Antonio Percesepe

List of Publications by Year
in descending order

97 papers	4,045 citations	201385 27 h-index	128067 60 g-index
101 all docs	101 docs citations	101 times ranked	4801 citing authors

#	Article	IF	Citations
1	Recurrent NF1 gene variants and their genotype/phenotype correlations in patients with Neurofibromatosis type I. Genes Chromosomes and Cancer, 2022, 61, 10-21.	1.5	6
2	The effect of carriersâ€™ reproductive choices and pregnancy history on sporadic severe haemophilia: A 20â€year retrospective study through a regional registry. Haemophilia, 2022, 28, 308-315.	1.0	3
3	Clinical and Genetic Findings in a Series of Eight Families with Arthrogryposis. Genes, 2022, 13, 29.	1.0	6
4	BCR-ABL1 compound mutants: prevalence, spectrum and correlation with tyrosine kinase inhibitor resistance in a consecutive series of Philadelphia chromosome-positive leukemia patients analyzed by NGS. Leukemia, 2021, 35, 2102-2107.	3.3	8
5	Liquid biopsy with cell free DNA: new horizons for prostate cancer. Critical Reviews in Clinical Laboratory Sciences, 2021, 58, 60-76.	2.7	27
6	Genetic Neonatal-Onset Epilepsies and Developmental/Epileptic Encephalopathies with Movement Disorders: A Systematic Review. International Journal of Molecular Sciences, 2021, 22, 4202.	1.8	10
7	Impact of the rs1024611 Polymorphism of CCL2 on the Pathophysiology and Outcome of Primary Myelofibrosis. Cancers, 2021, 13, 2552.	1.7	9
8	Filling the gap: A thorough investigation for the genetic diagnosis of unsolved polyposis patients with monoallelic MUTYH pathogenic variants. Molecular Genetics & Genomic Medicine, 2021, 9, e1831.	0.6	3
9	Deciphering the pathogenesis of the COL4â€related hematuric nephritis: A genotype/phenotype study. Molecular Genetics & Genomic Medicine, 2021, 9, e1576.	0.6	2
10	Results and Clinical Interpretation of Germline RET Analysis in a Series of Patients with Medullary Thyroid Carcinoma: The Challenge of the Variants of Uncertain Significance. Cancers, 2020, 12, 3268.	1.7	2
11	Unmasking selective path integration deficits in Alzheimerâ€™s disease risk carriers. Science Advances, 2020, 6, eaba1394.	4.7	55
12	FCGR3B polymorphism predicts relapse risk in eosinophilic granulomatosis with polyangiitis. Rheumatology, 2020, 59, 3563-3566.	0.9	8
13	Prospective assessment of NGS-detectable mutations in CML patients with nonoptimal response: the NEXT-in-CML study. Blood, 2020, 135, 534-541.	0.6	61
14	Melorheostosis and Osteopoikilosis Clinical and Molecular Description of an Italian Case Series. Calcified Tissue International, 2019, 105, 215-221.	1.5	7
15	Challenges in the clinical interpretation of small de novo copy number variants in neurodevelopmental disorders. Gene, 2019, 706, 162-171.	1.0	9
16	Spectrum of X-linked intellectual disabilities and psychiatric symptoms in a family harbouring a Xp22.12 microduplication encompassing the RPS6KA3 gene. Journal of Genetics, 2019, 98, 1.	0.4	5
17	Detection of Actionable BCR-ABL1 Kinase Domain (KD) Mutations in Chronic Myeloid Leukemia (CML) Patients with Failure and Warning Response to Tyrosine Kinase Inhibitors (TKIs): Potential Impact of Next-Generation Sequencing (NGS) and Droplet Digital PCR (ddPCR) on Clinical Decision Making. Blood, 2019, 134, 661-661.	0.6	5
18	Relationship between atopy, asthma and alpha-1 antitrypsin deficiency. , 2019, , .		0

