Francesco Cali

List of Publications by Year
in descending order

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Version: 2024-02-01

78	3,481	331670 21	149698 56
papers	citations	h-index	g-index

80	80	80	5484
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	KCNQ2-Related Neonatal Epilepsy Treated With Vitamin B6: A Report of Two Cases and Literature Review. Frontiers in Neurology, 2022, 13, 826225.	2.4	1
2	Sensory-Adapted Dental Environment for the Treatment of Patients with Autism Spectrum Disorder. Children, 2022, 9, 393.	1.5	9
3	Boolean Networks: A Primer. , 2021, , 41-53.		0
4	A Novel Homozygous <i>ALG12</i> Mutation in a Patient with CDG Type Ig: New Report of a Case with a Mild Phenotype. Molecular Syndromology, 2021, 12, 327-332.	0.8	1
5	Archaeogenetics and Landscape Dynamics in Sicily during the Holocene: A Review. Sustainability, 2021, 13, 9469.	3.2	7
6	Role of COMT V158M Polymorphism in the Development of Dystonia after Administration of Antipsychotic Drugs. Brain Sciences, 2021, 11, 1293.	2.3	5
7	Implementation of Sample Pooling Procedure Using a Rapid SARS-CoV-2 Diagnostic Real-Time PCR Test Performed Prior to Hospital Admission of People with Intellectual Disabilities. International Journal of Environmental Research and Public Health, 2021, 18, 9317.	2.6	5
8	Letter to the Editor Regarding the Article â€œWhole-Exome Sequencing in NF1-Related West's Syndrome Leads to the Identification of KCNC2 as a Novel Candidate Gene for Epilepsyâ€• Neuropediatrics, 2021, 52, 153-153.	0.6	0
9	An odd precocious case of progressive osseous heteroplasia. Italian Journal of Dermatology and Venereology, 2021, 156, 409-411.	0.2	0
10	TBC1D24 gene mRNA expression in a boy with early infantile epileptic encephalopathy-16. Acta Neurologica Belgica, 2020, 120, 381-383.	1.1	3
11	Are Mutations in the DHRS9 Gene Causally Linked to Epilepsy? A Case Report. Medicina (Lithuania), 2020, 56, 387.	2.0	2
12	A Customized Next-Generation Sequencing-Based Panel to Identify Novel Genetic Variants in Dementing Disorders: A Pilot Study. Neural Plasticity, 2020, 2020, 1-10.	2.2	6
13	A de novo heterozygous mutation in KCNC2 gene implicated in severe developmental and epileptic encephalopathy. European Journal of Medical Genetics, 2020, 63, 103848.	1.3	24
14	Late-onset oro-facial dyskinesia in Spinocerebellar Ataxia type 2: a case report. BMC Neurology, 2020, 20, 156.	1.8	7
15	Novel compound heterozygous mutation in NPC1 gene cause Niemannâ€“Pick disease type C with juvenile onset. Journal of Genetics, 2020, 99, 1.	0.7	2
16	Novel SPINK5 variants in a patient with Netherton syndrome and intellectual disability. The diagnostic value of trichoscopy. Giornale Italiano Di Dermatologia E Venereologia, 2020, 155, 239-240.	0.8	1
17	Pigmented porokeratosis with dermal deposits of amyloid: the different chromatic features. Giornale Italiano Di Dermatologia E Venereologia, 2020, 155, 240-241.	0.8	0
18	Possible implication of undescribed SMN1-SMN2 genotype in chronic EMG-pattern of SMA with transitory acute denervation. Journal of Musculoskeletal Neuronal Interactions, 2020, 20, 610-613.	0.1	0

