Grazia Mancini

List of Publications by Year
in descending order

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45 papers	3,445 citations	172457 29 h-index	233421 45 g-index
48 all docs	48 docs citations	48 times ranked	6673 citing authors

#	Article	IF	Citations
1	Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. Brain, 2017, 140, 1316-1336.	7.6	426
2	A new gene, encoding an anion transporter, is mutated in sialic acid storage diseases. Nature Genetics, 1999, 23, 462-465.	21.4	252
3	GermlineBRAFmutations in Noonan, LEOPARD, and cardiofaciocutaneous syndromes: Molecular diversity and associated phenotypic spectrum. Human Mutation, 2009, 30, 695-702.	2.5	251
4	Human USP18 deficiency underlies type 1 interferonopathy leading to severe pseudo-TORCH syndrome. Journal of Experimental Medicine, 2016, 213, 1163-1174.	8.5	224
5	Mutations in the Gene Encoding Capillary Morphogenesis Protein 2 Cause Juvenile Hyaline Fibromatosis and Infantile Systemic Hyalinosis. American Journal of Human Genetics, 2003, 73, 791-800.	6.2	209
6	Targeted loss of Arx results in a developmental epilepsy mouse model and recapitulates the human phenotype in heterozygous females. Brain, 2009, 132, 1563-1576.	7.6	178
7	<scp>USP</scp> 18 lack in microglia causes destructive interferonopathy of the mouse brain. EMBO Journal, 2015, 34, 1612-1629.	7.8	178
8	Mutation in the AP4M1 Gene Provides a Model for Neuroaxonal Injury in Cerebral Palsy. American Journal of Human Genetics, 2009, 85, 40-52.	6.2	156
9	Homozygous Nonsense Mutations in KIAA1279 Are Associated with Malformations of the Central and Enteric Nervous Systems. American Journal of Human Genetics, 2005, 77, 120-126.	6.2	138
10	Homozygous MTTP and APOB mutations may lead to hepatic steatosis and fibrosis despite metabolic differences in congenital hypocholesterolemia. Journal of Hepatology, 2014, 61, 891-902.	3.7	116
11	Baraitserâ€“Winter cerebrofrontofacial syndrome: delineation of the spectrum in 42 cases. European Journal of Human Genetics, 2015, 23, 292-301.	2.8	115
12	The Spectrum of SLC17A5-Gene Mutations Resulting in Free Sialic Acidâ€“Storage Diseases Indicates Some Genotype-Phenotype Correlation. American Journal of Human Genetics, 2000, 67, 832-840.	6.2	107
13	Three new families with arterial tortuosity syndrome. American Journal of Medical Genetics Part A, 2004, 131A, 134-143.	2.4	85
14	COL4A2 mutation associated with familial porencephaly and small-vessel disease. European Journal of Human Genetics, 2012, 20, 844-851.	2.8	84
15	The Gene for Juvenile Hyaline Fibromatosis Maps to Chromosome 4q21. American Journal of Human Genetics, 2002, 71, 975-980.	6.2	71
16	Expanding the Phenotype Associated with NAA10â€Related Nâ€Terminal Acetylation Deficiency. Human Mutation, 2016, 37, 755-764.	2.5	70
17	The clinical spectrum of complete FBN1 allele deletions. European Journal of Human Genetics, 2011, 19, 247-252.	2.8	65
18	Molecular and functional analysis ofSUMF1 mutations in multiple sulfatase deficiency. Human Mutation, 2004, 23, 576-581.	2.5	63

