

# Francesco Cali

## List of Publications by Year in descending order

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78  
papers

3,481  
citations

331670

21  
h-index

149698

56  
g-index

80  
all docs

80  
docs citations

80  
times ranked

5484  
citing authors

#	ARTICLE	IF	CITATIONS
1	Ancient human genomes suggest three ancestral populations for present-day Europeans. <i>Nature</i> , 2014, 513, 409-413.	27.8	1,179
2	Tracing European Founder Lineages in the Near Eastern mtDNA Pool. <i>American Journal of Human Genetics</i> , 2000, 67, 1251-1276.	6.2	837
3	Phylogeographic Analysis of Haplogroup E3b (E-M215) Y Chromosomes Reveals Multiple Migratory Events Within and Out Of Africa. <i>American Journal of Human Genetics</i> , 2004, 74, 1014-1022.	6.2	197
4	Cell line DNA typing in forensic genetics—the necessity of reliable standards. <i>Forensic Science International</i> , 2003, 138, 37-43.	2.2	102
5	Dramatic brain aminergic deficit in a genetic mouse model of phenylketonuria. <i>NeuroReport</i> , 2000, 11, 1361-1364.	1.2	100
6	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. <i>Journal of Vascular Surgery</i> , 2011, 54, 993-999.	1.1	69
7	The Role of Recent Admixture in Forming the Contemporary West Eurasian Genomic Landscape. <i>Current Biology</i> , 2015, 25, 2518-2526.	3.9	68
8	Identification of human D lactate dehydrogenase deficiency. <i>Nature Communications</i> , 2019, 10, 1477.	12.8	62
9	Population Structure in the Mediterranean Basin: A Y Chromosome Perspective. <i>Annals of Human Genetics</i> , 2006, 70, 207-225.	0.8	56
10	1.5 Mb de novo 22q11.21 microduplication in a patient with cognitive deficits and dysmorphic facial features. <i>Clinical Genetics</i> , 2007, 71, 177-182.	2.0	52
11	Mutational Analysis of EFHC1 Gene in Italian Families with Juvenile Myoclonic Epilepsy. <i>Epilepsia</i> , 2007, 48, 1686-1690.	5.1	44
12	Differential Greek and northern African migrations to Sicily are supported by genetic evidence from the Y chromosome. <i>European Journal of Human Genetics</i> , 2009, 17, 91-99.	2.8	43
13	Continental and subcontinental distributions of mtDNA control region types. <i>International Journal of Legal Medicine</i> , 2002, 116, 99-108.	2.2	40
14	MtDNA control region and RFLP data for Sicily and France. <i>International Journal of Legal Medicine</i> , 2001, 114, 229-231.	2.2	37
15	Moors and Saracens in Europe: estimating the medieval North African male legacy in southern Europe. <i>European Journal of Human Genetics</i> , 2009, 17, 848-852.	2.8	37
16	Genetic diversity within the R408W phenylketonuria mutation lineages in Europe. <i>Human Mutation</i> , 2003, 21, 387-393.	2.5	32
17	Mutation spectrum of NF1 gene in Italian patients with neurofibromatosis type 1 using Ion Torrent PGM <sub>seq</sub> platform. <i>European Journal of Medical Genetics</i> , 2017, 60, 93-99.	1.3	30
18	Functional Annotation of Genes Overlapping Copy Number Variants in Autistic Patients: Focus on Axon Pathfinding. <i>Current Genomics</i> , 2010, 11, 136-145.	1.6	29