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19	GRIN2A: involvement in movement disorders and intellectual disability without seizures. Neurological Sciences, 2019, 40, 2405-2406.	1.9	3
20	Identification of human D lactate dehydrogenase deficiency. Nature Communications, 2019, 10, 1477.	12.8	62
21	Interpreting Genetic Variants: Hints from a Family Cluster of Parkinsonâ€™s Disease. Journal of Parkinson's Disease, 2019, 9, 203-206.	2.8	11
22	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
23	Novel c.C2254T (p.Q752*) mutation in ZFYVE26 (SPG15) gene in a patient with hereditary spastic paraparesis. Journal of Genetics, 2018, 97, 1469-1472.	0.7	3
24	Novel c.C2254T (p.Q752*) mutation in (SPG15) gene in a patient with hereditary spastic paraparesis. Journal of Genetics, 2018, 97, 1469-1472.	0.7	0
25	Aninteresting case of Piebaldism with cafÃ©-au-lait macules and freckling: the use of targeted next-generation sequencing for molecular diagnosis. European Journal of Dermatology, 2018, 28, 119-120.	0.6	1
26	Mutation spectrum of NF1 gene in Italian patients with neurofibromatosis type 1 using Ion Torrent PGMâ„¢ platform. European Journal of Medical Genetics, 2017, 60, 93-99.	1.3	30
27	Dental anxiety in patients with borderline intellectual functioning and patients with intellectual disabilities. BMC Oral Health, 2016, 16, 114.	2.3	18
28	The Greeks in the West: genetic signatures of the Hellenic colonisation in southern Italy and Sicily. European Journal of Human Genetics, 2016, 24, 429-436.	2.8	26
29	A novel splice acceptor site mutation in the ATP2A2 gene in a family with Darier disease. Giornale Italiano Di Dermatologia E Venereologia, 2016, 151, 582-5.	0.8	0
30	The Role of Recent Admixture in Forming the Contemporary West Eurasian Genomic Landscape. Current Biology, 2015, 25, 2518-2526.	3.9	68
31	Assessing the Impact of Copy Number Variants on miRNA Genes in Autism by Monte Carlo Simulation. PLoS ONE, 2014, 9, e90947.	2.5	25
32	Carrier screening for spinal muscular atrophy in Italian population. Journal of Genetics, 2014, 93, 179-181.	0.7	8
33	Ancient human genomes suggest three ancestral populations for present-day Europeans. Nature, 2014, 513, 409-413.	27.8	1,179
34	GENETIC RELATIONSHIPS OF BRASSICA VEGETABLES AND WILD RELATIVES IN SOUTHERN ITALY DETERMINED BY FIVE SSR. Acta Horticulturae, 2013, , 189-196.	0.2	8
35	Comparative multiplex dosage analysis in spinocerebellar ataxia type 2 patients. Genetics and Molecular Research, 2013, 12, 1176-1181.	0.2	1
36	Multiplex ligation-dependent probe amplification detection of an unknown large deletion of the CREB-binding protein gene in a patient with Rubinstein-Taybi Syndrome. Genetics and Molecular Research, 2013, 12, 2809-15.	0.2	5

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37	Short-term results of a randomized trial examining timing of carotid endarterectomy in patients with severe asymptomatic unilateral carotid stenosis undergoing coronary artery bypass grafting. Journal of Vascular Surgery, 2011, 54, 993-999.	1.1	69
38	Functional Annotation of Genes Overlapping Copy Number Variants in Autistic Patients: Focus on Axon Pathfinding. Current Genomics, 2010, 11, 136-145.	1.6	29
39	Exon deletions of the phenylalanine hydroxylase gene in Italian hyperphenylalaninemics. Experimental and Molecular Medicine, 2010, 42, 81.	7.7	13
40	Novel deletion of the E3A ubiquitin protein ligase gene detected by multiplex ligation-dependent probe amplification in a patient with Angelman syndrome. Experimental and Molecular Medicine, 2010, 42, 842.	7.7	5
41	SPANX-B and SPANX-C (Xq27 region) gene dosage analysis in Downâ€™s syndrome subjects with undescended testes. Journal of Genetics, 2009, 88, 93-97.	0.7	3
42	Differential Greek and northern African migrations to Sicily are supported by genetic evidence from the Y chromosome. European Journal of Human Genetics, 2009, 17, 91-99.	2.8	43
43	Moors and Saracens in Europe: estimating the medieval North African male legacy in southern Europe. European Journal of Human Genetics, 2009, 17, 848-852.	2.8	37
44	Analysis of the gastrinâ€releasing peptide receptor gene in Italian patients with autism spectrum disorders. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2008, 147B, 807-813.	1.7	10
45	SPANX-B and SPANX-C (Xq27 region) gene dosage analysis in Sicilian patients with melanoma. Melanoma Research, 2008, 18, 295-299.	1.2	7
46	1.5 Mb de novo 22q11.21 microduplication in a patient with cognitive deficits and dysmorphic facial features. Clinical Genetics, 2007, 71, 177-182.	2.0	52
47	Mutational Analysis of EFHC1 Gene in Italian Families with Juvenile Myoclonic Epilepsy. Epilepsia, 2007, 48, 1686-1690.	5.1	44
48	Juvenile myoclonic epilepsy with generalised and focal electroencephalographic abnormalities: a case report with a molecular genetic study. Neurological Sciences, 2007, 28, 276-278.	1.9	2
49	Population Structure in the Mediterranean Basin: A Y Chromosome Perspective. Annals of Human Genetics, 2006, 70, 207-225.	0.8	56
50	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
51	Suggestive evidence for association of D2S2188 marker (2q31.1) with autism in 143 Sicilian (Italian) TRIO families. Psychiatric Genetics, 2005, 15, 149-150.	1.1	6
52	mtDNA analysis ofÂtheÂhuman remains buried inÂtheÂsarcophagus ofÂFederico II. Journal of Cultural Heritage, 2005, 6, 313-319.	3.3	3
53	Skewed X-inactivation in a family with mental retardation and PQBP1 gene mutation. Clinical Genetics, 2005, 67, 446-447.	2.0	11
54	Phylogeographic Analysis of Haplogroup E3b (E-M215) Y Chromosomes Reveals Multiple Migratory Events Within and Out Of Africa. American Journal of Human Genetics, 2004, 74, 1014-1022.	6.2	197

