Marcello Niceta

List of Publications by Year
in descending order

51 papers	1,414 citations	331670 21 h-index	361022 35 g-index
52 all docs	52 docs citations	52 times ranked	3470 citing authors

#	Article	IF	Citations
1	A novel disorder involving dyshematopoiesis, inflammation, and HLH due to aberrant CDC42 function. Journal of Experimental Medicine, 2019, 216, 2778-2799.	8.5	132
2	Mutations Impairing GSK3-Mediated MAF Phosphorylation Cause Cataract, Deafness, Intellectual Disability, Seizures, and a Down Syndrome-like Facies. American Journal of Human Genetics, 2015, 96, 816-825.	6.2	102
3	Persistent episomal transgene expression in liver following delivery of a scaffold/matrix attachment region containing non-viral vector. Gene Therapy, 2008, 15, 1593-1605.	4.5	91
4	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
5	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
6	Development of S/MAR minicircles for enhanced and persistent transgene expression in the mouse liver. Journal of Molecular Medicine, 2011, 89, 515-529.	3.9	60
7	Childhood onset tubular aggregate myopathy associated with de novo STIM1 mutations. Journal of Neurology, 2014, 261, 870-876.	3.6	56
8	LYRM7mutations cause a multifocal cavitating leukoencephalopathy with distinct MRI appearance. Brain, 2016, 139, 782-794.	7.6	51
9	TBCE Mutations Cause Early-Onset Progressive Encephalopathy with Distal Spinal Muscular Atrophy. American Journal of Human Genetics, 2016, 99, 974-983.	6.2	49
10	Not only dominant, not only optic atrophy: expanding the clinical spectrum associated with OPA1 mutations. Orphanet Journal of Rare Diseases, 2017, 12, 89.	2.7	39
11	Novel mutations in IBA57 are associated with leukodystrophy and variable clinical phenotypes. Journal of Neurology, 2017, 264, 102-111.	3.6	38
12	Mutations in Fibronectin Cause a Subtype of Spondylometaphyseal Dysplasia with â€œCorner Fracturesâ€• American Journal of Human Genetics, 2017, 101, 815-823.	6.2	37
13	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
14	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
15	<scp>DJ</scp>â€1 modulates mitochondrial response to oxidative stress: clues from a novel diagnosis of <scp>PARK7</scp>. Clinical Genetics, 2017, 92, 18-25.	2.0	34
16	Mutations in the <scp>IRBIT</scp> domain of <scp>ITPR1</scp> are a frequent cause of autosomal dominant nonprogressive congenital ataxia. Clinical Genetics, 2017, 91, 86-91.	2.0	30
17	Anti-Hypothalamus and Anti-Pituitary AutoÂantibodies in ROHHAD Syndrome: Additional Evidence Supporting an Autoimmune Etiopathogenesis. Hormone Research in Paediatrics, 2019, 92, 124-132.	1.8	27
18	Evaluation of serum CA 125 levels in patients with pelvic pain related to endometriosis. International Journal of Biological Markers, 2007, 22, 200-202.	1.8	27

