Gerarda Cappuccio

List of Publications by Year
in descending order

77 papers	1,194 citations	471509 17 h-index	552781 26 g-index
82 all docs	82 docs citations	82 times ranked	2511 citing authors

#	Article	IF	Citations
1	Genotypes and phenotypes heterogeneity in PIK3CA-related overgrowth spectrum and overlapping conditions: 150 novel patients and systematic review of 1007 patients with PIK3CA pathogenetic variants. Journal of Medical Genetics, 2023, 60, 163-173.	3.2	15
2	Recurrent de novo missense variants in GNB2 can cause syndromic intellectual disability. Journal of Medical Genetics, 2022, 59, 511-516.	3.2	4
3	Clinical heterogeneity of Kabuki syndrome in a cohort of Italian patients and review of the literature. European Journal of Pediatrics, 2022, 181, 171-187.	2.7	13
4	Long-Term Efficacy of T3 Analogue Triac in Children and Adults With MCT8 Deficiency: A Real-Life Retrospective Cohort Study. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e1136-e1147.	3.6	15
5	Epilepsy in KAT6A syndrome: Description of two individuals and revision of the literature. European Journal of Medical Genetics, 2022, 65, 104380.	1.3	5
6	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders. Human Genetics and Genomics Advances, 2022, 3, 100075.	1.7	42
7	Expanded cardiovascular phenotype of Myhre syndrome includes tetralogy of Fallot suggesting a role for <scp>SMAD4</scp> in human neural crest defects. American Journal of Medical Genetics, Part A, 2022, 188, 1384-1395.	1.2	2
8	De Novo ATP1A1 Variants in an Early-Onset Complex Neurodevelopmental Syndrome. Neurology, 2022, 98, 440-445.	1.1	5
9	Expanding the phenotype of <scp>HNRNPU</scp>â€related neurodevelopmental disorder with emphasis on seizure phenotype and review of literature. American Journal of Medical Genetics, Part A, 2022, 188, 1497-1514.	1.2	6
10	Mild neurological phenotype in a family carrying a novel N-terminal null GRIN2A variant. European Journal of Medical Genetics, 2022, 65, 104500.	1.3	1
11	Biallelic variants in <scp> CENPF </scp> causing a phenotype distinct from StrÃ¸mme syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2022, , .	1.6	3
12	Clinical and Functional Consequences of C-Terminal Variants in MCT8: A Case Series. Journal of Clinical Endocrinology and Metabolism, 2021, 106, 539-553.	3.6	4
13	Milder presentation of TELO2-related syndrome in two sisters homozygous for the p.Arg609His pathogenic variant. European Journal of Medical Genetics, 2021, 64, 104116.	1.3	5
14	FBXO28 causes developmental and epileptic encephalopathy with profound intellectual disability. Epilepsia, 2021, 62, e13-e21.	5.1	8
15	DNA methylation epi-signature is associated with two molecularly and phenotypically distinct clinical subtypes of Phelan-McDermid syndrome. Clinical Epigenetics, 2021, 13, 2.	4.1	27
16	Peculiar footprints in a child with agenesis of corpus callosum. Journal of Paediatrics and Child Health, 2021, 57, 450-451.	0.8	0
17	Biâ€allelic KARS1 pathogenic variants affecting functions of cytosolic and mitochondrial isoforms are associated with a progressive and multisystem disease. Human Mutation, 2021, 42, 745-761.	2.5	7
18	Variants in the degron of AFF3 are associated with intellectual disability, mesomelic dysplasia, horseshoe kidney, and epileptic encephalopathy. American Journal of Human Genetics, 2021, 108, 857-873.	6.2	19

