Nataliya Di Donato

List of Publications by Year
in descending order

72	4,156	201674 27	123424 61
papers	citations	h-index	g-index

77	77	77	8730
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Range of genetic mutations associated with severe non-syndromic sporadic intellectual disability: an exome sequencing study. Lancet, The, 2012, 380, 1674-1682.	13.7	940
2	HDAC8 mutations in Cornelia de Lange syndrome affect the cohesin acetylation cycle. Nature, 2012, 489, 313-317.	27.8	488
3	Mutations in genes encoding the cadherin receptor-ligand pair DCHS1 and FAT4 disrupt cerebral cortical development. Nature Genetics, 2013, 45, 1300-1308.	21.4	247
4	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. American Journal of Human Genetics, 2015, 97, 343-352.	6.2	230
5	GRIN2Bencephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. Journal of Medical Genetics, 2017, 54, 460-470.	3.2	190
6	Mutation Update for Kabuki Syndrome GenesKMT2DandKDM6Aand Further Delineation of X-Linked Kabuki Syndrome Subtype 2. Human Mutation, 2016, 37, 847-864.	2.5	134
7	Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. Neuron, 2020, 106, 404-420.e8.	8.1	121
8	Loss-of-function HDAC8 mutations cause a phenotypic spectrum of Cornelia de Lange syndrome-like features, ocular hypertelorism, large fontanelle and X-linked inheritance. Human Molecular Genetics, 2014, 23, 2888-2900.	2.9	120
9	Baraitserâ€“Winter cerebrofrontofacial syndrome: delineation of the spectrum in 42 cases. European Journal of Human Genetics, 2015, 23, 292-301.	2.8	115
10	Next-generation sequencing in X-linked intellectual disability. European Journal of Human Genetics, 2015, 23, 1513-1518.	2.8	112
11	Lissencephaly: Expanded imaging and clinical classification. American Journal of Medical Genetics, Part A, 2017, 173, 1473-1488.	1.2	104
12	Analysis of 17 genes detects mutations in 81% of 811 patients with lissencephaly. Genetics in Medicine, 2018, 20, 1354-1364.	2.4	92
13	Mutations inEXOSC2are 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
14	Severe forms of Baraitserâ€“Winter syndrome are caused by ACTB mutations rather than ACTG1 mutations. European Journal of Human Genetics, 2014, 22, 179-183.	2.8	67
15	Phenotype and genotype in 103 patients with tricho-rhino-phalangeal syndrome. European Journal of Medical Genetics, 2015, 58, 279-292.	1.3	62
16	International consensus recommendations on the diagnostic work-up for malformations of cortical development. Nature Reviews Neurology, 2020, 16, 618-635.	10.1	53
17	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
18	FOXP1-related intellectual disability syndrome: a recognisable entity. Journal of Medical Genetics, 2017, 54, 613-623.	3.2	48

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19	Recurrent de novo BICD2 mutation associated with arthrogryposis multiplex congenita and bilateral perisylvian polymicrogyria. Neuromuscular Disorders, 2016, 26, 744-748.	0.6	44
20	De Novo Variants in MAPK8IP3 Cause Intellectual Disability with Variable Brain Anomalies. American Journal of Human Genetics, 2019, 104, 203-212.	6.2	44
21	Further delineation of Malan syndrome. Human Mutation, 2018, 39, 1226-1237.	2.5	42
22	Targeted and Genomewide NGS Data Disqualify Mutations inMYO1A, the â€œDFNA48Geneâ€, as a Cause of Deafness. Human Mutation, 2014, 35, 565-570.	2.5	38
23	Variants in exons 5 and 6 of ACTB cause syndromic thrombocytopenia. Nature Communications, 2018, 9, 4250.	12.8	38
24	Diagnostic value of partial exome sequencing in developmental disorders. PLoS ONE, 2018, 13, e0201041.	2.5	36
25	De Novo Variants in TAOK1 Cause Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 105, 213-220.	6.2	36
26	Heterozygous truncation mutations of the <scp>SMC</scp>1A 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
27	Overlapping SETBP1 gain-of-function mutations in Schinzel-Giedion syndrome and hematologic malignancies. PLoS Genetics, 2017, 13, e1006683.	3.5	35
28	Tubulinopathies continued: refining the phenotypic spectrum associated with variants in TUBG1. European Journal of Human Genetics, 2018, 26, 1132-1142.	2.8	30
29	The clinical-phenotype continuum in DYNC1H1-related disordersâ€”genomic profiling and proposal for a novel classification. Journal of Human Genetics, 2020, 65, 1003-1017.	2.3	30
30	Parental mosaicism in epilepsies due to alleged de novo variants. Epilepsia, 2019, 60, e63-e66.	5.1	29
31	Clinical phenotypes of MAGEL2 mutations and deletions. Orphanet Journal of Rare Diseases, 2014, 9, 40.	2.7	28
32	Update on the ACTG1â€associated Baraitserâ€“Winter cerebrofrontofacial syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2644-2651.	1.2	28
33	Females with de novo aberrations in PHF6: Clinical overlap of Borjesonâ€“Forssmanâ€“Lehmann with Coffinâ€“Siris syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 290-301.	1.6	27
34	Partial deletion of GLRB and GRIA2 in a patient with intellectual disability. European Journal of Human Genetics, 2013, 21, 112-114.	2.8	22
35	A child with Liâ€“Fraumeni syndrome: Modes to inactivate the second allele of TP53 in three different malignancies. Pediatric Blood and Cancer, 2015, 62, 1481-1484.	1.5	22
36	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