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19	NBAS pathogenic variants: Defining the associated clinical and facial phenotype and genotypeâ€“phenotype correlations. Human Mutation, 2019, 40, 721-728.	2.5	26
20	Microcephaly, intractable seizures and developmental delay caused by biallelic variants in <scp>TBCD</scp>: further delineation of a new chaperoneâ€mediated tubulinopathy. Clinical Genetics, 2017, 91, 725-738.	2.0	25
21	Expanding the molecular diversity and phenotypic spectrum of glycerol 3â€phosphate dehydrogenase 1 deficiency. Journal of Inherited Metabolic Disease, 2016, 39, 689-695.	3.6	24
22	A novel mutation in <scp>NDUFB11</scp> unveils a new clinical phenotype associated with lactic acidosis and sideroblastic anemia. Clinical Genetics, 2017, 91, 441-447.	2.0	24
23	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
24	Clinical spectrum of Kabukiâ€like syndrome caused by <scp>HNRNPK</scp> haploinsufficiency. Clinical Genetics, 2018, 93, 401-407.	2.0	23
25	Biallelic mutations in <scp>DYNC2LI1</scp> are a rare cause of Ellisâ€van Creveld syndrome. Clinical Genetics, 2018, 93, 632-639.	2.0	23
26	Bi-allelic Variants in the GPI Transamidase Subunit PIGK Cause a Neurodevelopmental Syndrome with Hypotonia, Cerebellar Atrophy, and Epilepsy. American Journal of Human Genetics, 2020, 106, 484-495.	6.2	22
27	Childhood-onset dystonia-causing KMT2B variants result in a distinctive genomic hypermethylation profile. Clinical Epigenetics, 2021, 13, 157.	4.1	22
28	Refractory Acne and 21-Hydroxylase Deficiency in a Selected Group of Female Patients. Dermatology, 2010, 220, 121-127.	2.1	21
29	Identification of two new mutations in TRPS 1 gene leading to the trichoâ€rhinoâ€phalangeal syndrome type I and III. American Journal of Medical Genetics, Part A, 2009, 149A, 1837-1841.	1.2	19
30	Biallelic mutations in the homeodomain of NKX6-2 underlie a severe hypomyelinating leukodystrophy. Brain, 2017, 140, 2550-2556.	7.6	18
31	Expanding the clinical spectrum associated with PACS2 mutations. Clinical Genetics, 2019, 95, 525-531.	2.0	18
32	De novo p.T362R mutation in MORC2 causes early onset cerebellar ataxia, axonal polyneuropathy and nocturnal hypoventilation. Brain, 2017, 140, e34-e34.	7.6	17
33	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
34	Q289P mutation in the FGFR2 gene: first report in a patient with type 1 Pfeiffer syndrome. European Journal of Pediatrics, 2009, 168, 1135-1139.	2.7	13
35	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
36	Cardiac Defects and Genetic Syndromes: Old Uncertainties and New Insights. Genes, 2021, 12, 1047.	2.4	12

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37	Skeletal abnormalities are common features in AymÃ©â€Gripp syndrome. Clinical Genetics, 2020, 97, 362-369.	2.0	10
38	A mild form of adenylosuccinate lyase deficiency in absence of typical brain MRI features diagnosed by whole exome sequencing. Italian Journal of Pediatrics, 2017, 43, 65.	2.6	9
39	Epidemiological study of nonsyndromic hearing loss in Sicilian newborns. American Journal of Medical Genetics, Part A, 2007, 143A, 1666-1670.	1.2	8
40	Clinical Utility of a Unique Genome-Wide DNA Methylation Signature for KMT2A-Related Syndrome. International Journal of Molecular Sciences, 2022, 23, 1815.	4.1	8
41	Expanding the <scp>KIF4A</scp>â€associated phenotype. American Journal of Medical Genetics, Part A, 2021, 185, 3728-3739.	1.2	6
42	Biallelic variants in <scp>ZNF142</scp> lead to a syndromic neurodevelopmental disorder. Clinical Genetics, 2022, 102, 98-109.	2.0	6
43	MTHFR C677T homozygous as risk factor for complications after OLT for cryptogenic cirrhosis. Clinical Transplantation, 2006, 20, 796-798.	1.6	5
44	Glucose 6â€phosphate dehydrogenase Palermo R257M: a novel variant associated with chronic nonâ€spherocytic haemolytic anaemia. British Journal of Haematology, 2010, 149, 296-297.	2.5	4
45	Somatic mosaicism represents an underestimated event underlying collagen 6-related disorders. European Journal of Paediatric Neurology, 2017, 21, 873-883.	1.6	4
46	Vitiligo susceptibility and catalase gene polymorphisms in Sicilian population. Giornale Italiano Di Dermatologia E Venereologia, 2018, 153, 619-623.	0.8	4
47	<scp>SHP2</scp>'s gainâ€ofâ€function in <scp>Werner</scp> syndrome causes childhood disease onset likely resulting from negative genetic interaction. Clinical Genetics, 2022, 102, 12-21.	2.0	2
48	A Child with Diminished Linear Growth and Waddling Gait. Journal of Pediatrics, 2018, 201, 297-297.e1.	1.8	1
49	Clinical Application of Easychip 8x15K Platform in 4106 Pregnancies Without Ultrasound Anomalies. Reproductive Sciences, 2021, 28, 1142-1149.	2.5	1
50	A novel nonsense mutation in exon 2 of the factor IX gene resulting in severe haemophilia B. Internal and Emergency Medicine, 2006, 1, 318-320.	2.0	0
51	A large view of CYP21 locus among Sicilians and other populations: identification of a novel CYP21A2 variant in Sicily. Journal of Endocrinological Investigation, 2011, 34, 847-54.	3.3	0