Maria Lisa Dentici

List of Publications by Year
in descending order

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78 papers	2,567 citations	236925 25 h-index	214800 47 g-index

78 all docs	78 docs citations	78 times ranked	5123 citing authors

#	Article	IF	Citations
1	A restricted spectrum of NRAS mutations causes Noonan syndrome. Nature Genetics, 2010, 42, 27-29.	21.4	271
2	GermlineBRAFmutations in Noonan, LEOPARD, and cardiofaciocutaneous syndromes: Molecular diversity and associated phenotypic spectrum. Human Mutation, 2009, 30, 695-702.	2.5	251
3	Mutations in KCNH1 and ATP6V1B2 cause Zimmermann-Laband syndrome. Nature Genetics, 2015, 47, 661-667.	21.4	177
4	Familial transposition of the great arteries caused by multiple mutations in laterality genes. Heart, 2010, 96, 673-677.	2.9	126
5	SOS1 mutations in Noonan syndrome: molecular spectrum, structural insights on pathogenic effects, and genotype-phenotype correlations. Human Mutation, 2011, 32, 760-772.	2.5	97
6	BRF1 mutations alter RNA polymerase IIIâ€“dependent transcription and cause neurodevelopmental anomalies. Genome Research, 2015, 25, 155-166.	5.5	85
7	Deficiency for the Ubiquitin Ligase UBE3B in a Blepharophimosis-Ptosis-Intellectual-Disability Syndrome. American Journal of Human Genetics, 2012, 91, 998-1010.	6.2	82
8	A Restricted Spectrum of Mutations in the SMAD4 Tumor-Suppressor Gene Underlies Myhre Syndrome. American Journal of Human Genetics, 2012, 90, 161-169.	6.2	77
9	Spectrum of MEK1 and MEK2 gene mutations in cardio-facio-cutaneous syndrome and genotypeâ€“phenotype correlations. European Journal of Human Genetics, 2009, 17, 733-740.	2.8	74
10	RASopathies: Clinical Diagnosis in the First Year of Life. Molecular Syndromology, 2010, 1, 282-289.	0.8	73
11	Mutations in KCNK4 that Affect Gating Cause a Recognizable Neurodevelopmental Syndrome. American Journal of Human Genetics, 2018, 103, 621-630.	6.2	73
12	New mutations inZFPM2/FOG2gene in tetralogy of Fallot and double outlet right ventricle. Clinical Genetics, 2011, 80, 184-190.	2.0	69
13	Kabuki syndrome: clinical and molecular diagnosis in the first year of life. Archives of Disease in Childhood, 2015, 100, 158-164.	1.9	69
14	Biallelic Mutations in TBCD , Encoding the Tubulin Folding Cofactor D, Perturb Microtubule Dynamics and Cause Early-Onset Encephalopathy. American Journal of Human Genetics, 2016, 99, 962-973.	6.2	66
15	Congenital heart defects in molecularly proven Kabuki syndrome patients. American Journal of Medical Genetics, Part A, 2017, 173, 2912-2922.	1.2	60
16	Diagnosis of Noonan syndrome and related disorders using target next generation sequencing. BMC Medical Genetics, 2014, 15, 14.	2.1	59
17	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	6.2	48
18	Mutation update for the SATB2 gene. Human Mutation, 2019, 40, 1013-1029.	2.5	38

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19	SCUBE3 loss-of-function causes a recognizable recessive developmental disorder due to defective bone morphogenetic protein signaling. American Journal of Human Genetics, 2021, 108, 115-133.	6.2	37
20	De Novo Variants Disturbing the Transactivation Capacity of POU3F3 Cause a Characteristic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 105, 403-412.	6.2	35
21	Epilepsy in KCNH1â€related syndromes. Epileptic Disorders, 2016, 18, 123-136.	1.3	34
22	Syndromic nonâ€compaction of the left ventricle: associated chromosomal anomalies. Clinical Genetics, 2013, 84, 362-367.	2.0	30
23	Clinical delineation, sex differences, and genotypeâ€“phenotype correlation in pathogenic KDM6A variants causing X-linked Kabuki syndrome type 2. Genetics in Medicine, 2021, 23, 1202-1210.	2.4	30
24	Expanding the clinical and molecular spectrum of PRMT7 mutations: 3 additional patients and review. Clinical Genetics, 2018, 93, 675-681.	2.0	28
25	Identification of novel and hotspot mutations in the channel domain of ITPR1 in two patients with Gillespie syndrome. Gene, 2017, 628, 141-145.	2.2	27
26	KBG syndrome: Common and uncommon clinical features based on 31 new patients. American Journal of Medical Genetics, Part A, 2020, 182, 1073-1083.	1.2	27
27	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. Genetics in Medicine, 2020, 22, 1338-1347.	2.4	25
28	Expanding the phenotypic spectrum of truncating POGZ mutations: Association with CNS malformations, skeletal abnormalities, and distinctive facial dysmorphism. American Journal of Medical Genetics, Part A, 2017, 173, 1965-1969.	1.2	23
29	Clinical spectrum of Kabukiâ€like syndrome caused by <scp>HNRNPK</scp> haploinsufficiency. Clinical Genetics, 2018, 93, 401-407.	2.0	23
30	Biallelic mutations in <scp>DYNC2LI1</scp> are a rare cause of Ellisâ€van Creveld syndrome. Clinical Genetics, 2018, 93, 632-639.	2.0	23
31	Atrioventricular canal defect in patients with RASopathies. European Journal of Human Genetics, 2013, 21, 200-204.	2.8	22
32	Expanding the spectrum of megalencephalic leukoencephalopathy with subcortical cysts in two patients with GLIALCAM mutations. Neurogenetics, 2014, 15, 41-48.	1.4	22
33	Atrioventricular canal defect and genetic syndromes: The unifying role of sonic hedgehog. Clinical Genetics, 2019, 95, 268-276.	2.0	20
34	TUBB Variants Underlying Different Phenotypes Result in Altered Vesicle Trafficking and Microtubule Dynamics. International Journal of Molecular Sciences, 2020, 21, 1385.	4.1	20
35	POGZâ€related epilepsy: Case report and review of the literature. American Journal of Medical Genetics, Part A, 2019, 179, 1631-1636.	1.2	19
36	Expanding the clinical spectrum associated with PACS2 mutations. Clinical Genetics, 2019, 95, 525-531.	2.0	18