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55	Genetic diversity within the R408W phenylketonuria mutation lineages in Europe. Human Mutation, 2003, 21, 387-393.	2.5	32
56	Cell line DNA typing in forensic geneticsâ€"the necessity of reliable standards. Forensic Science International, 2003, 138, 37-43.	2.2	102
57	Autosomal Microsatellite and mtDNA Genetic Analysis in Sicily (Italy). Annals of Human Genetics, 2003, 67, 42-53.	0.8	17
58	Lack of association of HOXA1 and HOXB1 mutations and autism in Sicilian (Italian) patients. Molecular Psychiatry, 2003, 8, 716-717.	7.9	18
59	Continental and subcontinental distributions of mtDNA control region types. International Journal of Legal Medicine, 2002, 116, 99-108.	2.2	40
60	DXYS156: a multi-purpose short tandem repeat locus for determination of sex, paternal and maternal geographic origins and DNA fingerprinting. International Journal of Legal Medicine, 2002, 116, 133-138.	2.2	24
61	A methodological strategy for PAH genotyping in populations with a marked molecular heterogeneity of hyperphenylalaninemia. Molecular and Cellular Probes, 2001, 15, 13-19.	2.1	6
62	PAH Gene Mutations in the Sicilian Population: Association with Minihaplotypes and Expression Analysis. Molecular Genetics and Metabolism, 2001, 74, 353-361.	1.1	16
63	MtDNA control region and RFLP data for Sicily and France. International Journal of Legal Medicine, 2001, 114, 229-231.	2.2	37
64	Genetic Heterogeneity in Five Italian Regions: Analysis of PAH Mutations and Minihaplotypes. Human Heredity, 2001, 52, 154-159.	0.8	20
65	Dramatic brain aminergic deficit in a genetic mouse model of phenylketonuria. NeuroReport, 2000, 11, 1361-1364.	1.2	100
66	Towards a genetic history of Sicily. Journal of Cultural Heritage, 2000, 1, S39-S42.	3.3	1
67	Mutations and polymorphisms of the PAH gene in Sicily: comparison with other DNA polymorphisms. Journal of Cultural Heritage, 2000, 1, S43-S45.	3.3	1
68	Molecular basis of mild hyperphenylalaninaemia in Turkey. Journal of Inherited Metabolic Disease, 2000, 23, 523-525.	3.6	10
69	Tracing European Founder Lineages in the Near Eastern mtDNA Pool. American Journal of Human Genetics, 2000, 67, 1251-1276.	6.2	837
70	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
71	Eight new mutations of the phenylalanine hydroxylase gene in Italian patients with hyperphenylalaninemia. Human Mutation, 1998, 11, 240-243.	2.5	7
72	Eight new mutations of the phenylalanine hydroxylase gene in Italian patients with hyperphenylalaninemia. Human Mutation, 1998, 11, 240-243.	2.5	0

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73	Two novel PAH gene mutations detected in Italian phenylketonuric patients. Human Genetics, 1997, 99, 275-278.	3.8	0
74	The STR252 - IVS10nt546 - VNTR7 phenylalanine hydroxylase minihaplotype in five Mediterranean samples. Human Genetics, 1997, 100, 350-355.	3.8	17
75	Preliminary studies on the molecular basis of hyperphenylalaninemia in Egypt. Human Genetics, 1996, 98, 3-6.	3.8	9
76	PAH deficiency in Italy: correlation of genotype with phenotype in the Sicilian population. Journal of Inherited Metabolic Disease, 1996, 19, 15-24.	3.6	20
77	Association between haplotypes, Hind III-VNTR alleles and mutations at the PAH locus in Sicily. Acta Paediatrica, International Journal of Paediatrics, 1994, 83, 39-40.	1.5	3
78	Exome sequencing in a child with neurodevelopmental disorder and epilepsy: Variant analysis of the <scp>AHNAK2</scp> gene. Molecular Genetics & Genomic Medicine, 0, , .	1.2	1