

# Grazia M S Mancini

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/2975277/publications.pdf>

Version: 2024-02-01

67  
papers

3,847  
citations

159585

30  
h-index

138484

58  
g-index

68  
all docs

68  
docs citations

68  
times ranked

8341  
citing authors

#	ARTICLE	IF	CITATIONS
1	De novo germline and postzygotic mutations in AKT3, PIK3R2 and PIK3CA cause a spectrum of related megalencephaly syndromes. <i>Nature Genetics</i> , 2012, 44, 934-940.	21.4	621
2	Human USP18 deficiency underlies type 1 interferonopathy leading to severe pseudo-TORCH syndrome. <i>Journal of Experimental Medicine</i> , 2016, 213, 1163-1174.	8.5	224
3	XRCC1 mutation is associated with PARP1 hyperactivation and cerebellar ataxia. <i>Nature</i> , 2017, 541, 87-91.	27.8	209
4	The expanding phenotype of COL4A1 and COL4A2 mutations: clinical data on 13 newly identified families and a review of the literature. <i>Genetics in Medicine</i> , 2015, 17, 843-853.	2.4	204
5	<scp>USP</scp> 18 lack in microglia causes destructive interferonopathy of the mouse brain. <i>EMBO Journal</i> , 2015, 34, 1612-1629.	7.8	178
6	Definitions and classification of malformations of cortical development: practical guidelines. <i>Brain</i> , 2020, 143, 2874-2894.	7.6	145
7	<i>SYNGAP1</i> encephalopathy. <i>Neurology</i> , 2019, 92, e96-e107.	1.1	131
8	RAC1 Missense Mutations in Developmental Disorders with Diverse Phenotypes. <i>American Journal of Human Genetics</i> , 2017, 101, 466-477.	6.2	119
9	De novo CCND2 mutations leading to stabilization of cyclin D2 cause megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome. <i>Nature Genetics</i> , 2014, 46, 510-515.	21.4	118
10	Sodium Channel SCN3A (NaV1.3) Regulation of Human Cerebral Cortical Folding and Oral Motor Development. <i>Neuron</i> , 2018, 99, 905-913.e7.	8.1	109
11	Loss-of-Function and Gain-of-Function Mutations in KCNQ5 Cause Intellectual Disability or Epileptic Encephalopathy. <i>American Journal of Human Genetics</i> , 2017, 101, 65-74.	6.2	99
12	SYT1-associated neurodevelopmental disorder: a case series. <i>Brain</i> , 2018, 141, 2576-2591.	7.6	98
13	Altered distribution of ATG9A and accumulation of axonal aggregates in neurons from a mouse model of AP-4 deficiency syndrome. <i>PLoS Genetics</i> , 2018, 14, e1007363.	3.5	85
14	COL4A2 mutation associated with familial porencephaly and small-vessel disease. <i>European Journal of Human Genetics</i> , 2012, 20, 844-851.	2.8	84
15	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. <i>Genetics in Medicine</i> , 2019, 21, 1295-1307.	2.4	80
16	De novo loss-of-function KCNMA1 variants are associated with a new multiple malformation syndrome and a broad spectrum of developmental and neurological phenotypes. <i>Human Molecular Genetics</i> , 2019, 28, 2937-2951.	2.9	76
17	Expanding the Phenotype Associated with NAA10â€“Related Nâ€“Terminal Acetylation Deficiency. <i>Human Mutation</i> , 2016, 37, 755-764.	2.5	70
18	Progressive cerebellar atrophy and polyneuropathy: expanding the spectrum of PNKP mutations. <i>Neurogenetics</i> , 2013, 14, 43-51.	1.4	66

