

Glenn E Palomaki

List of Publications by Year in descending order

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109
papers

7,672
citations

126708

33
h-index

51492

86
g-index

112
all docs

112
docs citations

112
times ranked

5373
citing authors

#	ARTICLE	IF	CITATIONS
1	First-trimester screening for pre-eclampsia: estimated vs measured mean arterial pressure. Ultrasound in Obstetrics and Gynecology, 2022, 59, 692-693.	0.9	0
2	Preeclampsia at delivery is associated with lower serum vitamin D and higher antiangiogenic factors: a case control study. Reproductive Biology and Endocrinology, 2022, 20, 8.	1.4	1
3	An Educational Assessment of Evidence Used for Variant Classification. Journal of Molecular Diagnostics, 2022, 24, 555-565.	1.2	3
4	International Society for Prenatal Diagnosis Position Statement: cell free (cf)DNA screening for Down syndrome in multiple pregnancies. Prenatal Diagnosis, 2021, 41, 1222-1232.	1.1	41
5	DNA-based screening and personal health: a points to consider statement for individuals and health-care providers from the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2021, 23, 979-988.	1.1	14
6	DNA-based screening and population health: a points to consider statement for programs and sponsoring organizations from the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2021, 23, 989-995.	1.1	43
7	Prenatal serum screening for Down syndrome and neural tube defects in the United States: Changes in utilization patterns from 2012 to 2020. Journal of Medical Screening, 2021, 28, 405-410.	1.1	4
8	Laboratory screening and diagnosis of open neural tube defects, 2019 revision: a technical standard of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2020, 22, 462-474.	1.1	23
9	Assessment of laboratories offering cell-free (cf) DNA screening for Down syndrome: results of the 2018 College of American Pathology External Educational Exercises. Genetics in Medicine, 2020, 22, 777-784.	1.1	5
10	Adjusting antimüllerian hormone levels for age and body mass index improves detection of polycystic ovary syndrome. Fertility and Sterility, 2020, 113, 876-884.e2.	0.5	7
11	Comment on "Expanded carrier screening for autosomal recessive conditions in health care: Arguments for a couple-based approach and examination of couples' views". Prenatal Diagnosis, 2019, 39, 1038-1038.	1.1	0
12	Maternal BMI, Peripheral Deiodinase Activity, and Plasma Glucose: Relationships Between White Women in the HAPO Study. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 2593-2600.	1.8	12
13	Levels of angiogenic markers in second-trimester maternal serum from in vitro fertilization pregnancies with oocyte donation. Fertility and Sterility, 2019, 112, 1112-1117.	0.5	1
14	Fewer women aged 35 and older choose serum screening for Down's syndrome: Impact and implications. Journal of Medical Screening, 2019, 26, 59-66.	1.1	1
15	Sequencing Cell-Free DNA in the Maternal Circulation to Screen for Down Syndrome, Other Common Trisomies, and Selected Genetic Disorders. , 2019, , 561-582.		3
16	Prenatal cell-free DNA screening test failures: a systematic review of failure rates, risks of Down syndrome, and impact of repeat testing. Genetics in Medicine, 2018, 20, 1312-1323.	1.1	40
17	CAP/ACMG proficiency testing for biochemical genetics laboratories: a summary of performance. Genetics in Medicine, 2018, 20, 83-90.	1.1	7
18	Relaxin-2 connecting peptide (pro-RLX2) levels in second trimester serum samples to predict preeclampsia. Pregnancy Hypertension, 2018, 11, 124-128.	0.6	6

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19	Snoring and markers of fetal and placental wellbeing. <i>Clinica Chimica Acta</i> , 2018, 485, 139-143.	0.5	9
20	The clinical utility of DNA-based screening for fetal aneuploidy by primary obstetrical care providers in the general pregnancy population. <i>Genetics in Medicine</i> , 2017, 19, 778-786.	1.1	36
21	Serum Progesterone Levels in Pregnant Women with Obstructive Sleep Apnea: A Case Control Study. <i>Journal of Women's Health</i> , 2017, 26, 259-265.	1.5	28
22	Nuchal translucency measurement in the era of prenatal screening for aneuploidy using cell free (cf)DNA. <i>Prenatal Diagnosis</i> , 2017, 37, 303-305.	1.1	7
23	Offering Prenatal Screening in the Age of Genomic Medicine: A Practical Guide. <i>Journal of Women's Health</i> , 2017, 26, 755-761.	1.5	9
24	Emerging Considerations for Noninvasive Prenatal Testing. <i>Clinical Chemistry</i> , 2017, 63, 946-953.	1.5	9
25	Measuring maternal serum screening markers for Down's syndrome in plasma collected for cell-free DNA testing. <i>Journal of Medical Screening</i> , 2017, 24, 113-119.	1.1	2
26	Where have all the trisomies gone?. <i>American Journal of Obstetrics and Gynecology</i> , 2016, 215, 583-587.e1.	0.7	13
27	Confusion between analytic validity and clinical validity. <i>American Journal of Obstetrics and Gynecology</i> , 2016, 215, 533-534.	0.7	1
28	Results from an external proficiency testing program: 11 years of molecular genetics testing for myotonic dystrophy type 1. <i>Genetics in Medicine</i> , 2016, 18, 1290-1294.	1.1	6
29	Evaluating first trimester maternal serum screening combinations for Down syndrome suitable for use with reflexive secondary screening via sequencing of cell free DNA: high detection with low rates of invasive procedures. <i>Prenatal Diagnosis</i> , 2015, 35, 789-796.	1.1	19
30	Prenatal serum screening markers may not require adjustment in former smokers. <i>Prenatal Diagnosis</i> , 2015, 35, 1371-1373.	1.1	0
31	A flawed challenge but valid recommendation: a response to Takoudes and Hamar. <i>Ultrasound in Obstetrics and Gynecology</i> , 2015, 45, 117-117.	0.9	2
32	Modeling risk for severe adverse outcomes using angiogenic factor measurements in women with suspected preterm preeclampsia. <i>Prenatal Diagnosis</i> , 2015, 35, 386-393.	1.1	28
33	Circulating cell free DNA testing: are some test failures informative?. <i>Prenatal Diagnosis</i> , 2015, 35, 289-293.	1.1	79
34	Evaluation of Patient Education Materials: The Example of Circulating cell free DNA Testing for Aneuploidy. <i>Journal of Genetic Counseling</i> , 2015, 24, 259-266.	0.9	24
35	Is maternal plasma DNA testing impacting serum-based screening for aneuploidy in the United States?. <i>Genetics in Medicine</i> , 2015, 17, 897-900.	1.1	4
36	Screening for breast cancer by molecular testing for three founder mutations in the BRCA1 and BRCA2 genes among women of Ashkenazi Jewish heritage. <i>Journal of Medical Screening</i> , 2015, 22, 109-111.	1.1	1