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37	GRIA3 missense mutation is cause of an x-linked developmental and epileptic encephalopathy. Seizure: the Journal of the British Epilepsy Association, 2020, 82, 1-6.	2.0	18
38	Infantile-Onset Syndromic Cerebellar Ataxia and CACNA1G Mutations. Pediatric Neurology, 2020, 104, 40-45.	2.1	17
39	Delineating the psychiatric and behavioral phenotype of recurrent 2q13 deletions and duplications. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 397-405.	1.7	16
40	TARP syndrome: Long-term survival, anatomic patterns of congenital heart defects, differential diagnosis and pathogenetic considerations. European Journal of Medical Genetics, 2019, 62, 103534.	1.3	16
41	New patients with Temple syndrome caused by 14q32 deletion: Genotypeâ€phenotype correlations and risk of thyroid cancer. American Journal of Medical Genetics, Part A, 2016, 170, 162-169.	1.2	15
42	TUBB3 E410K syndrome: Case report and review of the clinical spectrum of TUBB3 mutations. American Journal of Medical Genetics, Part A, 2020, 182, 1977-1984.	1.2	15
43	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
44	The difficult nosology of blepharophimosisâ€“mental retardation syndromes: Report on two siblings. American Journal of Medical Genetics, Part A, 2011, 155, 459-465.	1.2	13
45	Confirmation of BRD4 haploinsufficiency role in Cornelia de Langeâ€“like phenotype and delineation of a 19p13.12p13.11 gene contiguous syndrome. Annals of Human Genetics, 2019, 83, 100-109.	0.8	13
46	9q34.3 microduplications lead to neurodevelopmental disorders through EHMT1 overexpression. Neurogenetics, 2019, 20, 145-154.	1.4	12
47	Co-occurrence of mutations in KIF7 and KIAA0556 in Joubert syndrome with ocular coloboma, pituitary malformation and growth hormone deficiency: a case report and literature review. BMC Pediatrics, 2020, 20, 120.	1.7	12
48	Intrafamiliar clinical variability of circumferential skin creases Kunze type caused by a novel heterozygous mutation of Nâ€terminal TUBB gene. Clinical Genetics, 2018, 93, 1223-1228.	2.0	11
49	<scp>TSPEAR</scp> variants are primarily associated with ectodermal dysplasia and tooth agenesis but not hearing loss: A novel cohort study. American Journal of Medical Genetics, Part A, 2021, 185, 2417-2433.	1.2	10
50	JAG1 Mutation in a patient with deletion 22q11.2 syndrome and tetralogy of Fallot. American Journal of Medical Genetics, Part A, 2013, 161, 3133-3136.	1.2	9
51	Congenital heart defects in the recurrent 2q13 deletion syndrome. European Journal of Medical Genetics, 2022, 65, 104381.	1.3	9
52	<scp>PPP1R21</scp>â€related syndromic intellectual disability: Report of an adult patient and review. American Journal of Medical Genetics, Part A, 2020, 182, 3014-3022.	1.2	8
53	Vascular Birthmarks as a Clue for Complex and Syndromic Vascular Anomalies. Frontiers in Pediatrics, 2021, 9, 730393.	1.9	8
54	A 6-year-old child with Fryns syndrome: Further delineation of the natural history of the condition in survivors. European Journal of Medical Genetics, 2009, 52, 421-425.	1.3	7