#	ARTICLE	IF	CITATIONS
19	Human TBK1 deficiency leads to autoinflammation driven by TNF-induced cell death. <i>Cell</i> , 2021, 184, 4447-4463.e20.	28.9	64
20	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. <i>American Journal of Human Genetics</i> , 2018, 102, 985-994.	6.2	59
21	MACF1 Mutations Encoding Highly Conserved Zinc-Binding Residues of the GAR Domain Cause Defects in Neuronal Migration and Axon Guidance. <i>American Journal of Human Genetics</i> , 2018, 103, 1009-1021.	6.2	57
22	International consensus recommendations on the diagnostic work-up for malformations of cortical development. <i>Nature Reviews Neurology</i> , 2020, 16, 618-635.	10.1	53
23	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogyposis. <i>American Journal of Human Genetics</i> , 2019, 105, 689-705.	6.2	48
24	Hypomorphic Recessive Variants in SUFU Impair the Sonic Hedgehog Pathway and Cause Joubert Syndrome with Cranio-facial and Skeletal Defects. <i>American Journal of Human Genetics</i> , 2017, 101, 552-563.	6.2	45
25	Combined cardiological and neurological abnormalities due to filamin A gene mutation. <i>Clinical Research in Cardiology</i> , 2011, 100, 45-50.	3.3	43
26	Histone H3.3 beyond cancer: Germline mutations in <i>Histone 3 Family 3A and 3B</i> cause a previously unidentified neurodegenerative disorder in 46 patients. <i>Science Advances</i> , 2020, 6, .	10.3	43
27	<i>WDR73</i> Mutations Cause Infantile Neurodegeneration and Variable Glomerular Kidney Disease. <i>Human Mutation</i> , 2015, 36, 1021-1028.	2.5	42
28	Germline activating AKT3 mutation associated with megalencephaly, polymicrogyria, epilepsy and hypoglycemia. <i>Molecular Genetics and Metabolism</i> , 2015, 114, 467-473.	1.1	42
29	<i>ACTA2</i> mutation with childhood cardiovascular, autonomic and brain anomalies and severe outcome. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 1376-1380.	1.2	36
30	<i>ATP1A2</i> and <i>ATP1A3</i> associated early profound epileptic encephalopathy and polymicrogyria. <i>Brain</i> , 2021, 144, 1435-1450.	7.6	35
31	Molybdenum cofactor deficiency: Identification of a patient with homozygote mutation in the <i>MOCS3</i> gene. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1601-1606.	1.2	33
32	CysteinyI-tRNA Synthetase Mutations Cause a Multi-System, Recessive Disease That Includes Microcephaly, Developmental Delay, and Brittle Hair and Nails. <i>American Journal of Human Genetics</i> , 2019, 104, 520-529.	6.2	31
33	Severe presentation of <i>WDR62</i> mutation: Is there a role for modifying genetic factors?. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2161-2171.	1.2	30
34	Linking the heart and the brain: Neurodevelopmental disorders in patients with catecholaminergic polymorphic ventricular tachycardia. <i>Heart Rhythm</i> , 2019, 16, 220-228.	0.7	29
35	Mutations in the Heterotopia Gene <i>Eml1/EML1</i> Severely Disrupt the Formation of Primary Cilia. <i>Cell Reports</i> , 2019, 28, 1596-1611.e10.	6.4	28
36	Genotype-phenotype correlation of contiguous gene deletions of <i>SLC6A8</i> , <i>BCAP31</i> and <i>ABCD1</i> . <i>Clinical Genetics</i> , 2015, 87, 141-147.	2.0	25