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19	The Greeks in the West: genetic signatures of the Hellenic colonisation in southern Italy and Sicily. <i>European Journal of Human Genetics</i> , 2016, 24, 429-436.	2.8	26
20	Assessing the Impact of Copy Number Variants on miRNA Genes in Autism by Monte Carlo Simulation. <i>PLoS ONE</i> , 2014, 9, e90947.	2.5	25
21	DXYS156: a multi-purpose short tandem repeat locus for determination of sex, paternal and maternal geographic origins and DNA fingerprinting. <i>International Journal of Legal Medicine</i> , 2002, 116, 133-138.	2.2	24
22	A de novo heterozygous mutation in KCNC2 gene implicated in severe developmental and epileptic encephalopathy. <i>European Journal of Medical Genetics</i> , 2020, 63, 103848.	1.3	24
23	PAH deficiency in Italy: correlation of genotype with phenotype in the Sicilian population. <i>Journal of Inherited Metabolic Disease</i> , 1996, 19, 15-24.	3.6	20
24	Genetic Heterogeneity in Five Italian Regions: Analysis of PAH Mutations and Minihaplotypes. <i>Human Heredity</i> , 2001, 52, 154-159.	0.8	20
25	Lack of association of HOXA1 and HOXB1 mutations and autism in Sicilian (Italian) patients. <i>Molecular Psychiatry</i> , 2003, 8, 716-717.	7.9	18
26	Dental anxiety in patients with borderline intellectual functioning and patients with intellectual disabilities. <i>BMC Oral Health</i> , 2016, 16, 114.	2.3	18
27	The STR252 - IVS10nt546 - VNTR7 phenylalanine hydroxylase minihaplotype in five Mediterranean samples. <i>Human Genetics</i> , 1997, 100, 350-355.	3.8	17
28	Autosomal Microsatellite and mtDNA Genetic Analysis in Sicily (Italy). <i>Annals of Human Genetics</i> , 2003, 67, 42-53.	0.8	17
29	PAH Gene Mutations in the Sicilian Population: Association with Minihaplotypes and Expression Analysis. <i>Molecular Genetics and Metabolism</i> , 2001, 74, 353-361.	1.1	16
30	Exon deletions of the phenylalanine hydroxylase gene in Italian hyperphenylalaninemics. <i>Experimental and Molecular Medicine</i> , 2010, 42, 81.	7.7	13
31	Screening of subtelomeric rearrangements in autistic disorder: Identification of a partial trisomy of 13q34 in a patient bearing a 13q;21p translocation. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2006, 141B, 584-590.	1.7	12
32	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. <i>Human Genetics</i> , 2019, 138, 187-198.	3.8	12
33	Skewed X-inactivation in a family with mental retardation and PQBP1 gene mutation. <i>Clinical Genetics</i> , 2005, 67, 446-447.	2.0	11
34	Interpreting Genetic Variants: Hints from a Family Cluster of Parkinson's Disease. <i>Journal of Parkinson's Disease</i> , 2019, 9, 203-206.	2.8	11
35	Molecular basis of mild hyperphenylalaninaemia in Turkey. <i>Journal of Inherited Metabolic Disease</i> , 2000, 23, 523-525.	3.6	10
36	Analysis of the gastrin-releasing peptide receptor gene in Italian patients with autism spectrum disorders. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2008, 147B, 807-813.	1.7	10

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37	Preliminary studies on the molecular basis of hyperphenylalaninemia in Egypt. <i>Human Genetics</i> , 1996, 98, 3-6.	3.8	9
38	Sensory-Adapted Dental Environment for the Treatment of Patients with Autism Spectrum Disorder. <i>Children</i> , 2022, 9, 393.	1.5	9
39	GENETIC RELATIONSHIPS OF BRASSICA VEGETABLES AND WILD RELATIVES IN SOUTHERN ITALY DETERMINED BY FIVE SSR. <i>Acta Horticulturae</i> , 2013, , 189-196.	0.2	8
40	Carrier screening for spinal muscular atrophy in Italian population. <i>Journal of Genetics</i> , 2014, 93, 179-181.	0.7	8
41	Eight new mutations of the phenylalanine hydroxylase gene in Italian patients with hyperphenylalaninemia. <i>Human Mutation</i> , 1998, 11, 240-243.	2.5	7
42	SPANX-B and SPANX-C (Xq27 region) gene dosage analysis in Sicilian patients with melanoma. <i>Melanoma Research</i> , 2008, 18, 295-299.	1.2	7
43	Late-onset oro-facial dyskinesia in Spinocerebellar Ataxia type 2: a case report. <i>BMC Neurology</i> , 2020, 20, 156.	1.8	7
44	Archaeogenetics and Landscape Dynamics in Sicily during the Holocene: A Review. <i>Sustainability</i> , 2021, 13, 9469.	3.2	7
45	A methodological strategy for PAH genotyping in populations with a marked molecular heterogeneity of hyperphenylalaninemia. <i>Molecular and Cellular Probes</i> , 2001, 15, 13-19.	2.1	6
46	Suggestive evidence for association of D2S2188 marker (2q31.1) with autism in 143 Sicilian (Italian) TRIO families. <i>Psychiatric Genetics</i> , 2005, 15, 149-150.	1.1	6
47	A Customized Next-Generation Sequencing-Based Panel to Identify Novel Genetic Variants in Dementing Disorders: A Pilot Study. <i>Neural Plasticity</i> , 2020, 2020, 1-10.	2.2	6
48	Novel deletion of the E3A ubiquitin protein ligase gene detected by multiplex ligation-dependent probe amplification in a patient with Angelman syndrome. <i>Experimental and Molecular Medicine</i> , 2010, 42, 842.	7.7	5
49	Role of COMT V158M Polymorphism in the Development of Dystonia after Administration of Antipsychotic Drugs. <i>Brain Sciences</i> , 2021, 11, 1293.	2.3	5
50	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. <i>International Journal of Environmental Research and Public Health</i> , 2021, 18, 9317.	2.6	5
51	Multiplex ligation-dependent probe amplification detection of an unknown large deletion of the CREB-binding protein gene in a patient with Rubinstein-Taybi Syndrome. <i>Genetics and Molecular Research</i> , 2013, 12, 2809-15.	0.2	5
52	Maternal phenylketonuria in two Sicilian families identified by maternal blood phenylalanine level screening and identification of a new phenylalanine hydroxylase gene mutation (P407L). <i>European Journal of Pediatrics</i> , 1999, 158, 83-84.	2.7	4
53	Association between haplotypes, Hind III-VNTR alleles and mutations at the PAH locus in Sicily. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 1994, 83, 39-40.	1.5	3
54	mtDNA analysis of the human remains buried in the sarcophagus of Federico II. <i>Journal of Cultural Heritage</i> , 2005, 6, 313-319.	3.3	3

