Berardino Porfirio

List of Publications by Year
in descending order

66	2,015	279798	243625
papers	citations	23 h-index	44 g-index

68	68	68	2955
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Normal tension glaucoma in CSF-shunted normal pressure hydrocephalus patients. An extended follow-up. Eye, 2022, , .	2.1	0
2	Aqueductal CSF stroke volume measurements may drive management of shunted idiopathic normal pressure hydrocephalus patients. Scientific Reports, 2021, 11, 7095.	3.3	2
3	Characterization of the Cancer Spectrum in Men With GermlineBRCA1andBRCA2Pathogenic Variants. JAMA Oncology, 2020, 6, 1218.	7.1	48
4	Large scale multifactorial likelihood quantitative analysis of BRCA1 and BRCA2 variants: An ENIGMA resource to support clinical variant classification. Human Mutation, 2019, 40, 1557-1578.	2.5	102
5	Evaluation of a Next-Generation Sequencing Assay for BRCA1 and BRCA2 Mutation Detection. Journal of Molecular Diagnostics, 2018, 20, 87-94.	2.8	11
6	Glaucoma in patients with shunt-treated normal pressure hydrocephalus. Journal of Neurosurgery, 2018, 129, 1078-1084.	1.6	30
7	A Comment on â€œJuvenile-onset Normal Tension Glaucoma From Chronic, Recurrent Low Cerebrospinal Fluid Pressure.â€J Glaucoma. 2016;25:e738â€“e740. Journal of Glaucoma, 2017, 26, e132-e133.	1.6	1
8	A Founder Effect for the HGD G360R Mutation in Italy: Implications for a Regional Screening of Alkaptonuria. JIMD Reports, 2016, 30, 45-52.	1.5	1
9	Re: Fast circulation of cerebrospinal fluid: an alternative perspective on the protective role of high intracranial pressure in ocular hypertension. Australasian journal of optometry, The, 2016, 99, 607-607.	1.3	1
10	A Comment on the â€œProgression of Normal-tension Glaucoma After Ventriculoperitoneal Shunt to Decrease Cerebrospinal Fluid Pressureâ€• J Glaucoma. 2016;25. Journal of Glaucoma, 2016, Publish Ahead of Print, .	1.6	0
11	Twelve novel HGD gene variants identified in 99 alkaptonuria patients: focus on â€˜black bone diseaseâ€™ in Italy. European Journal of Human Genetics, 2016, 24, 66-72.	2.8	87
12	Donor-Specific Anti-HLA Antibodies in Huntington's Disease Recipients of Human Fetal Striatal Grafts. Cell Transplantation, 2015, 24, 811-817.	2.5	12
13	AÎ² Clearance, â€œhubâ€•of Multiple Deficiencies Leading to Alzheimer Disease. Frontiers in Aging Neuroscience, 2015, 7, 200.	3.4	18
14	A commentary on â€œDifferentiation of pluripotent stem cells into striatal projection neurons: a pure MSN fate may not be sufficientâ€• Frontiers in Cellular Neuroscience, 2015, 9, 177.	3.7	0
15	Fetal striatal grafting slows motor and cognitive decline of Huntington's disease. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 974-981.	1.9	34
16	Human Striatum Remodelling after Neurotransplantation in Huntington's Disease. Stereotactic and Functional Neurosurgery, 2014, 92, 211-217.	1.5	10
17	Multifaceted roles of BDNF and FGF2 in human striatal primordium development. An in vitro study. Experimental Neurology, 2014, 257, 130-147.	4.1	23
18	High resolution melting analysis of deletion/insertion polymorphisms: A new method for the detection and quantification of mixed chimerism in allogeneic stem cell transplantation. Molecular and Cellular Probes, 2014, 28, 19-24.	2.1	5

