

# Nataliya Di Donato

## List of Publications by Year in descending order

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72  
papers

4,156  
citations

201674

27  
h-index

123424

61  
g-index

77  
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77  
docs citations

77  
times ranked

8730  
citing authors

#	ARTICLE	IF	CITATIONS
1	Response on EJPN-S-21-00580. European Journal of Paediatric Neurology, 2022, , .	1.6	0
2	Diagnostic pitfalls in patients with malformations of cortical development. European Journal of Paediatric Neurology, 2022, 37, 123-128.	1.6	1
3	Frameshift mutation S368fs in the gene encoding cytoskeletal $\beta$ -actin leads to ACTB-associated syndromic thrombocytopenia by impairing actin dynamics. European Journal of Cell Biology, 2022, 101, 151216.	3.6	7
4	Monoallelic and biallelic mutations in <i>RELN</i> underlie a graded series of neurodevelopmental disorders. Brain, 2022, 145, 3274-3287.	7.6	6
5	Biallelic UBE4A loss-of-function variants cause intellectual disability and global developmental delay. Genetics in Medicine, 2021, 23, 661-668.	2.4	2
6	Generation of iPSC lines from CPVT patient carrying heterozygous mutation p.A2254V in the ryanodine receptor 2 gene. Stem Cell Research, 2021, 53, 102259.	0.7	0
7	Proximal variants in <i>CCND2</i> associated with microcephaly, short stature, and developmental delay: A case series and review of inverse brain growth phenotypes. American Journal of Medical Genetics, Part A, 2021, 185, 2719-2738.	1.2	14
8	Lissencephaly: Update on diagnostics and clinical management. European Journal of Paediatric Neurology, 2021, 35, 147-152.	1.6	16
9	Congenital hiatal hernia segregating with a duplication in 9q22.31q22.32 in two families. American Journal of Medical Genetics, Part A, 2020, 182, 3040-3047.	1.2	1
10	The clinical-phenotype continuum in DYNC1H1-related disorders—genomic profiling and proposal for a novel classification. Journal of Human Genetics, 2020, 65, 1003-1017.	2.3	30
11	International consensus recommendations on the diagnostic work-up for malformations of cortical development. Nature Reviews Neurology, 2020, 16, 618-635.	10.1	53
12	Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. Neuron, 2020, 106, 404-420.e8.	8.1	121
13	Novel truncating PPM1D mutation in a patient with intellectual disability. European Journal of Medical Genetics, 2019, 62, 70-72.	1.3	10
14	De Novo Variants in TAOK1 Cause Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 105, 213-220.	6.2	36
15	Missense Mutations in NKAP Cause a Disorder of Transcriptional Regulation Characterized by Marfanoid Habitus and Cognitive Impairment. American Journal of Human Genetics, 2019, 105, 987-995.	6.2	11
16	First confirmatory study on PTPRQ as an autosomal dominant non-syndromic hearing loss gene. Journal of Translational Medicine, 2019, 17, 351.	4.4	10
17	Bi-allelic Loss of Human APC2, Encoding Adenomatous Polyposis Coli Protein 2, Leads to Lissencephaly, Subcortical Heterotopia, and Global Developmental Delay. American Journal of Human Genetics, 2019, 105, 844-853.	6.2	17
18	Parental mosaicism in epilepsies due to alleged de novo variants. Epilepsia, 2019, 60, e63-e66.	5.1	29

