Donatella Greco

List of Publications by Year
in descending order

40 papers	1,177 citations	567247 15 h-index	395678 33 g-index
41 all docs	41 docs citations	41 times ranked	2262 citing authors

#	Article	IF	Citations
1	Clinical spectrum and followâ€up in six individuals with Lambâ€“Shaffer syndrome (<scp>SOX5</scp>). American Journal of Medical Genetics, Part A, 2021, 185, 608-613.	1.2	6
2	Clinical management of individuals with Intellectual Disability: The outbreak of Covid-19 pandemic as experienced in a clinical and research center Research in Developmental Disabilities. Research in Developmental Disabilities, 2021, 110, 103856.	2.2	11
3	Praderâ€“Willi Syndrome with Angelman Syndrome in the Offspring. Medicina (Lithuania), 2021, 57, 460.	2.0	3
4	Impact of daytime routine modifications on people with severe intellectual disability amid COVIDâ€19 pandemic. Perspectives in Psychiatric Care, 2021, 57, 1536-1537.	1.9	4
5	Caring and living with Prader-Willi syndrome in Italy: integrating children, adults and parentsâ€™ experiences through a multicentre narrative medicine research. BMJ Open, 2020, 10, e036502.	1.9	13
6	Structural brain anomalies in Cri-du-Chat syndrome: MRI findings in 14 patients and possible genotype-phenotype correlations. European Journal of Paediatric Neurology, 2020, 28, 110-119.	1.6	3
7	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	12.8	105
8	Mutations in ACTL6B, coding for a subunit of the neuron-specific chromatin remodeling complex nBAF, cause early onset severe developmental and epileptic encephalopathy with brain hypomyelination and cerebellar atrophy. Human Genetics, 2019, 138, 187-198.	3.8	12
9	Interstitial 22q13 deletions not involving SHANK3 gene: A new contiguous gene syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 1666-1676.	1.2	49
10	Increased FGF3 and FGF4 gene dosage is a risk factor for craniosynostosis. Gene, 2014, 534, 435-439.	2.2	19
11	Identification of pathogenic gene variants in small families with intellectually disabled siblings by exome sequencing. Journal of Medical Genetics, 2013, 50, 802-811.	3.2	93
12	3q29 microdeletion syndrome: Cognitive and behavioral phenotype in four patients. American Journal of Medical Genetics, Part A, 2013, 161, 3018-3022.	1.2	19
13	The duplication 17p13.3 phenotype: Analysis of 21 families delineates developmental, behavioral and brain abnormalities, and rare variant phenotypes. American Journal of Medical Genetics, Part A, 2013, 161, 1833-1852.	1.2	53
14	Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. European Journal of Human Genetics, 2013, 21, 1074-1078.	2.8	64
15	Cutaneous Atrophic Guttate Lesions in a Linear and Reticulate Pattern: A Quiz. Acta Dermato-Venereologica, 2013, 93, 124-127.	1.3	3
16	The Pittâ€Hopkins syndrome: Report of 16 new patients and clinical diagnostic criteria. American Journal of Medical Genetics, Part A, 2011, 155, 1536-1545.	1.2	55
17	Molecular Mechanisms Generating and Stabilizing Terminal 22q13 Deletions in 44 Subjects with Phelan/McDermid Syndrome. PLoS Genetics, 2011, 7, e1002173.	3.5	172
18	NovelTMEM67mutations and genotype-phenotype correlates in meckelin-related ciliopathies. Human Mutation, 2010, 31, n/a-n/a.	2.5	77

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19	Expanding CEP290 mutational spectrum in ciliopathies. American Journal of Medical Genetics, Part A, 2009, 149A, 2173-2180.	1.2	38
20	MKS3/TMEM67mutations are a major cause of COACH Syndrome, a Joubert Syndrome related disorder with liver involvement. Human Mutation, 2009, 30, E432-E442.	2.5	96
21	Three new patients with dup(17)(p11.2p11.2) without autism. Clinical Genetics, 2008, 73, 294-296.	2.0	7
22	Cerebriform plantar hyperplasia: the major cutaneous feature of Proteus syndrome. International Journal of Dermatology, 2008, 47, 374-376.	1.0	10
23	Psychological consequences of prenatal diagnosis in a case of familial Angelman Syndrome. Prenatal Diagnosis, 2006, 26, 1156-1159.	2.3	2
24	Screening of subtelomeric rearrangements in autistic disorder: Identification of a partial trisomy of 13q34 in a patient bearing a 13q;21p translocation. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2006, 141B, 584-590.	1.7	12
25	Piezogenic pedal papules during Praderâ€“Willi syndrome. Journal of the European Academy of Dermatology and Venereology, 2005, 19, 136-137.	2.4	17
26	Identification of Novel Mutations in Patients with Coffinâ€“Lowry Syndrome by a Denaturing HPLC-Based Assay. Clinical Chemistry, 2005, 51, 2356-2358.	3.2	5
27	Medial Telangiectatic Sacral Nevi and Mca/Mr Syndromes. Pediatric Dermatology, 2003, 20, 370-371.	0.9	6
28	Facial and Skeletal Malformations, Mental Retardation, Aganglionosis, and Neurogenic Muscle Weakness: A Variant of Niikawa-Kuroki Syndrome or a New Syndrome?. Journal of Child Neurology, 2001, 16, 296-296.	1.4	0
29	Rubinsteinâ€“Taybi Syndrome with Epidermal Nevus: A Case Report. Pediatric Dermatology, 2001, 18, 34-37.	0.9	10
30	Cryptic telomeric rearrangements in subjects with mental retardation associated with dysmorphism and congenital malformations. Journal of Medical Genetics, 2001, 38, 417-420.	3.2	114
31	Medial Telangiectatic Sacral Nevi (Types A and C) Associated with Williams Syndrome. Dermatology, 2000, 201, 285-286.	2.1	6
32	Cardiofaciocutaneous (CFC) syndrome. Australasian Journal of Dermatology, 1999, 40, 111-113.	0.7	7
33	Maternal phenylketonuria in two Sicilian families identified by maternal blood phenylalanine level screening and identification of a new phenylalanine hydroxylase gene mutation (P407L). European Journal of Pediatrics, 1999, 158, 83-84.	2.7	4
34	An intriguing case of LEOPARD syndrome.. Pediatric Dermatology, 1998, 15, 125-128.	0.9	13
35	Eight new mutations of the phenylalanine hydroxylase gene in Italian patients with hyperphenylalaninemia. Human Mutation, 1998, 11, 240-243.	2.5	0
36	Leigh syndrome and partial deficit of cytochrome c oxidase associated with epilepsia partialis continua. Brain and Development, 1996, 18, 207-211.	1.1	37

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37	Saethre-Chotzen syndrome: a clinical, EEG and neuroradiological study. Child's Nervous System, 1996, 12, 699-704.	1.1	9
38	Phenotypic and phoniatric findings in mosaic cri du chat syndrome. American Journal of Medical Genetics Part A, 1991, 39, 391-395.	2.4	12
39	RELIABILITY OF INTRADERMAL VACCINATION AGAINST HEPATITIS B FOR ACCELERATED PROPHYLAXIS. Pediatric Infectious Disease Journal, 1990, 9, 520.	2.0	4
40	PEDIATRIC ACQUIRED IMMUNODEFICIENCY SYNDROME IN ITALY. Pediatric Infectious Disease Journal, 1987, 6, 863-864.	2.0	1