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19	Truncating SRCAP variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature. American Journal of Human Genetics, 2021, 108, 1053-1068.	6.2	31
20	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. American Journal of Human Genetics, 2021, 108, 1138-1150.	6.2	17
21	ZTTK syndrome: Clinical and molecular findings ofÂ15 cases and a review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 3740-3753.	1.2	11
22	Mild Clinical Presentation of Joubert Syndrome in a Male Adult Carrying Biallelic MKS1 Truncating Variants. Diagnostics, 2021, 11, 1218.	2.6	4
23	Rare and de novo coding variants in chromodomain genes in Chiari I malformation. American Journal of Human Genetics, 2021, 108, 100-114.	6.2	17
24	A pilot clinical trial with losartan in Myhre syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 702-709.	1.2	6
25	RARS1â€related hypomyelinating leukodystrophy: Expanding the spectrum. Annals of Clinical and Translational Neurology, 2020, 7, 83-93.	3.7	18
26	Rubinsteinâ€“Taybi syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2020, 182, 2939-2950.	1.2	16
27	Disease characteristics of MCT8 deficiency: an international, retrospective, multicentre cohort study. Lancet Diabetes and Endocrinology,the, 2020, 8, 594-605.	11.4	50
28	De novo SMARCA2 variants clustered outside the helicase domain cause a new recognizable syndrome with intellectual disability and blepharophimosis distinct from Nicolaidesâ€“Baraitser syndrome. Genetics in Medicine, 2020, 22, 1838-1850.	2.4	31
29	Refinement of the clinical and mutational spectrum of <scp>UBE2A</scp> deficiency syndrome. Clinical Genetics, 2020, 98, 172-178.	2.0	5
30	An Alu-mediated duplication in NMNAT1, involved in NAD biosynthesis, causes a novel syndrome, SHILCA, affecting multiple tissues and organs. Human Molecular Genetics, 2020, 29, 2250-2260.	2.9	14
31	A systematic cross-sectional survey of multiple sulfatase deficiency. Molecular Genetics and Metabolism, 2020, 130, 283-288.	1.1	10
32	Longâ€term followâ€up of an individual with <scp>ITPR1</scp>â€related disorder. American Journal of Medical Genetics, Part A, 2020, 182, 1846-1847.	1.2	0
33	Expansion of the phenotype of lateral meningocele syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1259-1262.	1.2	9
34	Mild form of Zellweger Spectrum Disorders (ZSD) due to variants in PEX1: Detailed clinical investigation in a 9-years-old female. Molecular Genetics and Metabolism Reports, 2020, 24, 100615.	1.1	12
35	Expansion of the phenotypic spectrum of de novo missense variants in kinesin family member 1A () Tj ETQq1 1 0.784314 rgBT /Overlo	2.5	16
36	Two cases of 16q12.1q21 deletions and refinement of the critical region. European Journal of Medical Genetics, 2020, 63, 103878.	1.3	3

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37	Cavitating and tigroidâ€like leukoencephalopathy in a case of NDUFA2â€related disorder. JIMD Reports, 2020, 52, 11-16.	1.5	7
38	A small 7q11.23 microduplication involving <scp>GTF2I</scp> in a family with intellectual disability. Clinical Genetics, 2020, 97, 940-942.	2.0	4
39	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	12.8	105
40	Consolidating the Role of TDP2 Mutations in Recessive Spinocerebellar Ataxia Associated with Pediatric Onset Drug Resistant Epilepsy and Intellectual Disability (SCAR23). Cerebellum, 2019, 18, 972-975.	2.5	12
41	De Novo Heterozygous POLR2A Variants Cause a Neurodevelopmental Syndrome with Profound Infantile-Onset Hypotonia. American Journal of Human Genetics, 2019, 105, 283-301.	6.2	46
42	Paralog Studies Augment Gene Discovery: DDX and DHX Genes. American Journal of Human Genetics, 2019, 105, 302-316.	6.2	56
43	Sphingolipid Metabolism Perturbations in Rett Syndrome. Metabolites, 2019, 9, 221.	2.9	12
44	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogryposis. American Journal of Human Genetics, 2019, 105, 689-705.	6.2	48
45	Opening a window on lysosomal acid lipase deficiency: Biochemical, molecular, and epidemiological insights. Journal of Inherited Metabolic Disease, 2019, 42, 509-518.	3.6	8
46	Microdeletion of pseudogene chr14.232.a affects LRFN5 expression in cells of a patient with autism spectrum disorder. European Journal of Human Genetics, 2019, 27, 1475-1480.	2.8	13
47	Severe presentation and complex brain malformations in an individual carrying a CCND2 variant. Molecular Genetics & Genomic Medicine, 2019, 7, e708.	1.2	7
48	Retinal dystrophy in an individual carrying a de novo missense variant of SMARCA4. Molecular Genetics & Genomic Medicine, 2019, 7, e682.	1.2	8
49	Three de novo DDX3X variants associated with distinctive brain developmental abnormalities and brain tumor in intellectually disabled females. European Journal of Human Genetics, 2019, 27, 1254-1259.	2.8	41
50	Pain and sleep disturbances in Rett syndrome and other neurodevelopmental disorders. Acta Paediatrica, International Journal of Paediatrics, 2019, 108, 171-172.	1.5	4
51	<scp>AP</scp>1S2â€truncating variant in a patient with severe neurodevelopmental disorder and cerebral folate deficiency. Acta Paediatrica, International Journal of Paediatrics, 2019, 108, 564-565.	1.5	2
52	The Treatment of Hypersalivation in Rett Syndrome with Botulinum Toxin: Efficacy and Clinical Implications. Neurology and Therapy, 2019, 8, 155-160.	3.2	6
53	A child with Myhre syndrome presenting with corectopia and tetralogy of Fallot. American Journal of Medical Genetics, Part A, 2018, 176, 426-430.	1.2	15
54	Treatment of hypersalivation in rett syndrome with botulinum toxin: Efficacy and clinical implications. Toxicon, 2018, 156, S28-S29.	1.6	0

