Paola Concolino

List of Publications by Year
in descending order

68	1,143	430874 18	454955 30
papers	citations	h-index	g-index

69	69	69	1539
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Multiple endocrine neoplasia type 1 (MEN1): An update of 208 new germline variants reported in the last nine years. Cancer Genetics, 2016, 209, 36-41.	0.4	118
2	Multiplex ligation-dependent probe amplification (MLPA) assay for the detection of CYP21A2 gene deletions/duplications in Congenital Adrenal Hyperplasia: First technical report. Clinica Chimica Acta, 2009, 402, 164-170.	1.1	58
3	Serum levels of seven cytokines in premature ventilated newborns: correlations with old and new forms of bronchopulmonary dysplasia. Intensive Care Medicine, 2006, 32, 723-730.	8.2	54
4	Functional effect of Saffron supplementation and risk genotypes in early age-related macular degeneration: a preliminary report. Journal of Translational Medicine, 2013, 11, 228.	4.4	49
5	Molecular diagnosis of congenital adrenal hyperplasia due to 21-hydroxylase deficiency: an update of new CYP21A2 mutations. Clinical Chemistry and Laboratory Medicine, 2010, 48, 1057-1062.	2.3	44
6	A new CYP21A1P/CYP21A2chimeric gene identified in an Italian woman suffering from classical congenital adrenal hyperplasia form. BMC Medical Genetics, 2009, 10, 72.	2.1	40
7	GSTM1-null polymorphism as possible risk marker for hypertension: Results from the aging and longevity study in the Sirente Geographic Area (ilSIRENTE study). Clinica Chimica Acta, 2009, 399, 92-96.	1.1	38
8	Vitamin D Receptor Polymorphisms and Falls Among Older Adults Living in the Community: Results From the ilSIRENTE Study. Journal of Bone and Mineral Research, 2008, 23, 1031-1036.	2.8	31
9	Clinical impact on ovarian cancer patients of massive parallel sequencing forBRCAmutation detection: the experience at Gemelli hospital and a literature review. Expert Review of Molecular Diagnostics, 2015, 15, 1383-1403.	3.1	30
10	Rapid UGT1A1 (TA)n genotyping by high resolution melting curve analysis for Gilbert's syndrome diagnosis. Clinica Chimica Acta, 2010, 411, 246-249.	1.1	29
11	A comprehensive BRCA1/2 NGS pipeline for an immediate Copy Number Variation (CNV) detection in breast and ovarian cancer molecular diagnosis. Clinica Chimica Acta, 2018, 480, 173-179.	1.1	28
12	Epithelial lining fluid free IGF-I-to-PAPP-A ratio is associated with bronchopulmonary dysplasia in preterm infants. American Journal of Physiology - Endocrinology and Metabolism, 2007, 292, E308-E313.	3.5	26
13	Mannose-binding lectin polymorphisms and pulmonary outcome in premature neonates: aÂpilot study. Intensive Care Medicine, 2007, 33, 1787-1794.	8.2	26
14	Identification of RFLP G6PD mutations by using microcapillary electrophoretic chips (Experion^{TM}). Journal of Separation Science, 2008, 31, 2694-2700.	2.5	26
15	Advanced tools for BRCA1/2 mutational screening: comparison between two methods for large genomic rearrangements (LGRs) detection. Clinical Chemistry and Laboratory Medicine, 2014, 52, 1119-27.	2.3	26
16	Association of periodontitis with GSTM1/GSTT1-null variantsâ€"A pilot study. Clinical Biochemistry, 2007, 40, 939-945.	1.9	24
17	Comparison between three molecular methods for detection of blood melanoma tyrosinase mRNA. Correlation with melanoma stages and S100B, LDH, NSE biochemical markers. Clinica Chimica Acta, 2005, 362, 85-93.	1.1	23
18	A preliminary Quality Control (QC) for next generation sequencing (NGS) library evaluation turns out to be a very useful tool for a rapid detection of BRCA1/2 deleterious mutations. Clinica Chimica Acta, 2014, 437, 72-77.	1.1	20