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19	Large-sized Fetal Striatal Grafts in Huntingtonâ€™s Disease Do Stop Growing. Long-term Monitoring in the Florence Experience. PLOS Currents, 2014, 6, .	1.4	3
20	Tag SNPs of the ancestral haplotype 57.1 do not substitute HLA-B*57:01 typing for eligibility to abacavir treatment in the Italian population. Pharmacogenomics, 2012, 13, 247-249.	1.3	17
21	Workshop report: extending the number of resources and bioinformatics analysis for the investigation of HLA rare alleles. International Journal of Immunogenetics, 2012, 40, n/a-n/a.	1.8	16
22	The conundrum of HLA-DRB1*14:01/*14:54 and HLA-DRB3*02:01/*02:02 mismatches in unrelated hematopoietic SCT. Bone Marrow Transplantation, 2011, 46, 916-922.	2.4	25
23	Schwannomatosis associated with multiple meningiomas due to a familial SMARCB1 mutation. Neurogenetics, 2010, 11, 73-80.	1.4	90
24	Human striatal neuroblasts develop and build a striatal-like structure into the brain of Huntington's disease patients after transplantation. Experimental Neurology, 2010, 222, 30-41.	4.1	74
25	Functional polymorphisms of the microsomal epoxide hydrolase gene: A reappraisal on a early-onset lung cancer patients series. Lung Cancer, 2009, 63, 187-193.	2.0	22
26	The Metabonomic Signature of Celiac Disease. Journal of Proteome Research, 2009, 8, 170-177.	3.7	160
27	Nonpermissive HLA-DPB1 disparity is a significant independent risk factor for mortality after unrelated hematopoietic stem cell transplantation. Blood, 2009, 114, 1437-1444.	1.4	131
28	Fatal hypertrophic cardiomyopathy and nemaline myopathy associated with ACTA1 K336E mutation. Journal of Molecular and Cellular Cardiology, 2007, 42, S73.	1.9	1
29	Genetic STRs variation in a large population from Tuscany (Italy). Forensic Science International: Genetics, 2007, 1, e10-e11.	3.1	7
30	Fatal hypertrophic cardiomyopathy and nemaline myopathy associated with ACTA1 K336E mutation. Neuromuscular Disorders, 2006, 16, 548-552.	0.6	83
31	Radiation-induced cutaneous carcinoma of the head and neck: is there an early role for p53 mutations?. Clinical and Experimental Dermatology, 2006, 31, 793-798.	1.3	13
32	De novoÎ±-actin mutations in monozygotic twins. Clinical Genetics, 2005, 68, 91-92.	2.0	3
33	Sequence diversity within the HA-1 gene as detected by melting temperature assay without oligonucleotide probes. BMC Medical Genetics, 2005, 6, 36.	2.1	5
34	Lack of association between the HLA-DRB1 locus and post-streptococcal reactive arthritis and acute rheumatic fever in italian children. Seminars in Arthritis and Rheumatism, 2004, 34, 553-558.	3.4	21
35	Shared-epitope HLA-DRB1 alleles and sex ratio in Italian patients with rheumatoid arthritis. Joint Bone Spine, 2004, 71, 24-28.	1.6	26
36	Alkaptonuria, ochronosis, and ochronotic arthropathy. Seminars in Arthritis and Rheumatism, 2004, 33, 239-248.	3.4	94

