

# Rosangela Ferese

## List of Publications by Year in descending order

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Version: 2024-02-01

36  
papers

984  
citations

516710

16  
h-index

434195

31  
g-index

37  
all docs

37  
docs citations

37  
times ranked

2044  
citing authors

#	ARTICLE	IF	CITATIONS
1	Familial transposition of the great arteries caused by multiple mutations in laterality genes. <i>Heart</i> , 2010, 96, 673-677.	2.9	126
2	Activating mutations in RRAS underlie a phenotype within the RASopathy spectrum and contribute to leukaemogenesis. <i>Human Molecular Genetics</i> , 2014, 23, 4315-4327.	2.9	114
3	ccf-mtDNA as a Potential Link Between the Brain and Immune System in Neuro-Immunological Disorders. <i>Frontiers in Immunology</i> , 2019, 10, 1064.	4.8	83
4	Spectrum of MEK1 and MEK2 gene mutations in cardio-facio-cutaneous syndrome and genotype-phenotype correlations. <i>European Journal of Human Genetics</i> , 2009, 17, 733-740.	2.8	74
5	RASopathies: Clinical Diagnosis in the First Year of Life. <i>Molecular Syndromology</i> , 2010, 1, 282-289.	0.8	73
6	New mutations in <i>ZFPM2/FOG2</i> gene in tetralogy of Fallot and double outlet right ventricle. <i>Clinical Genetics</i> , 2011, 80, 184-190.	2.0	69
7	Novel and recurrent EVC and EVC2 mutations in Ellis-van Creveld syndrome and Weyers acrofacial dysostosis. <i>European Journal of Medical Genetics</i> , 2013, 56, 80-87.	1.3	64
8	Four Copies of <i>SNCA</i> Responsible for Autosomal Dominant Parkinson's Disease in Two Italian Siblings. <i>Parkinson's Disease</i> , 2015, 2015, 1-6.	1.1	41
9	A variant in the carboxyl-terminus of connexin 40 alters GAP junctions and increases risk for tetralogy of Fallot. <i>European Journal of Human Genetics</i> , 2013, 21, 69-75.	2.8	36
10	Congenital heart defects in recurrent reciprocal 1q21.1 deletion and duplication syndromes: Rare association with pulmonary valve stenosis. <i>European Journal of Medical Genetics</i> , 2013, 56, 144-149.	1.3	33
11	Methamphetamine persistently increases alpha-synuclein and suppresses gene promoter methylation within striatal neurons. <i>Brain Research</i> , 2019, 1719, 157-175.	2.2	28
12	Using global team science to identify genetic parkinson's disease worldwide. <i>Annals of Neurology</i> , 2019, 86, 153-157.	5.3	26
13	Rapamycin promotes differentiation increasing $\beta$ -tubulin, NeuN, and NeuroD while suppressing nestin expression in glioblastoma cells. <i>Oncotarget</i> , 2017, 8, 29574-29599.	1.8	24
14	Rapamycin Ameliorates Defects in Mitochondrial Fission and Mitophagy in Glioblastoma Cells. <i>International Journal of Molecular Sciences</i> , 2021, 22, 5379.	4.1	22
15	Heterozygous PLA2G6 Mutation Leads to Iron Accumulation Within Basal Ganglia and Parkinson's Disease. <i>Frontiers in Neurology</i> , 2018, 9, 536.	2.4	20
16	Dissecting Molecular Features of Gliomas: Genetic Loci and Validated Biomarkers. <i>International Journal of Molecular Sciences</i> , 2020, 21, 685.	4.1	18
17	A New Splicing Mutation in the L1CAM Gene Responsible for X-Linked Hydrocephalus (HSAS). <i>Journal of Molecular Neuroscience</i> , 2016, 59, 376-381.	2.3	16
18	Vacuolar Protein Sorting Genes in Parkinson's Disease: A Re-appraisal of Mutations Detection Rate and Neurobiology of Disease. <i>Frontiers in Neuroscience</i> , 2016, 10, 532.	2.8	15

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19	Heterozygous missense mutations in <i>NFATC1</i> are associated with atrioventricular septal defect. <i>Human Mutation</i> , 2018, 39, 1428-1441.	2.5	15
20	Quantitative Ultrastructural Morphometry and Gene Expression of mTOR-Related Mitochondriogenesis within Glioblastoma Cells. <i>International Journal of Molecular Sciences</i> , 2020, 21, 4570.	4.1	14
21	Novel and recurrent JAG1 mutations in patients with tetralogy of Fallot. <i>Clinical Genetics</i> , 2011, 80, 591-594.	2.0	11
22	<i>JAG1</i> Mutation in a patient with deletion 22q11.2 syndrome and tetralogy of Fallot. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 3133-3136.	1.2	9
23	The Monoamine Brainstem Reticular Formation as a Paradigm for Re-Defining Various Phenotypes of Parkinson's Disease Owing Genetic and Anatomical Specificity. <i>Frontiers in Cellular Neuroscience</i> , 2017, 11, 102.	3.7	9
24	Developmental Coordination Disorder in a Patient with Mental Disability and a Mild Phenotype Carrying Terminal 6q26-qter Deletion. <i>Frontiers in Genetics</i> , 2017, 8, 206.	2.3	7
25	Corticosterone Upregulates Gene and Protein Expression of Catecholamine Markers in Organotypic Brainstem Cultures. <i>International Journal of Molecular Sciences</i> , 2019, 20, 2901.	4.1	7
26	A Single Nucleotide ADA Genetic Variant Is Associated to Central Inflammation and Clinical Presentation in MS: Implications for Cladribine Treatment. <i>Genes</i> , 2020, 11, 1152.	2.4	5
27	The Autophagy-Related Organelle Autophagosome Is Suppressed within Ischemic Penumbra. <i>International Journal of Molecular Sciences</i> , 2021, 22, 10364.	4.1	5
28	The BDNF Val66Met Polymorphism (rs6265) Modulates Inflammation and Neurodegeneration in the Early Phases of Multiple Sclerosis. <i>Genes</i> , 2022, 13, 332.	2.4	5
29	A Large Family with p.Arg554His Mutation in ABCD1: Clinical Features and Genotype/Phenotype Correlation in Female Carriers. <i>Genes</i> , 2021, 12, 775.	2.4	4
30	Occurrence of Total and Proteinase K-Resistant Alpha-Synuclein in Glioblastoma Cells Depends on mTOR Activity. <i>Cancers</i> , 2022, 14, 1382.	3.7	4
31	Interleukin 6 SNP rs1818879 Regulates Radiological and Inflammatory Activity in Multiple Sclerosis. <i>Genes</i> , 2022, 13, 897.	2.4	3
32	Mitochondrial Serine Protease HTRA2 p.G399S in a Female with Di George Syndrome and Parkinson's Disease. <i>Parkinson's Disease</i> , 2018, 2018, 1-6.	1.1	2
33	PCR-based approach for qualitative molecular analysis of six neurotropic pathogens. <i>Acta Virologica</i> , 2017, 61, 273-279.	0.8	1
34	Next Generation Sequencing and ALS: known genes, different phenotypes. <i>Archives Italiennes De Biologie</i> , 2018, 155, 159-166.	0.4	1
35	Short history of the "Genomic Revolution" and implication for neurological institutes. <i>Rivista Italiana Della Medicina Di Laboratorio</i> , 2015, 11, 1-13.	0.4	0
36	Structural modeling of altered CLCN1 conformation following a novel mutation in a patient affected by autosomal dominant myotonia congenita (Thomsen disease). <i>Archives Italiennes De Biologie</i> , 2018, 155, 275-278.	0.4	0