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19	Homozygosity for -2518 G Allele Variant of MCP-1 Predisposes to Adverse Presentation and Outcome in Primary Myelofibrosis. Blood, 2019, 134, 1689-1689.	0.6	0
20	Spectrum of X-linked intellectual disabilities and psychiatric symptoms in a family harbouring a Xp22.12 microduplication encompassing the gene. Journal of Genetics, 2019, 98, .	0.4	0
21	Genetic diagnosis in neonatal-onset epilepsies: Back to the future. European Journal of Paediatric Neurology, 2018, 22, 354-357.	0.7	14
22	The -2518 A/G polymorphism of the monocyte chemoattractant protein-1 as a candidate genetic predisposition factor for secondary myelofibrosis and biomarker of disease severity. Leukemia, 2018, 32, 2266-2270.	3.3	16
23	Gain-of-function mutations in DNMT3A in patients with paraganglioma. Genetics in Medicine, 2018, 20, 1644-1651.	1.1	73
24	Next Generation Sequencing-Based BCR-ABL1 Kinase Domain Mutation Screening in De Novo and Tyrosine Kinase Inhibitor-Resistant Philadelphia Chromosome-Positive Acute Lymphoblastic Leukemia: Results of a Prospective Study. Blood, 2018, 132, 4078-4078.	0.6	1
25	Compound BCR-ABL1 Kinase Domain Mutants: Prevalence, Spectrum and Correlation with Tyrosine Kinase Inhibitor Resistance in a Prospective Series of Philadelphia Chromosome-Positive Leukemia Patients Analyzed By Next Generation Sequencing. Blood, 2018, 132, 789-789.	0.6	3
26	The genetic and clinical spectrum of a large cohort of patients with distal renal tubular acidosis. Kidney International, 2017, 91, 1243-1255.	2.6	79
27	Self-Amputation of the Extra Digit in a Fetus with Polydactyly: First Ultrasound Demonstration. Fetal Diagnosis and Therapy, 2017, 41, 314-316.	0.6	1
28	MicroRNA Expression in Malignant Pleural Mesothelioma and Asbestosis: A Pilot Study. Disease Markers, 2017, 2017, 1-10.	0.6	37
29	Patterns of Novel Alleles and Genotype/Phenotype Correlations Resulting from the Analysis of 108 Previously Undetected Mutations in Patients Affected by Neurofibromatosis Type I. International Journal of Molecular Sciences, 2017, 18, 2071.	1.8	11
30	Monogenic Autoinflammatory Diseases with Mendelian Inheritance: Genes, Mutations, and Genotype/Phenotype Correlations. Frontiers in Immunology, 2017, 8, 344.	2.2	37
31	Unravelling the Complexity of Inherited Retinal Dystrophies Molecular Testing: Added Value of Targeted Next-Generation Sequencing. BioMed Research International, 2016, 2016, 1-14.	0.9	47
32	Reverse phenotyping comes of age. Molecular Genetics and Metabolism, 2016, 118, 230-231.	0.5	9
33	Mutation screening of the Otop1 gene in familial benign positional paroxysmal vertigo. Hearing, Balance and Communication, 2016, 14, 1-7.	0.1	0
34	Pre- and post-natal growth in two sisters with 3-M syndrome. European Journal of Medical Genetics, 2016, 59, 232-236.	0.7	13
35	Prenatal diagnosis and followâ€up of a case of branchioâ€otoâ€renal syndrome displays renal growth impairment after the second trimester. Journal of Obstetrics and Gynaecology Research, 2015, 41, 1831-1834.	0.6	4
36	Early diagnosis of branchio-oculo-facial syndrome in a patient with inner ear malformation and mild ocular involvement. Clinical Dysmorphology, 2015, 24, 17-20.	0.1	1

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37	Exome sequencing in a patient with Catelâ€“Manzke-like syndrome excludes the involvement of the known genes and reveals a possible candidate. European Journal of Medical Genetics, 2015, 58, 597-602.	0.7	3
38	Involvement of MBD4 inactivation in mismatch repair-deficient tumorigenesis. Oncotarget, 2015, 6, 42892-42904.	0.8	43
39	Role of Genetic Factors in the Pathogenesis of Radial Deficiencies in Humans. Current Genomics, 2015, 16, 264-278.	0.7	16
40	Hypomelanosis of Ito with a trisomy 2 mosaicism: a case report. Journal of Medical Case Reports, 2014, 8, 333.	0.4	8
41	New and Rare GJB2 Alleles in Patients with Nonsyndromic Sensorineural Hearing Impairment: A Genotype/Auditory Phenotype Correlation. Genetic Testing and Molecular Biomarkers, 2014, 18, 839-844.	0.3	5
42	Mutations Affecting the BHLHA9 DNA-Binding Domain Cause MSSD, Mesoaxial Synostotic Syndactyly with Phalangeal Reduction, Malik-Percin Type. American Journal of Human Genetics, 2014, 95, 649-659.	2.6	27
43	A syndromic form of Pierre Robin sequence is caused by 5q23 deletions encompassing FBN2 and PHAX. European Journal of Medical Genetics, 2014, 57, 587-595.	0.7	18
44	Predictive diagnostic value for the clinical features accompanying intellectual disability in children with pathogenic copy number variations: a multivariate analysis. Italian Journal of Pediatrics, 2014, 40, 39.	1.0	10
45	VACTERL (Vertebral Defects, Anal Atresia, Tracheoesophageal Fistula withÂEsophageal Atresia, Cardiac) Tj ETQq1 1 0.784314 rgBT /O UpperÂLimb Involvement. Journal of Pediatrics, 2014, 164, 458-462.e2.	0.9	21
46	A three-generation family with terminal microdeletion involving 5p15.33â€“32 due to a whole-arm 5;15 chromosomal translocation with a steady phenotype of atypical cri du chat syndrome. European Journal of Medical Genetics, 2014, 57, 145-150.	0.7	27
47	Holoprosencephaly: report of four cases and genotypeâ€“phenotype correlations. Journal of Genetics, 2013, 92, 97-101.	0.4	12
48	Biological parameters determining the clinical outcome of autologous cultures of limbal stem cells. Regenerative Medicine, 2013, 8, 553-567.	0.8	117
49	Structural chromosomal abnormalities detected during CVS analysis and their role in the prenatal ascertainment of cryptic subtelomeric rearrangements. American Journal of Medical Genetics, Part A, 2013, 161, 2559-2563.	0.7	0
50	Surface Rendering of External Genitalia of a Fetus at the 32nd Week of Gestation Affected by Partial Androgen Insensitivity Syndrome. Case Reports in Obstetrics and Gynecology, 2013, 2013, 1-3.	0.2	3
51	Genetic Basis of Congenital Upper Limb Anomalies: Analysis of 487 Cases of a Specialized Clinic. Birth Defects Research Part A: Clinical and Molecular Teratology, 2013, 97, 798-805.	1.6	15
52	Facioscapulohumeral muscular dystrophy: new insights from compound heterozygotes and implication for prenatal genetic counselling. Journal of Medical Genetics, 2012, 49, 171-178.	1.5	53
53	A 12.4ÂMb direct duplication in 19q12-q13 in a boy with cardiac and CNS malformations and developmental delay. Journal of Applied Genetics, 2011, 52, 335-339.	1.0	7
54	Molecular Genetic Analysis of the PLP1 Gene in 38 Families with PLP1-related disorders: Identification and Functional Characterization of 11 Novel PLP1 Mutations. Orphanet Journal of Rare Diseases, 2011, 6, 40.	1.2	32