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55	CantÃº syndrome versus Zimmermann-Laband syndrome: Report of nine individuals with ABCC9 variants. European Journal of Medical Genetics, 2020, 63, 103996.	1.3	7
56	Atypical 7q11.23 deletions excluding <scp>ELN</scp> gene result in <scp>Williamsâ€“Beuren</scp> syndrome craniofacial features and neurocognitive profile. American Journal of Medical Genetics, Part A, 2021, 185, 242-249.	1.2	7
57	Association of DiGeorge anomaly and caudal dysplasia sequence in a neonate born to a diabetic mother. Cardiology in the Young, 2013, 23, 14-17.	0.8	6
58	CHARGE syndrome due to deletion of region upstream of CHD7 gene START codon. BMC Medical Genetics, 2015, 16, 78.	2.1	6
59	7q11.23 Microduplication Syndrome: Clinical and Neurobehavioral Profiling. Brain Sciences, 2020, 10, 839.	2.3	6
60	A new 1p36.13â€1p36.12 microdeletion syndrome characterized by learning disability, behavioral abnormalities, and ptosis. Clinical Genetics, 2020, 97, 927-932.	2.0	6
61	Biallelic variants in <scp>ZNF142</scp> lead to a syndromic neurodevelopmental disorder. Clinical Genetics, 2022, 102, 98-109.	2.0	6
62	Biallelic variants in ZNF526 cause a severe neurodevelopmental disorder with microcephaly, bilateral cataract, epilepsy and simplified gyration. Journal of Medical Genetics, 2021, , jmedgenet-2020-107430.	3.2	5
63	Congenital heart defects in molecularly confirmed <scp>KBG</scp> syndrome patients. American Journal of Medical Genetics, Part A, 2022, 188, 1149-1159.	1.2	5
64	Hypopigmented skin patches in 17q21.31 microdeletion syndrome. Clinical Dysmorphology, 2014, 23, 32-34.	0.3	4
65	Unclassifiable pattern of hypopigmentation in a patient with mosaic partial 12p tetrasomy without Pallisterâ€“Killian syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 1943-1946.	1.2	4
66	Familial aggregation of â€œapple peelâ€•intestinal atresia and cardiac leftâ€sided obstructive lesions: A possible causal relationship with NOTCH1 gene mutations. American Journal of Medical Genetics, Part A, 2019, 179, 1570-1574.	1.2	4
67	Sprengel anomaly in deletion 22q11.2 (DiGeorge/Veloâ€“Cardioâ€“Facial) syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 661-664.	1.2	3
68	Ectodermal Dysplasia-Syndactyly Syndrome with Toe-Only Minimal Syndactyly Due to a Novel Mutation in NECTIN4: A Case Report and Literature Review. Genes, 2021, 12, 748.	2.4	3
69	First Report of Low-Rate Mosaicism for 20q11.21q12 Deletion and Delineation of the Associated Disorder. Cytogenetic and Genome Research, 2018, 156, 87-94.	1.1	2
70	Microcephalic osteodysplastic primordial dwarfism type II and pachygyria: Morphometric analysis in a 2â€yearâ€old girl. American Journal of Medical Genetics, Part A, 2020, 182, 2372-2376.	1.2	2
71	Cognitive and Adaptive Characterization of Children and Adolescents with KBG Syndrome: An Explorative Study. Journal of Clinical Medicine, 2021, 10, 1523.	2.4	2
72	Expanding the novel <scp> MAPKAPK5 </scp> â€“related developmental disorderâ€™s genotypeâ€phenotype correlation: patient report and 19 months followâ€up. Clinical Genetics, 2022, , .	2.0	2

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73	Genotypeâ€“phenotype spectrum and correlations in <scp>Xiaâ€Gibbs</scp> syndrome: Report of five novel cases and literature review. Birth Defects Research, 0, , .	1.5	2
74	RDDR: a dysmorphology diagnostic network for newborns in central Italy. Journal of Maternal-Fetal and Neonatal Medicine, 2012, 25, 113-115.	1.5	1
75	Interstitial 10q21.1q23.31 Duplication due to Meiotic Recombination of a Paternal Balanced Complex Rearrangement: Cytogenetic and Molecular Characterization. Cytogenetic and Genome Research, 2017, 151, 179-185.	1.1	1
76	A Child with Diminished Linear Growth and Waddling Gait. Journal of Pediatrics, 2018, 201, 297-297.e1.	1.8	1
77	Homozygous HESX1 and COL1A1 Gene Variants in a Boy with Growth Hormone Deficiency and Early Onset Osteoporosis. International Journal of Molecular Sciences, 2021, 22, 750.	4.1	1
78	A Comparison of Adaptive Functioning Between Children With Duplication 7 Syndrome and Williams-Beuren Syndrome: A Pilot Investigation. Frontiers in Psychiatry, 2022, 13, .	2.6	0