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19	Glucose-6-phosphate dehydrogenase Buenos Aires: A novel de novo missense mutation associated with severe enzyme deficiency. Clinical Biochemistry, 2008, 41, 742-745.	1.9	19
20	Differentiated Thyroid Cancer in Two Patients with Resistance to Thyroid Hormone. Thyroid, 2011, 21, 793-797.	4.5	18
21	Multiplex Ligation-Dependent Probe Amplification Analysis Is Useful for Diagnosing Congenital Adrenal Hyperplasia but Requires a Deep Knowledge of CYP21A2 Genetics. Clinical Chemistry, 2011, 57, 1079-1080.	3.2	18
22	Insight into a Novel p53 Single Point Mutation (G389E) by Molecular Dynamics Simulations. International Journal of Molecular Sciences, 2011, 12, 128-140.	4.1	18
23	Droplet digital PCR for large genomic rearrangements detection: A promising strategy in tissue BRCA1 testing. Clinica Chimica Acta, 2021, 513, 17-24.	1.1	18
24	Genetic analysis of the dystroglycan gene in bronchopulmonary dysplasia affected premature newborns. Clinica Chimica Acta, 2007, 378, 164-167.	1.1	17
25	Reverse-hybridization assay for rapid detection of common CYP21A2 mutations in dried blood spots from newborns with elevated 17-OH progesterone. Clinica Chimica Acta, 2012, 414, 211-214.	1.1	17
26	The unsolved enigma of CDH1 down-regulation in hereditary diffuse gastric cancer. Journal of Surgical Research, 2004, 121, 50-55.	1.6	16
27	A novel MEN1 frameshift germline mutation in two Italian monozygotic twins. Clinical Chemistry and Laboratory Medicine, 2008, 46, 824-6.	2.3	16
28	Genes, pseudogenes and like genes: The case of 21-hydroxylase in Italian population. Clinica Chimica Acta, 2013, 424, 85-89.	1.1	16
29	Characterization of a new BRCA1 rearrangement in an Italian woman with hereditary breast and ovarian cancer syndrome. Breast Cancer Research and Treatment, 2017, 164, 497-503.	2.5	16
30	GSTT1 and GSTM1 allelic polymorphisms in head and neck cancer patients from Italian Lazio Region. Clinica Chimica Acta, 2007, 376, 174-178.	1.1	14
31	CYP21A2 intronic variants causing 21-hydroxylase deficiency. Metabolism: Clinical and Experimental, 2017, 71, 46-51.	3.4	13
32	Iodothyronine deiodinases and reduced sensitivity to thyroid hormones. Frontiers in Bioscience - Landmark, 2020, 25, 201-228.	3.0	13
33	Description of a novel missense mutation of glucose-6-phosphate dehydrogenase gene associated with asymptomatic high enzyme deficiency. Clinical Biochemistry, 2007, 40, 856-858.	1.9	12
34	Functional analysis of two rare CYP21A2 mutations detected in Italian patients with a mildest form of congenital adrenal hyperplasia. Clinical Endocrinology, 2009, 71, 470-476.	2.4	12
35	p.H282N and p.Y191H: 2 novel CYP21A2 mutations in Italian congenital adrenal hyperplasia patients. Metabolism: Clinical and Experimental, 2012, 61, 519-524.	3.4	12
36	Identification and Characterization of a New BRCA2 Rearrangement in an Italian Family with Hereditary Breast and Ovarian Cancer Syndrome. Molecular Diagnosis and Therapy, 2017, 21, 539-545.	3.8	12