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19	De Novo Variants in MAPK8IP3 Cause Intellectual Disability with Variable Brain Anomalies. American Journal of Human Genetics, 2019, 104, 203-212.	6.2	44
20	PUF60-SCRIB fusion transcript in a patient with 8q24.3 microdeletion and atypical Verheij syndrome. European Journal of Medical Genetics, 2019, 62, 103587.	1.8	7
21	Analysis of 17 genes detects mutations in 81% of 811 patients with lissencephaly. Genetics in Medicine, 2018, 20, 1354-1364.	2.4	92
22	Tubulinopathies continued: refining the phenotypic spectrum associated with variants in TUBG1. European Journal of Human Genetics, 2018, 26, 1132-1142.	2.8	30
23	A C-terminal nonsense mutation links PTPRQ with autosomal-dominant hearing loss, DFNA73. Genetics in Medicine, 2018, 20, 614-621.	2.4	21
24	Variable clinical phenotype in two siblings with Aicardi-Goutières syndrome type 6 and a novel mutation in the ADAR gene. European Journal of Paediatric Neurology, 2018, 22, 186-189.	1.6	18
25	Tagungsbericht Syndromtag 2018. Medizinische Genetik, 2018, 30, 533-540.	0.2	0
26	Novel VPS33B mutation in a patient with autosomal recessive keratoderma ichthyosis deafness syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2862-2866.	1.2	8
27	Variants in exons 5 and 6 of ACTB cause syndromic thrombocytopenia. Nature Communications, 2018, 9, 4250.	12.8	38
28	Diagnostic value of partial exome sequencing in developmental disorders. PLoS ONE, 2018, 13, e0201041.	2.5	36
29	Further delineation of Malan syndrome. Human Mutation, 2018, 39, 1226-1237.	2.5	42
30	Constitutional de novo and postzygotic mutations in isolated cases of cerebral cavernous malformations. Molecular Genetics & Genomic Medicine, 2017, 5, 21-27.	1.2	4
31	Heterozygous truncation mutations of the <i>SMC1A</i> gene cause a severe early onset epilepsy with cluster seizures in females: Detailed phenotyping of 10 new cases. Epilepsia, 2017, 58, 565-575.	5.1	35
32	Functional monosomy of 6q27qter and functional disomy of Xpterp22.11 due to X;6 translocation with an atypical X inactivation pattern. American Journal of Medical Genetics, Part A, 2017, 173, 1334-1341.	1.2	5
33	Lissencephaly: Expanded imaging and clinical classification. American Journal of Medical Genetics, Part A, 2017, 173, 1473-1488.	1.2	104
34	Pierpont syndrome: report of a new patient. Clinical Dysmorphology, 2017, 26, 205-208.	0.3	12
35	<i>GRIN2B</i> encephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. Journal of Medical Genetics, 2017, 54, 460-470.	3.2	190
36	Central nervous system anomalies in two females with Borjeson-Forsman-Lehmann syndrome. Epilepsy and Behavior, 2017, 69, 104-109.	1.7	7

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37	Novel PRPS1 gain-of-function mutation in a patient with congenital hyperuricemia and facial anomalies. , 2017, 173, 2736-2742.		11
38	Skewed X-inactivation in a family with <i>DLG3</i> associated X-linked intellectual disability. American Journal of Medical Genetics, Part A, 2017, 173, 2545-2550.	1.2	12
39	<i>FOXP1</i> -related intellectual disability syndrome: a recognisable entity. Journal of Medical Genetics, 2017, 54, 613-623.	3.2	48
40	Overlapping SETBP1 gain-of-function mutations in Schinzel-Giedion syndrome and hematologic malignancies. PLoS Genetics, 2017, 13, e1006683.	3.5	35
41	Identification and Functional Testing of ERCC2 Mutations in a Multi-national Cohort of Patients with Familial Breast- and Ovarian Cancer. PLoS Genetics, 2016, 12, e1006248.	3.5	22
42	Mutation Update for Kabuki Syndrome Genes <i>KMT2D</i> and <i>KDM6A</i> and Further Delineation of X-Linked Kabuki Syndrome Subtype 2. Human Mutation, 2016, 37, 847-864.	2.5	134
43	Update on the <i>ACTG1</i> associated Baraitser-Winter cerebrofrontofacial syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2644-2651.	1.2	28
44	Identification and Characterization of a Novel Constitutional PIK3CA Mutation in a Child Lacking the Typical Segmental Overgrowth of PIK3CA-Related Overgrowth Spectrum. Human Mutation, 2016, 37, 242-245.	2.5	11
45	Novel Mutation in the DKC1 Gene: Neonatal Hoyeraal-Hreidarsson Syndrome As a Rare Differential Diagnosis in Pontocerebellar Hypoplasia, Primary Microcephaly, and Progressive Bone Marrow Failure. Neuropediatrics, 2016, 47, 182-186.	0.6	8
46	Ready to clone: CNV detection and breakpoint fine-mapping in breast and ovarian cancer susceptibility genes by high-resolution array CGH. Breast Cancer Research and Treatment, 2016, 159, 585-590.	2.5	15
47	Tentative clinical diagnosis of Lujan-Fryns syndrome? A conglomeration of different genetic entities?. American Journal of Medical Genetics, Part A, 2016, 170, 94-102.	1.2	11
48	Recurrent de novo BICD2 mutation associated with arthrogryposis multiplex congenita and bilateral perisylvian polymicrogyria. Neuromuscular Disorders, 2016, 26, 744-748.	0.6	44
49	Mutations in CRADD Result in Reduced Caspase-2-Mediated Neuronal Apoptosis and Cause Megalencephaly with a Rare Lissencephaly Variant. American Journal of Human Genetics, 2016, 99, 1117-1129.	6.2	50
50	Mutations in <i>EXOSC2</i> are associated with a novel syndrome characterised by retinitis pigmentosa, progressive hearing loss, premature ageing, short stature, mild intellectual disability and distinctive gestalt. Journal of Medical Genetics, 2016, 53, 419-425.	3.2	69
51	Mammalian cadherins DCHS1-FAT4 affect functional cerebral architecture. Brain Structure and Function, 2016, 221, 2487-2491.	2.3	12
52	A child with Li-Fraumeni syndrome: Modes to inactivate the second allele of <i>TP53</i> in three different malignancies. Pediatric Blood and Cancer, 2015, 62, 1481-1484.	1.5	22
53	6q22.33 microdeletion in a family with intellectual disability, variable major anomalies, and behavioral abnormalities. American Journal of Medical Genetics, Part A, 2015, 167, 2800-2807.	1.2	3
54	Next-generation sequencing in X-linked intellectual disability. European Journal of Human Genetics, 2015, 23, 1513-1518.	2.8	112