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55	SPANX-B and SPANX-C (Xq27 region) gene dosage analysis in Down's syndrome subjects with undescended testes. <i>Journal of Genetics</i> , 2009, 88, 93-97.	0.7	3
56	Novel c.C2254T (p.Q752*) mutation in ZFYVE26 (SPG15) gene in a patient with hereditary spastic paraparesis. <i>Journal of Genetics</i> , 2018, 97, 1469-1472.	0.7	3
57	GRIN2A: involvement in movement disorders and intellectual disability without seizures. <i>Neurological Sciences</i> , 2019, 40, 2405-2406.	1.9	3
58	TBC1D24 gene mRNA expression in a boy with early infantile epileptic encephalopathy-16. <i>Acta Neurologica Belgica</i> , 2020, 120, 381-383.	1.1	3
59	Juvenile myoclonic epilepsy with generalised and focal electroencephalographic abnormalities: a case report with a molecular genetic study. <i>Neurological Sciences</i> , 2007, 28, 276-278.	1.9	2
60	Are Mutations in the DHRS9 Gene Causally Linked to Epilepsy? A Case Report. <i>Medicina (Lithuania)</i> , 2020, 56, 387.	2.0	2
61	Novel compound heterozygous mutation in NPC1 gene cause Niemann-Pick disease type C with juvenile onset. <i>Journal of Genetics</i> , 2020, 99, 1.	0.7	2
62	Towards a genetic history of Sicily. <i>Journal of Cultural Heritage</i> , 2000, 1, S39-S42.	3.3	1
63	Mutations and polymorphisms of the PAH gene in Sicily: comparison with other DNA polymorphisms. <i>Journal of Cultural Heritage</i> , 2000, 1, S43-S45.	3.3	1
64	Comparative multiplex dosage analysis in spinocerebellar ataxia type 2 patients. <i>Genetics and Molecular Research</i> , 2013, 12, 1176-1181.	0.2	1
65	A Novel Homozygous <i>ALG12</i> Mutation in a Patient with CDG Type Ig: New Report of a Case with a Mild Phenotype. <i>Molecular Syndromology</i> , 2021, 12, 327-332.	0.8	1
66	Novel SPINK5 variants in a patient with Netherton syndrome and intellectual disability. The diagnostic value of trichoscopy. <i>Giornale Italiano Di Dermatologia E Venereologia</i> , 2020, 155, 239-240.	0.8	1
67	KCNQ2-Related Neonatal Epilepsy Treated With Vitamin B6: A Report of Two Cases and Literature Review. <i>Frontiers in Neurology</i> , 2022, 13, 826225.	2.4	1
68	An interesting case of Piebaldism with café-au-lait macules and freckling: the use of targeted next-generation sequencing for molecular diagnosis. <i>European Journal of Dermatology</i> , 2018, 28, 119-120.	0.6	1
69	Exome sequencing in a child with neurodevelopmental disorder and epilepsy: Variant analysis of the <i>AHNAK2</i> gene. <i>Molecular Genetics &amp; Genomic Medicine</i> , 0, , .	1.2	1
70	Two novel PAH gene mutations detected in Italian phenylketonuric patients. <i>Human Genetics</i> , 1997, 99, 275-278.	3.8	0
71	Boolean Networks: A Primer. , 2021, , 41-53.		0
72	Pigmented porokeratosis with dermal deposits of amyloid: the different chromatic features. <i>Giornale Italiano Di Dermatologia E Venereologia</i> , 2020, 155, 240-241.	0.8	0

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73	Letter to the Editor Regarding the Article "Whole-Exome Sequencing in NF1-Related West's Syndrome Leads to the Identification of KCNC2 as a Novel Candidate Gene for Epilepsy" Neuropediatrics, 2021, 52, 153-153.	0.6	0
74	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
75	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
76	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
77	An odd precocious case of progressive osseous heteroplasia. Italian Journal of Dermatology and Venereology, 2021, 156, 409-411.	0.2	0
78	Eight new mutations of the phenylalanine hydroxylase gene in Italian patients with hyperphenylalaninemia. Human Mutation, 1998, 11, 240-243.	2.5	0