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37	A Whole Germline BRCA2 Gene Deletion: How to Learn from CNV In Silico Analysis. International Journal of Molecular Sciences, 2018, 19, 961.	4.1	12
38	Spectrum of DICER1 Germline Pathogenic Variants in Ovarian Sertoliâ€“Leydig Cell Tumor. Journal of Clinical Medicine, 2021, 10, 1845.	2.4	12
39	DNA from buccal swab is suitable for rapid genotyping of angiotensin-converting enzyme insertion/deletion (I/D) polymorphism. Clinica Chimica Acta, 2014, 431, 125-130.	1.1	11
40	Linkage between I172N mutation, a marker of 21-hydroxylase deficiency, and a single nucleotide polymorphism in Int6 of CYP21B gene: A genetic study of Sardinian family. Clinica Chimica Acta, 2006, 364, 298-302.	1.1	10
41	Performance of multiplicom's BRCA MASTR Dx kit on the detection of BRCA1 and BRCA2 mutations in fresh frozen ovarian and breast tumor samples. Oncotarget, 2016, 7, 81357-81366.	1.8	10
42	Two novel CYP21A2 missense mutations in Italian patients with 21â€hydroxylase deficiency: Identification and functional characterisation. IUBMB Life, 2009, 61, 229-235.	3.4	9
43	BRCA1 and BRCA2 Testing through Next Generation Sequencing in a Small Cohort of Italian Breast/Ovarian Cancer Patients: Novel Pathogenic and Unknown Clinical Significance Variants. International Journal of Molecular Sciences, 2019, 20, 3442.	4.1	9
44	Insulin-like growth factor I (CA) repeats are associated with higher melanoma's Breslow index but not associated with the presence of the melanoma. A pilot study. Clinica Chimica Acta, 2008, 390, 104-109.	1.1	8
45	A new CYP21A2 nonsense mutation causing severe 21-hydroxylase deficiency. Clinical Chemistry and Laboratory Medicine, 2009, 47, 824-5.	2.3	8
46	A new standardized absolute quantitative RT-PCR method for detection of tyrosinase mRNAs in melanoma patients: Technical and operative instructions. Clinica Chimica Acta, 2009, 409, 100-105.	1.1	8
47	The First Case of Association Between Postpartum Thyroiditis and Thyroid Hormone Resistance in an Italian Patient Showing a Novel p.V283A THRB Mutation. Thyroid, 2013, 23, 506-510.	4.5	8
48	A prolonged neonatal jaundice associated with a rare G6PD mutation. Pediatric Blood and Cancer, 2009, 53, 475-478.	1.5	7
49	Rapid detection of CFH (p.Y402H) and ARMS2 (p.A69S) polymorphisms in age-related macular degeneration using high-resolution melting analysis. Clinical Chemistry and Laboratory Medicine, 2012, 50, 1031-4.	2.3	7
50	Is there a relationship between ELF free-IGF-1 levels and fibrotic process enhancement characterizing CLD development in neutropenic premature babies?. Pediatric Pulmonology, 2006, 41, 286-287.	2.0	5
51	Acute haemolytic crisis due to concomitant presence of infection and possible altered acetaminophen catabolism in a Philipino child carrying the G6PD-Vanua Lava mutation. Annals of Clinical Biochemistry, 2011, 48, 282-285.	1.6	5
52	Preliminary molecular evidence associating a novel BRCA1 synonymous variant with hereditary ovarian cancer syndrome. Human Genome Variation, 2018, 5, 2.	0.7	5
53	46,XY Disorder of Sex Development Caused by 17Î±-Hydroxylase/17,20-Lyase Deficiency due to Homozygous Mutation ofCYP17A1Gene: Consequences of Late Diagnosis. Case Reports in Endocrinology, 2018, 2018, 1-6.	0.4	5
54	Additional molecular and clinical evidence open the way to definitive IARC classification of the BRCA1 c.5332Gâ€›â€¯A (p.Asp1778Asn) variant. Clinical Biochemistry, 2019, 63, 54-58.	1.9	5

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55	Interaction between GSTM1 genotype and IL-6 on mortality in older adults: Results from the ilSIRENTE study. Cytokine, 2011, 53, 301-305.	3.2	4
56	Retinal function and CFH-ARMS2 polymorphisms analysis: a pilot study in Italian AMD patients. Neurobiology of Aging, 2012, 33, 1852.e5-1852.e12.	3.1	4
57	Prevalence of CAH-X Syndrome in Italian Patients with Congenital Adrenal Hyperplasia (CAH) Due to 21-Hydroxylase Deficiency. Journal of Clinical Medicine, 2022, 11, 3818.	2.4	4
58	CYP21A2 p.E238 Deletion as Result of Multiple Microconversion Events. Diagnostic Molecular Pathology, 2013, 22, 48-51.	2.1	3
59	CYP21A2 genetics: When genotype does not fit phenotype. Clinical Biochemistry, 2016, 49, 524-525.	1.9	3
60	A novel germline mutation at exon 10 of MEN1 gene: a clinical survey and positive genotype-phenotype analysis of a MEN1 Italian family, including monozygotic twins. Hormones, 2018, 17, 427-435.	1.9	3
61	Novel human pathological mutations. Gene Symbol: CYP21A2. Disease: Non-classic 21-hydroxylase deficiency. Human Genetics, 2007, 122, 559.	3.8	3
62	A case of patient affected by hirsutism carrying the P482S CYP21 gene mutation associated with loss of heterozygosity (LOH). Clinica Chimica Acta, 2006, 370, 201-202.	1.1	2
63	High Resolution Melting Analysis (HRMA) for the identification of a rare UGT1A1 promoter polymorphism. Clinical Biochemistry, 2011, 44, 1359-1360.	1.9	2
64	A Novel ATM Pathogenic Variant in an Italian Woman with Gallbladder Cancer. Genes, 2021, 12, 313.	2.4	2
65	Genetic cystic fibrosis transmembrane regulator 4016insT D1152H compound heterozygosity and male infertility: an Italian case report. Clinical Chemistry and Laboratory Medicine, 2007, 45, 923-4.	2.3	1
66	First case of V281+I172N/V281L CYP21A2 genotype associated with congenital adrenal hyperplasia form. A case report from South Italy. Clinical Biochemistry, 2007, 40, 1435-1436.	1.9	0
67	How the â€œAâ€•to â€œCâ€•conversion may create a new splice acceptor site?. Metabolism: Clinical and Experimental, 2010, 59, e11-e12.	3.4	0
68	A novel MEN1 pathogenic variant in an Italian patient with multiple endocrine neoplasia type 1. Molecular Biology Reports, 2020, 47, 7313-7316.	2.3	0