Maria Piane

List of Publications by Citations

30 papers	739 citations	12 h-index	27 g-index
34 ext. papers	834 ext. citations	4.9 avg, IF	2.72 L-index

#	Paper	IF	Citations
30	Genotype-phenotype relationships in ataxia-telangiectasia and variants. *American Journal of Human Genetics*, **1998**, 62, 551-61	11	211
29	Ataxia with oculomotor apraxia type 2: a clinical, pathologic, and genetic study. *Neurology*, **2006**, 66, 1207-10	6.5	99
28	MRE11 mutations and impaired ATM-dependent responses in an Italian family with ataxia-telangiectasia-like disorder. *Human Molecular Genetics*, **2004**, 13, 2155-63	5.6	92
27	The novel human gene aprataxin is directly involved in DNA single-strand-break repair. *Cellular and Molecular Life Sciences*, **2005**, 62, 485-91	10.3	69
26	Genetics of migraine and pharmacogenomics: some considerations. *Journal of Headache and Pain*, **2007**, 8, 334-9	8.8	33
25	Role of senataxin in DNA damage and telomeric stability. *DNA Repair*, **2011**, 10, 199-209	4.3	27
24	Majewski osteodysplastic primordial dwarfism type II (MOPD II) syndrome previously diagnosed as Seckel syndrome: report of a novel mutation of the PCNT gene. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 2452-6	2.5	25
23	Different clinical and immunological presentation of ataxia-telangiectasia within the same family. *Neuropediatrics*, **2008**, 39, 43-5	1.6	25
22	p53 centrosomal localization diagnoses ataxia-telangiectasia homozygotes and heterozygotes. *Journal of Clinical Investigation*, **2013**, 123, 1335-42	15.9	17
21	DHPLC screening of ATM gene in Italian patients affected by ataxia-telangiectasia: fourteen novel ATM mutations. *Disease Markers*, **2006**, 22, 257-64	3.2	15
20	Cutaneous venous malformations related to KRIT1 mutation: case report and literature review. *Journal of Molecular Neuroscience*, **2013**, 51, 442-5	3.3	13
19	A Next-Generation Sequencing Approach to Identify Gene Mutations in Early- and Late-Onset Hypertrophic Cardiomyopathy Patients of an Italian Cohort. *International Journal of Molecular Sciences*, **2016**, 17,	6.3	13
18	Founder effects for ATM gene mutations in Italian Ataxia Telangiectasia families. *Annals of Human Genetics*, **2009**, 73, 532-9	2.2	12
17	Molecular prenatal diagnosis of ataxia telangiectasia heterozygosity by direct mutational assays. *Prenatal Diagnosis*, **1999**, 19, 542-5	3.2	12
16	Genetic Testing and Clinical Management Practices for Variants in Non-BRCA1/2 Breast (and Breast/Ovarian) Cancer Susceptibility Genes: An International Survey by the Evidence-Based Network for the Interpretation of Germline Mutant Alleles (ENIGMA) Clinical Working Group. *JCO Precision Oncology*, **2018**, 2,	3.6	12
15	Homozygosity for c 6325T>G transition in the ATM gene causes an atypical, late-onset variant form of ataxia-telangiectasia. *Journal of Neurology*, **2010**, 257, 1738-40	5.5	10
14	Risk Stratification in Hypertrophic Cardiomyopathy. Insights from Genetic Analysis and Cardiopulmonary Exercise Testing. *Journal of Clinical Medicine*, **2020**, 9,	5.1	9

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13	Control of cell respiration by nitric oxide in Ataxia Telangiectasia lymphoblastoid cells. *Biochimica Et Biophysica Acta - Bioenergetics*, **2008**, 1777, 66-73	4.6	9
12	Rapid detection of copy number variations and point mutations in genes using a single workflow by ion semiconductor sequencing pipeline. *Oncotarget*, **2018**, 9, 33648-33655	3.3	9
11	Novel compound heterozygous mutations in a child with Ataxia-Telangiectasia showing unrelated cerebellar disorders. *Journal of the Neurological Sciences*, **2016**, 371, 48-53	3.2	6
10	Impact of Cystic Fibrosis Transmembrane Regulator (CFTR) gene mutations on male infertility. *Archivio Italiano Di Urologia Andrologia*, **2014**, 86, 171-4	1.6	4
9	Six novel ATM mutations in Italian patients with classical ataxia-telangiectasia. *Human Mutation*, **2003**, 21, 450	4.7	4
8	Modulation of hypersensitivity to oxidative DNA damage in ATM defective cells induced by potassium bromate by inhibition of the Poly (ADP-ribose) polymerase (PARP). *Mutation Research - Genetic Toxicology and Environmental Mutagenesis*, **2018**, 836, 117-123	3	3
7	Clinical and genetic studies in a family with a new splice-site mutation in the choroideremia gene. *Molecular Vision*, **2014**, 20, 325-33	2.3	3
6	Genotype-Phenotype Correlations in Monogenic Parkinson Disease: A Review on Clinical and Molecular Findings. *Frontiers in Neurology*, **2021**, 12, 648588	4.1	3
5	Cytotoxic T lymphocyte antigen 4 polymorphism 49 (A>G) and migraine. *Journal of Headache and Pain*, **2005**, 6, 188-90	8.8	2
4	Phenotype expression in a case of adult cystic fibrosis caused by an extremely rare compound heterozygous genotype (2183AA>G/2789+5G>A). *Pancreas*, **2009**, 38, 599-601	2.6	1
3	Impact of the COVID-19 Pandemic on Clinical Pathways for Non-SARS-CoV-2 Related Diseases in the Lazio Region, Italy.. *International Journal of Environmental Research and Public Health*, **2022**, 19,	4.6	1
2	TNNI3 and KCNQ1 co-inherited variants in a family with hypertrophic cardiomyopathy and long QT phenotypes: A case report. *Molecular Genetics and Metabolism Reports*, **2021**, 27, 100743	1.8	0
1	Altered NK-cell compartment and dysfunctional NKG2D/NKG2D-ligand axis in patients with ataxia-telangiectasia. *Clinical Immunology*, **2021**, 230, 108802	9	