#	ARTICLE	IF	CITATIONS
37	<i>CSTB</i> null mutation associated with microcephaly, early developmental delay, and severe dyskinesia. <i>Neurology</i> , 2016, 86, 877-878.	1.1	25
38	Altered synaptobrevin-II trafficking in neurons expressing a synaptophysin mutation associated with a severe neurodevelopmental disorder. <i>Neurobiology of Disease</i> , 2017, 108, 298-306.	4.4	25
39	TMX2 Is a Crucial Regulator of Cellular Redox State, and Its Dysfunction Causes Severe Brain Developmental Abnormalities. <i>American Journal of Human Genetics</i> , 2019, 105, 1126-1147.	6.2	25
40	A recurrent de novo missense pathogenic variant in <i>SMARCB1</i> causes severe intellectual disability and choroid plexus hyperplasia with resultant hydrocephalus. <i>Genetics in Medicine</i> , 2019, 21, 572-579.	2.4	24
41	The expanding phenotypic spectrum of <i>ARFGEF2</i> gene mutation: Cardiomyopathy and movement disorder. <i>Brain and Development</i> , 2016, 38, 124-127.	1.1	22
42	Advanced genomic testing may aid in counseling of isolated agenesis of the corpus callosum on prenatal ultrasound. <i>Prenatal Diagnosis</i> , 2017, 37, 1191-1197.	2.3	22
43	Heterogeneous clinical phenotypes and cerebral malformations reflected by rotatin cellular dynamics. <i>Brain</i> , 2019, 142, 867-884.	7.6	22
44	Human <i>RAD50</i> deficiency: Confirmation of a distinctive phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1378-1386.	1.2	21
45	The potential diagnostic yield of whole exome sequencing in pregnancies complicated by fetal ultrasound anomalies. <i>Acta Obstetrica Et Gynecologica Scandinavica</i> , 2021, 100, 1106-1115.	2.8	21
46	A standard of care for individuals with <i>PIK3CA</i> -related disorders: An international expert consensus statement. <i>Clinical Genetics</i> , 2022, 101, 32-47.	2.0	21
47	<i>EML1</i> -associated brain overgrowth syndrome with ribbon-like heterotopia. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2019, 181, 627-637.	1.6	17
48	Bi-allelic Loss of Human <i>APC2</i> , Encoding Adenomatous Polyposis Coli Protein 2, Leads to Lissencephaly, Subcortical Heterotopia, and Global Developmental Delay. <i>American Journal of Human Genetics</i> , 2019, 105, 844-853.	6.2	17
49	Heterozygous <i>ANKRD17</i> loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. <i>American Journal of Human Genetics</i> , 2021, 108, 1138-1150.	6.2	17
50	Loss of <i>USP18</i> in microglia induces white matter pathology. <i>Acta Neuropathologica Communications</i> , 2019, 7, 106.	5.2	15
51	Defining the phenotypical spectrum associated with variants in <i>TUBB2A</i> . <i>Journal of Medical Genetics</i> , 2021, 58, 33-40.	3.2	11
52	Progressive leukoencephalopathy impairs neurobehavioral development in sialin-deficient mice. <i>Experimental Neurology</i> , 2017, 291, 106-119.	4.1	10
53	Mutated zinc finger protein of the cerebellum 1 leads to microcephaly, cortical malformation, callosal agenesis, cerebellar dysplasia, tethered cord and scoliosis. <i>European Journal of Medical Genetics</i> , 2018, 61, 783-789.	1.3	10
54	Ehlers Danlos syndrome, kyphoscoliotic type due to Lysyl Hydroxylase 1 deficiency in two children without congenital or early onset kyphoscoliosis. <i>European Journal of Medical Genetics</i> , 2017, 60, 536-540.	1.3	9

#	ARTICLE	IF	CITATIONS
55	Asymmetric polymicrogyria and periventricular nodular heterotopia due to mutation in <i>ARX</i> . American Journal of Medical Genetics, Part A, 2012, 158A, 1472-1476.	1.2	7
56	Cystoid maculopathy is a frequent feature of Cohen syndrome-associated retinopathy. Scientific Reports, 2021, 11, 16412.	3.3	7
57	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
58	Neuro-MIG: A European network on brain malformations. European Journal of Medical Genetics, 2018, 61, 741-743.	1.3	6
59	De novo <i>TRPV4</i> Leu619Pro variant causes a new channelopathy characterised by giant cell lesions of the jaws and skull, skeletal abnormalities and polyneuropathy. Journal of Medical Genetics, 2022, 59, 305-312.	3.2	6
60	Late-onset phenotype associated with a homozygous GJC2 missense mutation in a Turkish family. Parkinsonism and Related Disorders, 2019, 66, 228-231.	2.2	5
61	Mixoploidy combined with aneuploidy in a 13 year-old patient with severe multiple congenital abnormalities and intellectual disability. American Journal of Medical Genetics, Part A, 2018, 176, 492-495.	1.2	3
62	Multidisciplinary interaction and MCD gene discovery. The perspective of the clinical geneticist. European Journal of Paediatric Neurology, 2021, 35, 27-34.	1.6	3
63	Periventricular nodular heterotopia and distal limb deficiency: A recurrent association. American Journal of Medical Genetics, Part A, 2010, 152A, 954-959.	1.2	2
64	A novel family illustrating the mild phenotypic spectrum of TUBB2B variants. European Journal of Paediatric Neurology, 2021, 35, 35-39.	1.6	2
65	Re: Polymicrogyria versus pachygyria in 22q11 microdeletion. American Journal of Medical Genetics, Part A, 2005, 136A, 419-419.	1.2	1
66	Overlapping cortical malformations in patients with pathogenic variants in <i>GRIN1</i> and <i>GRIN2B</i> . Journal of Medical Genetics, 2023, 60, 183-192.	3.2	1
67	[P2.75]: Malformations of cortical development: Genomic analysis by high density microarrays in a large patient cohort. International Journal of Developmental Neuroscience, 2010, 28, 712-713.	1.6	0