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19	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. American Journal of Human Genetics, 2018, 102, 985-994.	6.2	59
20	MACF1 Mutations Encoding Highly Conserved Zinc-Binding Residues of the GAR Domain Cause Defects in Neuronal Migration and Axon Guidance. American Journal of Human Genetics, 2018, 103, 1009-1021.	6.2	57
21	Phenotypic variability of atypical 22q11.2 deletions not including TBX1. American Journal of Medical Genetics, Part A, 2012, 158A, 2412-2420.	1.2	53
22	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogryposis. American Journal of Human Genetics, 2019, 105, 689-705.	6.2	48
23	Purification of the Lysosomal Sialic Acid Transporter. Journal of Biological Chemistry, 1998, 273, 34568-34574.	3.4	45
24	Expanded mutational spectrum in Cohen syndrome, tissue expression, and transcript variants ofCOH1. Human Mutation, 2009, 30, E404-E420.	2.5	44
25	Characterization of a heavy metal ion transporter in the lysosomal membrane. FEBS Letters, 1998, 436, 223-227.	2.8	37
26	ACTA2 mutation with childhood cardiovascular, autonomic and brain anomalies and severe outcome. American Journal of Medical Genetics, Part A, 2013, 161, 1376-1380.	1.2	36
27	Pitfalls in the Diagnosis of Multiple Sulfatase Deficiency. Neuropediatrics, 2001, 32, 38-40.	0.6	34
28	Novel noâ€stop FLNA mutation causes multiâ€organ involvement in males. American Journal of Medical Genetics, Part A, 2013, 161, 2376-2384.	1.2	33
29	DNA analysis of AHI1, NPHP1 and CYCLIN D1 in Joubert syndrome patients from the Netherlands. European Journal of Medical Genetics, 2008, 51, 24-34.	1.3	32
30	Sialin, an anion transporter defective in sialic acid storage diseases, shows highly variable expression in adult mouse brain, and is developmentally regulated. Neurobiology of Disease, 2005, 19, 351-365.	4.4	28
31	TMX2 Is a Crucial Regulator of Cellular Redox State, and Its Dysfunction Causes Severe Brain Developmental Abnormalities. American Journal of Human Genetics, 2019, 105, 1126-1147.	6.2	25
32	Nucleotide-activated chloride channels in lysosomal membranes. Biochemical and Biophysical Research Communications, 1992, 187, 254-260.	2.1	20
33	Transport of organic anions by the lysosomal sialic acid transporter: a functional approach towards the gene for sialic acid storage disease. FEBS Letters, 1999, 446, 65-68.	2.8	19
34	Long-term follow-up of type 1 lissencephaly: survival is related to neuroimaging abnormalities. Developmental Medicine and Child Neurology, 2011, 53, 417-421.	2.1	15
35	Progressive leukoencephalopathy impairs neurobehavioral development in sialin-deficient mice. Experimental Neurology, 2017, 291, 106-119.	4.1	10
36	Mutated zinc finger protein of the cerebellum 1 leads to microcephaly, cortical malformation, callosal agenesis, cerebellar dysplasia, tethered cord and scoliosis. European Journal of Medical Genetics, 2018, 61, 783-789.	1.3	10

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37	Is hearing loss a feature of Joubert syndrome, a ciliopathy?. International Journal of Pediatric Otorhinolaryngology, 2010, 74, 1034-1038.	1.0	8
38	A fetus with de novo 2q33.2q35 deletion including MAP2 with brain anomalies, esophageal atresia, and laryngeal stenosis. American Journal of Medical Genetics, Part A, 2014, 164, 194-198.	1.2	8
39	Asymmetric polymicrogyria and periventricular nodular heterotopia due to mutation in ARX. American Journal of Medical Genetics, Part A, 2012, 158A, 1472-1476.	1.2	7
40	Biallelic DAB1 Variants Are Associated With Mild Lissencephaly and Cerebellar Hypoplasia. Neurology: Genetics, 2021, 7, e558.	1.9	7
41	Unbalanced der(5)t(5;20) translocation associated with megalencephaly, perisylvian polymicrogyria, polydactyly and hydrocephalus. American Journal of Medical Genetics, Part A, 2010, 152A, 1488-1497.	1.2	6
42	The Phenotypic Spectrum of PNKP-Associated Disease and the Absence of Immunodeficiency and Cancer Predisposition in a Dutch Cohort. Pediatric Neurology, 2020, 113, 26-32.	2.1	6
43	Multidisciplinary interaction and MCD gene discovery. The perspective of the clinical geneticist. European Journal of Paediatric Neurology, 2021, 35, 27-34.	1.6	3
44	Periventricular nodular heterotopia and distal limb deficiency: A recurrent association. American Journal of Medical Genetics, Part A, 2010, 152A, 954-959.	1.2	2
45	Re: Polymicrogyria versus pachygyria in 22q11 microdeletion. American Journal of Medical Genetics, Part A, 2005, 136A, 419-419.	1.2	1