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	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
2	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
3	A Novel ATM Pathogenic Variant in an Italian Woman with Gallbladder Cancer. Genes, 2021, 12, 313.	2.4	2
4	Spectrum of DICER1 Germline Pathogenic Variants in Ovarian Sertoliâ€“Leydig Cell Tumor. Journal of Clinical Medicine, 2021, 10, 1845.	2.4	12
5	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
6	Iodothyronine deiodinases and reduced sensitivity to thyroid hormones. Frontiers in Bioscience - Landmark, 2020, 25, 201-228.	3.0	13
7	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
8	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
9	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
10	Preliminary molecular evidence associating a novel BRCA1 synonymous variant with hereditary ovarian cancer syndrome. Human Genome Variation, 2018, 5, 2.	0.7	5
11	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
12	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
13	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
14	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
15	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
16	CYP21A2 intronic variants causing 21-hydroxylase deficiency. Metabolism: Clinical and Experimental, 2017, 71, 46-51.	3.4	13
17	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
18	CYP21A2 genetics: When genotype does not fit phenotype. Clinical Biochemistry, 2016, 49, 524-525.	1.9	3

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19	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
20	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
21	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
22	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
23	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
24	Genes, pseudogenes and like genes: The case of 21-hydroxylase in Italian population. Clinica Chimica Acta, 2013, 424, 85-89.	1.1	16
25	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
26	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
27	CYP21A2 p.E238 Deletion as Result of Multiple Microconversion Events. Diagnostic Molecular Pathology, 2013, 22, 48-51.	2.1	3
28	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
29	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
30	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
31	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
32	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
33	High Resolution Melting Analysis (HRMA) for the identification of a rare UGT1A1 promoter polymorphism. Clinical Biochemistry, 2011, 44, 1359-1360.	1.9	2
34	Differentiated Thyroid Cancer in Two Patients with Resistance to Thyroid Hormone. Thyroid, 2011, 21, 793-797.	4.5	18
35	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
36	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

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37	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
38	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
39	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
40	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
41	A new CYP21A2 nonsense mutation causing severe 21-hydroxylase deficiency. Clinical Chemistry and Laboratory Medicine, 2009, 47, 824-5.	2.3	8
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	A prolonged neonatal jaundice associated with a rare G6PD mutation. Pediatric Blood and Cancer, 2009, 53, 475-478.	1.5	7
44	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
45	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
46	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
47	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
48	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
49	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
50	Identification of RFLP G6PD mutations by using microcapillary electrophoretic chips (Experion^{TM}). Journal of Separation Science, 2008, 31, 2694-2700.	2.5	26
51	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
52	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
53	A novel MEN1 frameshift germline mutation in two Italian monozygotic twins. Clinical Chemistry and Laboratory Medicine, 2008, 46, 824-6.	2.3	16
54	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

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55	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
56	Genetic analysis of the dystroglycan gene in bronchopulmonary dysplasia affected premature newborns. Clinica Chimica Acta, 2007, 378, 164-167.	1.1	17
57	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
58	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
59	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
60	Mannose-binding lectin polymorphisms and pulmonary outcome in premature neonates: aÂpilot study. Intensive Care Medicine, 2007, 33, 1787-1794.	8.2	26
61	Association of periodontitis with GSTM1/GSTT1-null variantsâ€"A pilot study. Clinical Biochemistry, 2007, 40, 939-945.	1.9	24
62	Novel human pathological mutations. Gene Symbol: CYP21A2. Disease: Non-classic 21-hydroxylase deficiency. Human Genetics, 2007, 122, 559.	3.8	3
63	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
64	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
65	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
66	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
67	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
68	The unsolved enigma of CDH1 down-regulation in hereditary diffuse gastric cancer. Journal of Surgical Research, 2004, 121, 50-55.	1.6	16