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37	A C-terminal nonsense mutation links PTPRQ with autosomal-dominant hearing loss, DFNA73. Genetics in Medicine, 2018, 20, 614-621.	2.4	21
38	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. European Journal of Medical Genetics, 2012, 55, 490-497.	1.3	20
39	Variable clinical phenotype in two siblings with Aicardi-GoutiÃ¨res syndrome type 6 and a novel mutation in the ADAR gene. European Journal of Paediatric Neurology, 2018, 22, 186-189.	1.6	18
40	Distinct phenotype of PHF6 deletions in females. European Journal of Medical Genetics, 2014, 57, 85-89.	1.3	17
41	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
42	Lissencephaly: Update on diagnostics and clinical management. European Journal of Paediatric Neurology, 2021, 35, 147-152.	1.6	16
43	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
44	Proximal variants in <scp>CCND2</scp> 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
45	Mammalian cadherins DCHS1-FAT4 affect functional cerebral architecture. Brain Structure and Function, 2016, 221, 2487-2491.	2.3	12
46	Pierpont syndrome: report of a new patient. Clinical Dysmorphology, 2017, 26, 205-208.	0.3	12
47	Skewed Xâ€inactivation in a family with DLG3â€associated Xâ€linked intellectual disability. American Journal of Medical Genetics, Part A, 2017, 173, 2545-2550.	1.2	12
48	Identification and Characterization of a Novel ConstitutionalPIK3CAMutation in a Child Lacking the Typical Segmental Overgrowth of â€œPIK3CA-Related Overgrowth Spectrumâ€• Human Mutation, 2016, 37, 242-245.	2.5	11
49	Tentative clinical diagnosis of Lujanâ€Fryns syndromeâ€”A conglomeration of different genetic entities?. American Journal of Medical Genetics, Part A, 2016, 170, 94-102.	1.2	11
50	Novel PRPS1 gain-of-function mutation in a patient with congenital hyperuricemia and facial anomalies. , 2017, 173, 2736-2742.		11
51	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
52	Novel truncating PPM1D mutation in a patient with intellectual disability. European Journal of Medical Genetics, 2019, 62, 70-72.	1.3	10
53	First confirmatory study on PTPRQ as an autosomal dominant non-syndromic hearing loss gene. Journal of Translational Medicine, 2019, 17, 351.	4.4	10
54	Microduplications of 3p26.3p26.2 containing CRBN gene in patients with intellectual disability and behavior abnormalities. European Journal of Medical Genetics, 2015, 58, 319-323.	1.3	9

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55	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
56	Novel VPS33B mutation in a patient with autosomal recessive keratodermaâ€ichthyosisâ€deafness syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2862-2866.	1.2	8
57	Macrocephaly, obesity, mental (intellectual) disability, and ocular abnormalities: Alternative definition and further delineation of MOMO syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 2857-2862.	1.2	7
58	Central nervous system anomalies in two females with Borjeson-Forssman-Lehmann syndrome. Epilepsy and Behavior, 2017, 69, 104-109.	1.7	7
59	PUF60-SCRIB fusion transcript in a patient with 8q24.3 microdeletion and atypical Verheij syndrome. European Journal of Medical Genetics, 2019, 62, 103587.	1.3	7
60	Frameshift mutation S368fs in the gene encoding cytoskeletal Î²-actin leads to ACTB-associated syndromic thrombocytopenia by impairing actin dynamics. European Journal of Cell Biology, 2022, 101, 151216.	3.6	7
61	Interstitial deletion 1p36.32 in two brothers with a distinct phenotype â€“ Overgrowth, macrocephaly and nearly normal intellectual function. European Journal of Medical Genetics, 2014, 57, 494-497.	1.3	6
62	Monoallelic and biallelic mutations inRELNunderlie a graded series of neurodevelopmental disorders. Brain, 2022, 145, 3274-3287.	7.6	6
63	Functional monosomy of 6q27â€qter and functional disomy of Xpterâ€p22.11 due to X;6 translocation with an atypical Xâ€inactivation pattern. American Journal of Medical Genetics, Part A, 2017, 173, 1334-1341.	1.2	5
64	Constitutional de novo and postzygotic mutations in isolated cases of cerebral cavernous malformations. Molecular Genetics & Genomic Medicine, 2017, 5, 21-27.	1.2	4
65	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
66	Biallelic UBE4A loss-of-function variants cause intellectual disability and global developmental delay. Genetics in Medicine, 2021, 23, 661-668.	2.4	2
67	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
68	Diagnostic pitfalls in patients with malformations of cortical development. European Journal of Paediatric Neurology, 2022, 37, 123-128.	1.6	1
69	Tagungsbericht Syndromtag 2018. Medizinische Genetik, 2018, 30, 533-540.	0.2	0
70	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
71	Abstract LB-044: Germline mutations in patients with hereditary breast and ovarian cancer establish ERCC2 as a cancer susceptibility gene. , 2015, , .		0
72	Response on EJPN-S-21-00580. European Journal of Paediatric Neurology, 2022, , .	1.6	0