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55	Sensorineural Hearing Loss in a Patient Affected by Congenital Cytomegalovirus Infection: Is It Useful to Identify Comorbid Pathologies?. Journal of Pediatric Genetics, 2017, 06, 181-185.	0.7	2
56	An extremely severe phenotype attributed to WDR81 nonsense mutations. Annals of Neurology, 2017, 82, 650-651.	5.3	11
57	Gait disturbance and lower limb pain in a patient with PIK3CA -related disorder. European Journal of Medical Genetics, 2017, 60, 655-657.	1.3	3
58	Expanding the phenotype of DSTâ€related disorder: A case report suggesting a genotype/phenotype correlation. American Journal of Medical Genetics, Part A, 2017, 173, 2743-2746.	1.2	23
59	Biochemical phenotyping unravels novel metabolic abnormalities and potential biomarkers associated with treatment of GLUT1 deficiency with ketogenic diet. PLoS ONE, 2017, 12, e0184022.	2.5	26
60	Expansion of the Phenotypic Spectrum of Propionic Acidemia with Isolated Elevated Propionylcarnitine. JIMD Reports, 2016, 35, 33-37.	1.5	10
61	Diagnosis of adenylosuccinate lyase deficiency by metabolomic profiling in plasma reveals a phenotypic spectrum. Molecular Genetics and Metabolism Reports, 2016, 8, 61-66.	1.1	48
62	New insights in the interpretation of array-CGH: autism spectrum disorder and positive family history for intellectual disability predict the detection of pathogenic variants. Italian Journal of Pediatrics, 2016, 42, 39.	2.6	21
63	De novo PIK3R2 variant causes polymicrogyria, corpus callosum hyperplasia and focal cortical dysplasia. European Journal of Human Genetics, 2016, 24, 1359-1362.	2.8	26
64	Early onset Charcotâ€Marieâ€Tooth neuropathy type <scp>2A</scp> and severe developmental delay: expanding the clinical phenotype of <scp>MFN2</scp>â€related neuropathy. Journal of the Peripheral Nervous System, 2015, 20, 415-418.	3.1	14
65	Giant breast tumors in a patient with Beckwithâ€“Wiedemann syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 182-185.	1.2	6
66	Identification of two novel spliceâ€site mutations in <scp>CHD7</scp> gene in two patients with classical and atypical <scp>CHARGE</scp> syndrome phenotype. Clinical Genetics, 2014, 85, 201-202.	2.0	1
67	Pearls & Oy-sters: Familial epileptic encephalopathy due to methylenetetrahydrofolate reductase deficiency. Neurology, 2014, 83, e41-e44.	1.1	9
68	Bronchial isomerism in a Kabuki syndrome patient with a novel mutation in MLL2gene. BMC Medical Genetics, 2014, 15, 15.	2.1	8
69	Loeysâ€“Dietz syndrome type 4, caused by chromothripsis, involving the TGFB2 gene. Gene, 2014, 538, 69-73.	2.2	19
70	Complex chromosomal rearrangements causing Langerâ€“Giedion syndrome atypical phenotype: Genotypeâ€“phenotype correlation and literature review. American Journal of Medical Genetics, Part A, 2014, 164, 753-759.	1.2	11
71	A case of 14q11.2 microdeletion with autistic features, severe obesity and facial dysmorphisms suggestive of Wolfâ€“Hirschhorn syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 190-193.	1.2	16
72	Low-Dose Amitriptyline-Induced Acute Dystonia in a Patient with Metachromatic Leukodystrophy. JIMD Reports, 2012, 9, 113-116.	1.5	3

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73	Cardiac valve disease: an unreported feature in Ehlers Danlos syndrome arthrocalasia type?. Italian Journal of Pediatrics, 2012, 38, 65.	2.6	12
74	Focal congenital lipoatrophy and vascular malformation: A mild form of inverse Klippelâ€“Trenaunay syndrome?. European Journal of Medical Genetics, 2012, 55, 705-707.	1.3	11
75	Clinical description of a patient carrying the smallest reported deletion involving 10p14 region. American Journal of Medical Genetics, Part A, 2012, 158A, 832-835.	1.2	24
76	Mental retardation, congenital heart malformation, and myelodysplasia in a patient with a complex chromosomal rearrangement involving the critical region 21q22. American Journal of Medical Genetics, Part A, 2011, 155, 1697-1705.	1.2	17
77	Variegated silencing through epigenetic modifications of a large Xq region in a case of balanced X;2 translocation with Incontinentia Pigmenti-like phenotype. Epigenetics, 2011, 6, 1242-1247.	2.7	14