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55	Firstâ€trimester ultrasonographic diagnosis of Langer mesomelic dysplasia in a previouslyaffected family. Prenatal Diagnosis, 2011, 31, 830-831.	1.1	0
56	Second Trimester Amniocentesis Is Not a Risk Factor for Very Low Birth Weight and Extremely Low Birth Weight. ISRN Obstetrics & Gynecology, 2011, 2011, 1-3.	1.2	1
57	Prenatal diagnosis and postnatal followâ€up of a child with mosaic trisomy 22 with several levels of mosaicism in different tissues. Journal of Obstetrics and Gynaecology Research, 2010, 36, 1116-1120.	0.6	15
58	Incidence of non-age-dependent chromosomal abnormalities: a population-based study on 88965 amniocenteses. European Journal of Human Genetics, 2009, 17, 897-903.	1.4	82
59	Familial Beckwithâ€“Wiedemann syndrome due to CDKN1C mutation manifesting with recurring omphalocele. Prenatal Diagnosis, 2008, 28, 447-449.	1.1	7
60	Pure segmental trisomy 1q42â€qter in a boy with a severe phenotype. American Journal of Medical Genetics, Part A, 2007, 143A, 2339-2342.	0.7	12
61	Age-specific risk of fetal loss post second trimester amniocentesis: analysis of 5043 cases. Prenatal Diagnosis, 2007, 27, 180-183.	1.1	89
62	The homozygous deletion of the 3â€² enhancer of the SHOX gene causes Langer mesomelic dysplasia. Clinical Genetics, 2007, 72, 490-491.	1.0	22
63	Quantitation of fetal DNA in maternal serum during the first trimester of pregnancy by the use of a DAZ repetitive probe. Molecular Human Reproduction, 2006, 12, 587-591.	1.3	34
64	Detection of a novel dystrophin gene mutation through carrier analysis performed during prenatal diagnosis in a case with intragenic recombination. Prenatal Diagnosis, 2005, 25, 1011-1014.	1.1	7
65	First-trimester prenatal screening for the common 35delG GJB2 mutation causing prelingual deafness. Prenatal Diagnosis, 2004, 24, 631-634.	1.1	8
66	Multimodal Molecular Screening Is Required to Improve the Sensitivity of MLH1 and MSH2 Mutation Analysis. Journal of Clinical Oncology, 2002, 20, 1705-1707.	0.8	1
67	Genes and translocations involved in POF. American Journal of Medical Genetics Part A, 2002, 111, 328-333.	2.4	146
68	Non-invasive first trimester fetal gender assignment in pregnancies at risk for X-linked recessive diseases. Prenatal Diagnosis, 2002, 22, 919-924.	1.1	25
69	Molecular Screening for Hereditary Nonpolyposis Colorectal Cancer: A Prospective, Population-Based Study. Journal of Clinical Oncology, 2001, 19, 3944-3950.	0.8	101
70	Phenotype-genotype correlations in an extended family with adenomatosis coli and an unusual APC gene mutation. Diseases of the Colon and Rectum, 2001, 44, 1597-1604.	0.7	9
71	Methylation pattern of different regions of theMLH1 promoter and silencing of gene expression in hereditary and sporadic colorectal cancer. Genes Chromosomes and Cancer, 2001, 31, 357-361.	1.5	53
72	Mutations of the 'minor' mismatch repair gene MSH6 in typical and atypical hereditary nonpolyposis colorectal cancer. Familial Cancer, 2001, 1, 95-101.	0.9	24