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37	Alpha-actin gene mutations and polymorphisms in Italian patients with nemaline myopathy. International Journal of Molecular Medicine, 2004, 13, 805-9.	4.0	15
38	Influence of Calcium-Sensing Receptor Gene on Urinary Calcium Excretion in Stone-Forming Patients. Journal of the American Society of Nephrology: JASN, 2002, 13, 2517-2523.	6.1	135
39	Alkaptonuria in Italy: polymorphic haplotype background, mutational profile, and description of four novel mutations in the homogentisate 1,2-dioxygenase gene. Journal of Medical Genetics, 2000, 37, 309-312.	3.2	22
40	Cumulative prognostic value ofp53 mutations and bcl-2 protein expression in head-and-neck cancer treated by radiotherapy. International Journal of Cancer, 1999, 84, 573-579.	5.1	66
41	Epstein-Barr virus infection and P53 expression in HIV-related oral large B cell lymphoma. , 1999, 21, 454-460.		15
42	Clinical and genetic heterogeneity in autosomal recessive nemaline myopathy. Neuromuscular Disorders, 1999, 9, 564-572.	0.6	84
43	Mutation and Polymorphism Analysis of the Human Homogentisate 1,2-Dioxygenase Gene in Alkaptonuria Patients. American Journal of Human Genetics, 1998, 62, 776-784.	6.2	79
44	A method for point mutation analysis that links SSCP and dye primer fluorescent sequencing. Molecular and Cellular Probes, 1998, 12, 125-131.	2.1	4
45	Potential Biomarkers in Predicting Progression of Epithelial Hyperplastic Lesions of the Larynx. Acta Oto-Laryngologica, 1997, 117, 30-38.	0.9	20
46	Immunohistochemical vs Molecular Biology Methods:Complementary Techniques for Effective Screening of p53 Alterations in Head and Neck Cancer. American Journal of Clinical Pathology, 1997, 107, 7-11.	0.7	43
47	HLA-B44 subtypes and the chance of finding HLA compatible donor/recipient pairs for bone marrow transplantation: a haplotype study of 303 Italian families. Tissue Antigens, 1997, 50, 602-609.	1.0	10
48	GENOTYPE-RELATED FINGERPRINTS FROM HLA-DPB1 EXON 2 LOW-STRINGENCY PCR. International Journal of Immunogenetics, 1996, 23, 451-457.	1.2	1
49	Hypersensitivity of lymphoblastoid lines derived from ataxia telangiectasia patients to the induction of chromosomal aberrations by etoposide (VP-16). Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 1993, 290, 265-272.	1.0	21
50	Expression of aphidicolin-induced fragile sites in lymphocytes of patients with breast cancer. Cancer Genetics and Cytogenetics, 1993, 67, 113-116.	1.0	24
51	Structural chromosomal rearrangements in HpaII-treated human lymphocytes. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 1991, 248, 115-121.	1.0	13
52	The distribution of Msp I-induced breaks in human lymphocyte chromosomes and its relationship to common fragile sites. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 1989, 213, 117-124.	1.0	16
53	Partial correction of chromosome instability in Fanconi anemia by desferrioxamine. Human Genetics, 1989, 83, 49-51.	3.8	20
54	Patients with different lung cancers show normal expression of fra(3)(p14.2) in aphidicolin-treated lymphocyte cultures. Cancer Genetics and Cytogenetics, 1989, 43, 95-101.	1.0	12

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55	128 Patients with different lung cancers show normal expression of aphidicolin induced FRA(3p14.2) in their normal cultured lymphocytes. Cancer Genetics and Cytogenetics, 1989, 38, 202.	1.0	0
56	Chromosome breakage induced by bleomycin in an ataxia telangiectasia lymphoblastoid line: correlation with fragile sites and Epstein-Barr virus DNA localization. Cytogenetic and Genome Research, 1989, 52, 180-185.	1.1	5
57	Sister-chromatid exchanges in human lymphocytes exposed to 1-p-(3-methyltriazeno)benzoic acid potassium salt. Mutation Research-Fundamental and Molecular Mechanisms of Mutagenesis, 1988, 208, 233-236.	1.1	4
58	Specific sites for EBV association in the Namalwa Burkitt lymphoma cell line and in a lymphoblastoid line transformed in vitro with EBV. Cytogenetic and Genome Research, 1988, 48, 220-223.	1.1	10
59	Ring 20 chromosome phenotype.. Journal of Medical Genetics, 1987, 24, 375-377.	3.2	30
60	Cytogenetic effects of 1-p-(3-methyltriazeno)benzoic acid potassium salt on human lymphocytes in vitro. Mutation Research - Genetic Toxicology Testing and Biomonitoring of Environmental Or Occupational Exposure, 1987, 189, 349-356.	1.2	7
61	Breakpoint distribution in constitutional chromosome rearrangements with respect to fragile sites. Annals of Human Genetics, 1987, 51, 329-336.	0.8	13
62	Common fragile sites: Their prevalence in subjects with constitutional and acquired chromosomal instability. American Journal of Medical Genetics Part A, 1987, 27, 471-482.	2.4	63
63	Effect of oxidants and antioxidants on chromosomal breakage in Fanconi anemia lymphocytes. Human Genetics, 1985, 69, 62-65.	3.8	69
64	The effect of aphidicolin on Fanconi's anemia lymphocyte chromosomes. Mutation Research-Fundamental and Molecular Mechanisms of Mutagenesis, 1985, 144, 257-263.	1.1	7
65	A live infant with trisomy 14 mosaicism and nuclear abnormalities of the neutrophils.. Journal of Medical Genetics, 1984, 21, 467-470.	3.2	13
66	Failure of diepoxybutane to enhance sister chromatid exchange levels in Fanconi's anemia patients and heterozygotes. Human Genetics, 1983, 63, 117-120.	3.8	18