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55	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. <i>American Journal of Human Genetics</i> , 2015, 97, 343-352.	6.2	230
56	Phenotype and genotype in 103 patients with tricho-rhino-phalangeal syndrome. <i>European Journal of Medical Genetics</i> , 2015, 58, 279-292.	1.3	62
57	Microduplications of 3p26.3p26.2 containing CRBN gene in patients with intellectual disability and behavior abnormalities. <i>European Journal of Medical Genetics</i> , 2015, 58, 319-323.	1.3	9
58	Baraitserâ€“Winter cerebrofrontofacial syndrome: delineation of the spectrum in 42 cases. <i>European Journal of Human Genetics</i> , 2015, 23, 292-301.	2.8	115
59	Abstract LB-044: Germline mutations in patients with hereditary breast and ovarian cancer establish ERCC2 as a cancer susceptibility gene. , 2015, , .		0
60	Distinct phenotype of PHF6 deletions in females. <i>European Journal of Medical Genetics</i> , 2014, 57, 85-89.	1.3	17
61	Targeted and Genomewide NGS Data Disqualify Mutations in<i>MYO1A</i>, the â€œ<i>DFNA48</i> Geneâ€™, as a Cause of Deafness. <i>Human Mutation</i> , 2014, 35, 565-570.	2.5	38
62	Severe forms of Baraitserâ€“Winter syndrome are caused by ACTB mutations rather than ACTG1 mutations. <i>European Journal of Human Genetics</i> , 2014, 22, 179-183.	2.8	67
63	Females with de novo aberrations in <i>PHF6</i>: Clinical overlap of Borjesonâ€“Forssmanâ€“Lehmann with Coffinâ€“Siris syndrome. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2014, 166, 290-301.	1.6	27
64	Clinical phenotypes of MAGEL2 mutations and deletions. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 40.	2.7	28
65	Loss-of-function HDAC8 mutations cause a phenotypic spectrum of Cornelia de Lange syndrome-like features, ocular hypertelorism, large fontanelle and X-linked inheritance. <i>Human Molecular Genetics</i> , 2014, 23, 2888-2900.	2.9	120
66	Interstitial deletion 1p36.32 in two brothers with a distinct phenotype â€“ Overgrowth, macrocephaly and nearly normal intellectual function. <i>European Journal of Medical Genetics</i> , 2014, 57, 494-497.	1.3	6
67	Mutations in genes encoding the cadherin receptor-ligand pair DCHS1 and FAT4 disrupt cerebral cortical development. <i>Nature Genetics</i> , 2013, 45, 1300-1308.	21.4	247
68	Partial deletion of GLRB and GRIA2 in a patient with intellectual disability. <i>European Journal of Human Genetics</i> , 2013, 21, 112-114.	2.8	22
69	Range of genetic mutations associated with severe non-syndromic sporadic intellectual disability: an exome sequencing study. <i>Lancet, The</i> , 2012, 380, 1674-1682.	13.7	940
70	De novo microdeletions of chromosome 6q14.1-q14.3 and 6q12.1-q14.1 in two patients with intellectual disability - further delineation of the 6q14 microdeletion syndrome and review of the literature. <i>European Journal of Medical Genetics</i> , 2012, 55, 490-497.	1.3	20
71	HDAC8 mutations in Cornelia de Lange syndrome affect the cohesin acetylation cycle. <i>Nature</i> , 2012, 489, 313-317.	27.8	488
72	Macrocephaly, obesity, mental (intellectual) disability, and ocular abnormalities: Alternative definition and further delineation of MOMO syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 2857-2862.	1.2	7