20	Co-segregation of LMNA and PMP22 gene mutations in the same family. Neuromuscular Disorders, 2005, 15, 858-862.	0.3	12
21	SCN5A Nonsense Mutation and NF1 Frameshift Mutation in a Family With Brugada Syndrome and Neurofibromatosis. Frontiers in Genetics, 2019, 10, 50.	1.1	12
22	Genotype/Phenotype Relationship in a Consanguineal Family With Brugada Syndrome Harboring the R1632C Missense Variant in the SCN5A Gene. Frontiers in Physiology, 2019, 10, 666.	1.3	11
23	A novel homozygous mutation in the TRDN gene causes a severe form of pediatric malignant ventricular arrhythmia. Heart Rhythm, 2020, 17, 296-304.	0.3	11
24	Novel SCN5A Frameshift Mutation in Brugada Syndrome Associated With Complex Arrhythmic Phenotype. Frontiers in Genetics, 2019, 10, 547.	1.1	10
25	A new double-trouble phenotype: fascioscapulohumeral muscular dystrophy ameliorates hereditary spastic paraparesis due to spastin mutation. Journal of Neurology, 2015, 262, 476-478.	1.8	8
26	The empowerment of translational research: lessons from laminopathies. Orphanet Journal of Rare Diseases, 2012, 7, 37.	1.2	7
27	Evaluation of damaging effects of splicing mutations: Validation of an in vitro method for diagnostic laboratories. Clinica Chimica Acta, 2014, 436, 276-282.	0.5	7
28	Novel SCN5A p.W697X Nonsense Mutation Segregation in a Family with Brugada Syndrome. International Journal of Molecular Sciences, 2019, 20, 4920.	1.8	7
29	Compound Heterozygous SCN5A Gene Mutations in Aasymptomatic Brugada Syndrome Child. Neurology International, 2012, 2, e11.	0.2	5
30	Novel SCN5A p.V1429M Variant Segregation in a Family with Brugada Syndrome. International Journal of Molecular Sciences, 2020, 21, 5902.	1.8	5
31	Novel SCN5A p.Val1667Asp Missense Variant Segregation and Characterization in a Family with Severe Brugada Syndrome and Multiple Sudden Deaths. International Journal of Molecular Sciences, 2021, 22, 4700.	1.8	5
32	Emerging perspectives on laminopathies. Cell Health and Cytoskeleton, 2016, , 25.	0.7	4
33	Expanding the central nervous system disease spectrum associated with <i>FLNC</i> mutation. Muscle and Nerve, 2019, 59, E33-E37.	1.0	3