

Marina Stroppiano

List of Publications by Year in descending order

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24
papers

676
citations

623734

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24
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all docs

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docs citations

24
times ranked

945
citing authors

#	ARTICLE	IF	CITATIONS
1	Predicting the probability of Gaucher disease in subjects with splenomegaly and thrombocytopenia. <i>Scientific Reports</i> , 2021, 11, 2594.	3.3	12
2	A multicentre observational study for early diagnosis of Gaucher disease in patients with Splenomegaly and/or Thrombocytopenia. <i>European Journal of Haematology</i> , 2016, 96, 352-359.	2.2	34
3	Glucocerebrosidase deficiency in zebrafish affects primary bone ossification through increased oxidative stress and reduced Wnt/ β 2-catenin signaling. <i>Human Molecular Genetics</i> , 2015, 24, 1280-1294.	2.9	46
4	A novel homozygous splicing mutation in PSAP gene causes metachromatic leukodystrophy in two Moroccan brothers. <i>Neurogenetics</i> , 2014, 15, 101-106.	1.4	8
5	Validity of β 2-d-glucosidase activity measured in dried blood samples for detection of potential Gaucher disease patients. <i>Clinical Biochemistry</i> , 2014, 47, 1293-1296.	1.9	16
6	A Multicenter Observational Study For Early Diagnosis Of Gaucher Disease In Patients With Splenomegaly and/Or Thrombocytopenia. <i>Blood</i> , 2013, 122, 4712-4712.	1.4	1
7	Identification and characterization of 15 novel GALC gene mutations causing Krabbe disease. <i>Human Mutation</i> , 2010, 31, E1894-E1914.	2.5	93
8	Molecular characterization of 22 novel UDP-N-acetylglucosamine-1-phosphate transferase β 1- and β 2-subunit (<i>GNPTAB</i>) gene mutations causing mucopolidosis types III β and III α in 46 patients. <i>Human Mutation</i> , 2009, 30, E956-E973.	2.5	38
9	Molecular and functional analysis of the HEXB gene in Italian patients affected with Sandhoff disease: identification of six novel alleles. <i>Neurogenetics</i> , 2009, 10, 49-58.	1.4	22
10	Identification of nine new IDS alleles in mucopolysaccharidosis II. Quantitative evaluation by real-time RT-PCR of mRNAs sensitive to nonsense-mediated and nonstop decay mechanisms. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2006, 1762, 478-484.	3.8	27
11	First-trimester fetal nuchal translucency and inherited metabolic disorders. <i>Prenatal Diagnosis</i> , 2006, 26, 77-80.	2.3	8
12	Characterization of iduronate-2-sulfatase gene-pseudogene recombinations in eight patients with Mucopolysaccharidosis type II revealed by a rapid PCR-based method. <i>Human Mutation</i> , 2005, 25, 491-497.	2.5	37
13	Homozygosity for a non-pseudogene complex glucocerebrosidase allele as cause of an atypical neuronopathic form of Gaucher disease. <i>American Journal of Medical Genetics, Part A</i> , 2005, 134A, 95-96.	1.2	10
14	Early Visual Seizures and Progressive Myoclonus Epilepsy in Neuronopathic Gaucher Disease Due to a Rare Compound Heterozygosity (N188S/S107L). <i>Epilepsia</i> , 2004, 45, 1154-1157.	5.1	148
15	Expression studies of two novel in CIS-mutations identified in an intermediate case of Hunter syndrome. <i>American Journal of Medical Genetics Part A</i> , 2003, 120A, 84-87.	2.4	14
16	Analysis of the glucocerebrosidase gene and mutation profile in 144 Italian gaucher patients. <i>Human Mutation</i> , 2002, 20, 234-235.	2.5	51
17	Contribution of arylsulfatase A mutations located on the same allele to enzyme activity reduction and metachromatic leukodystrophy severity. <i>Human Genetics</i> , 2002, 110, 351-355.	3.8	30
18	Prenatal diagnosis of Pelizaeus-Merzbacher disease: detection of proteolipid protein gene duplication by quantitative fluorescent multiplex PCR. <i>Prenatal Diagnosis</i> , 2001, 21, 668-671.	2.3	19

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19	Aberrant splicing at catalytic site as cause of infantile onset glycogen storage disease type II (GSDII): Molecular identification of a novel IVS9 (+2GT?GC) in combination with rare IVS10 (+1GT?CT). American Journal of Medical Genetics Part A, 2001, 101, 55-58.	2.4	10
20	Evidence for a Founder Effect in Sicilian Patients with Glycogen Storage Disease Type II. Human Heredity, 2000, 50, 331-333.	0.8	18
21	Somatic Mosaicism in a Patient with Gaucher Disease Type 2: Implication for Genetic Counseling and Therapeutic Decision-Making. Blood Cells, Molecules, and Diseases, 2000, 26, 611-612.	1.4	11
22	New mutation (S298P) in a patient with glycogen storage disease type IA. Human Mutation, 1998, 11, S329-S329.	2.5	2
23	A 9-bp deletion (2320del9) on the background of the arylsulfatase A pseudodeficiency allele in a metachromatic leukodystrophy patient and in a patient with nonprogressive neurological symptoms. Human Genetics, 1998, 102, 50-53.	3.8	16
24	A T > C transition causing a Leu > Pro substitution in a conserved region of the arylsulfatase A gene in a late infantile metachromatic leukodystrophy patient. Clinical Genetics, 1997, 52, 65-67.	2.0	5