Marina Stroppiano

List of Publications by Year
in descending order

24 papers	676 citations	623734 14 h-index	610901 24 g-index
24 all docs	24 docs citations	24 times ranked	945 citing authors

#	Article	IF	Citations
1	Early Visual Seizures and Progressive Myoclonus Epilepsy in Neuronopathic Gaucher Disease Due to a Rare Compound Heterozygosity (N188S/S107L). Epilepsia, 2004, 45, 1154-1157.	5.1	148
2	Identification and characterization of 15 novel GALC gene mutations causing Krabbe disease. Human Mutation, 2010, 31, E1894-E1914.	2.5	93
3	Analysis of the glucocerebrosidase gene and mutation profile in 144 Italian gaucher patients. Human Mutation, 2002, 20, 234-235.	2.5	51
4	Glucocerebrosidase deficiency in zebrafish affects primary bone ossification through increased oxidative stress and reduced Wnt/Î²-catenin signaling. Human Molecular Genetics, 2015, 24, 1280-1294.	2.9	46
5	Molecular characterization of 22 novel UDP-N-acetylglucosamine-1-phosphate transferase Î±- and Î²-subunit (GNPTAB) gene mutations causing mucolipidosis types IIÎ±/Î² and IIIÎ±/Î² in 46 patients. Human Mutation, 2009, 30, E956-E973.	2.5	38
6	Characterization of iduronate-2-sulfatase gene-pseudogene recombinations in eight patients with Mucopolysaccharidosis type II revealed by a rapid PCR-based method. Human Mutation, 2005, 25, 491-497.	2.5	37
7	A multicentre observational study for early diagnosis of Gaucher disease in patients with Splenomegaly and/or Thrombocytopenia. European Journal of Haematology, 2016, 96, 352-359.	2.2	34
8	Contribution of arylsulfatase A mutations located on the same allele to enzyme activity reduction and metachromatic leukodystrophy severity. Human Genetics, 2002, 110, 351-355.	3.8	30
9	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. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2006, 1762, 478-484.	3.8	27
10	Molecular and functional analysis of the HEXB gene in Italian patients affected with Sandhoff disease: identification of six novel alleles. Neurogenetics, 2009, 10, 49-58.	1.4	22
11	Prenatal diagnosis of Pelizaeus-Merzbacher disease: detection of proteolipid protein gene duplication by quantitative fluorescent multiplex PCR. Prenatal Diagnosis, 2001, 21, 668-671.	2.3	19
12	Evidence for a Founder Effect in Sicilian Patients with Glycogen Storage Disease Type II. Human Heredity, 2000, 50, 331-333.	0.8	18
13	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
14	Validity of Î²-d-glucosidase activity measured in dried blood samples for detection of potential Gaucher disease patients. Clinical Biochemistry, 2014, 47, 1293-1296.	1.9	16
15	Expression studies of two novel in CIS-mutations identified in an intermediate case of Hunter syndrome. American Journal of Medical Genetics Part A, 2003, 120A, 84-87.	2.4	14
16	Predicting the probability of Gaucher disease in subjects with splenomegaly and thrombocytopenia. Scientific Reports, 2021, 11, 2594.	3.3	12
17	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
18	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

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19	Homozygosity for a non-pseudogene complex glucocerebrosidase allele as cause of an atypical neuronopathic form of Gaucher disease. American Journal of Medical Genetics, Part A, 2005, 134A, 95-96.	1.2	10
20	First-trimester fetal nuchal translucency and inherited metabolic disorders. Prenatal Diagnosis, 2006, 26, 77-80.	2.3	8
21	A novel homozygous splicing mutation in PSAP gene causes metachromatic leukodystrophy in two Moroccan brothers. Neurogenetics, 2014, 15, 101-106.	1.4	8
22	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
23	New mutation (S298P) in a patient with glycogen storage disease type IA. Human Mutation, 1998, 11, S329-S329.	2.5	2
24	A Multicenter Observational Study For Early Diagnosis Of Gaucher Disease In Patients With Splenomegaly and/Or Thrombocytopenia. Blood, 2013, 122, 4712-4712.	1.4	1