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73	Clinical and biologic heterogeneity of hereditary nonpolyposis colorectal cancer. International Journal of Cancer, 2001, 95, 323-328.	2.3	19
74	Genomic instability and target gene mutations in colon cancers with different degrees of allelic shifts. , 2000, 27, 424-429.		19
75	Problems in the identification of hereditary nonpolyposis colorectal cancer in two families with late development of full-blown clinical spectrum. American Journal of Gastroenterology, 2000, 95, 2110-2115.	0.2	3
76	Problems in the identification of hereditary nonpolyposis colorectal cancer in two families with late development of full-blown clinical spectrum. American Journal of Gastroenterology, 2000, 95, 2110-2115.	0.2	2
77	Genomic instability and target gene mutations in colon cancers with different degrees of allelic shifts. , 2000, 27, 424.		1
78	The DNA repair gene MBD4 (MED1) is mutated in human carcinomas with microsatellite instability. Nature Genetics, 1999, 23, 266-268.	9.4	211
79	Microsatellite instability in multiple colorectal tumors. , 1999, 81, 1-5.		72
80	The I1307K polymorphism of the APC gene in colorectal cancer. Gastroenterology, 1999, 116, 58-63.	0.6	65
81	Mismatch repair genes and mononucleotide tracts as mutation targets in colorectal tumors with different degrees of microsatellite instability. Oncogene, 1998, 17, 157-163.	2.6	68
82	MLH1 and MSH2 constitutinal mutations in colorectal cancer families not meeting the standard criteria for hereditary nonpolyposis colorectal cancer. , 1998, 75, 835-839.		50
83	Incidence of Hereditary Nonpolyposis Colorectal Cancer and the Feasibility of Molecular Screening for the Disease. New England Journal of Medicine, 1998, 338, 1481-1487.	13.9	1,048
84	Small bowel carcinoma in hereditary nonpolyposis colorectal cancer. American Journal of Gastroenterology, 1998, 93, 2219-2222.	0.2	15
85	Hereditary Nonpolyposis Colorectal Cancer: An Approach to the Selection of Candidates to Genetic TestingBased on Clinical and MolecularCharacteristics. Public Health Genomics, 1998, 1, 229-236.	0.6	10
86	Characterization ofMSH2 andMLH1 mutations in Italian families with hereditary nonpolyposis colorectal cancer. , 1997, 18, 8-18.		67
87	K-ras andp53 mutations in hereditary non-polyposis colorectal cancers. International Journal of Cancer, 1997, 74, 94-96.	2.3	80
88	Survival analysis in families affected by hereditary non-polyposis colorectal cancer. , 1997, 71, 373-376.		50
89	Clinical features, frequency and prognosis of Dukes' A colorectal carcinoma: A population-based investigation. European Journal of Cancer, 1996, 32, 1957-1962.	1.3	15
90	Collection of Italian Hereditary Non-Polyposis Colorectal Cancer (HNPCC) Pedigrees. Tumori, 1996, 82, 151-179.	0.6	2

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91	Frequency and clinical features of multiple tumors of the large bowel in the general population and in patients with hereditary colorectal carcinoma. , 1996, 77, 2013-2021.		61
92	DNA ploidy pattern in human chronic liver diseases and hepatic nodular lesions. Flow cytometric analysis on echo-guided needle liver biopsy. Cancer, 1994, 73, 281-288.	2.0	44
93	Role of clinical criteria in the diagnosis of hereditary non-polyposis colorectal cancer (HNPCC): Results of a multivariate analysis. International Journal of Cancer, 1994, 58, 799-802.	2.3	19
94	Circadian variations of epithelial cell proliferation in human rectal crypts. Gastroenterology, 1994, 106, 982-987.	0.6	63
95	Effects of different doses of fish oil on rectal cell proliferation in patients with sporadic colonic adenomas. Gastroenterology, 1994, 107, 1709-1718.	0.6	180
96	CowdenÊ¼s Disease with Extensive Gastrointestinal Polyposis. Journal of Clinical Gastroenterology, 1994, 18, 42-47.	1.1	52
97	Effect of Ï‰-3 fatty acids on rectal mucosal cell proliferation in subjects at risk for colon cancer. Gastroenterology, 1992, 103, 883-891.	0.6	199