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37	Is it time for BRCA1/2 mutation screening in the general adult population?: impact of population characteristics. <i>Genetics in Medicine</i> , 2015, 17, 24-26.	1.1	16
38	Molecular genetic testing for cystic fibrosis: laboratory performance on the College of American Pathologists external proficiency surveys. <i>Genetics in Medicine</i> , 2015, 17, 219-225.	1.1	13
39	Molecular testing for the BRCA1 and BRCA2 Ashkenazi Jewish founder mutations: a report on the College of American Pathologists proficiency testing surveys. <i>Genetics in Medicine</i> , 2015, 17, 58-62.	1.1	16
40	Sequencing Cell Free DNA in the Maternal Circulation to Screen for Down Syndrome and Other Common Aneuploidies. , 2015, , 563-580.		0
41	Results of the College of American Pathology/American College of Medical Genetics and Genomics external proficiency testing from 2006 to 2013 for three conditions prevalent in the Ashkenazi Jewish population. <i>Genetics in Medicine</i> , 2014, 16, 695-702.	1.1	15
42	Three-year experience of a CAP/ACMG methods-based external proficiency testing program for laboratories offering DNA sequencing for rare inherited disorders. <i>Genetics in Medicine</i> , 2014, 16, 25-32.	1.1	21
43	Maternal plasma DNA testing for aneuploidy in pregnancies achieved by assisted reproductive technologies. <i>Genetics in Medicine</i> , 2014, 16, 419-422.	1.1	12
44	Down syndrome screening: Suitability of a WHO 5 standardized total hCG assay. <i>Clinical Biochemistry</i> , 2014, 47, 629-631.	0.8	9
45	Maternal Plasma DNA Testing: Experience of Women Counseled at a Prenatal Diagnosis Center. <i>Genetic Testing and Molecular Biomarkers</i> , 2014, 18, 665-669.	0.3	2
46	Use of first or second trimester serum markers, or both, to predict preeclampsia. <i>Pregnancy Hypertension</i> , 2014, 4, 271-278.	0.6	14
47	Noninvasive prenatal detection of sex chromosomal aneuploidies by sequencing circulating cell-free DNA from maternal plasma. <i>Prenatal Diagnosis</i> , 2013, 33, 591-597.	1.1	173
48	Use of genomic panels to determine risk of developing type 2 diabetes in the general population: a targeted evidence-based review. <i>Genetics in Medicine</i> , 2013, 15, 600-611.	1.1	12
49	The impact of maternal plasma DNA fetal fraction on next generation sequencing tests for common fetal aneuploidies. <i>Prenatal Diagnosis</i> , 2013, 33, 667-674.	1.1	310
50	Screening for Down Syndrome in the United States: Results of Surveys in 2011 and 2012. <i>Archives of Pathology and Laboratory Medicine</i> , 2013, 137, 921-926.	1.2	30
51	High-Throughput Massively Parallel Sequencing for Fetal Aneuploidy Detection from Maternal Plasma. <i>PLoS ONE</i> , 2013, 8, e57381.	1.1	86
52	Feasibility of Using Plasma Rather Than Serum in First and Second Trimester Multiple Marker Down's Syndrome Screening. <i>Journal of Medical Screening</i> , 2012, 19, 164-170.	1.1	1
53	Assessing the analytic validity of molecular testing for Huntington disease using data from an external proficiency testing survey. <i>Genetics in Medicine</i> , 2012, 14, 69-75.	1.1	19
54	Maternal plasma DNA: A major step forward in prenatal testing. <i>Journal of Medical Screening</i> , 2012, 19, 57-59.	1.1	63

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55	DNA sequencing of maternal plasma reliably identifies trisomy 18 and trisomy 13 as well as Down syndrome: an international collaborative study. <i>Genetics in Medicine</i> , 2012, 14, 296-305.	1.1	471
56	DNA sequencing of maternal plasma to identify Down syndrome and other trisomies in multiple gestations. <i>Prenatal Diagnosis</i> , 2012, 32, 730-734.	1.1	153
57	DNA sequencing of maternal plasma to detect Down syndrome: An international clinical validation study. <i>Genetics in Medicine</i> , 2011, 13, 913-920.	1.1	809
58	Impact of smoking on maternal serum markers and prenatal screening in the first and second trimesters. <i>Prenatal Diagnosis</i> , 2011, 31, 583-588.	1.1	24
59	Noninvasive Fetal Sex Determination Using Cell-Free Fetal DNA. <i>JAMA - Journal of the American Medical Association</i> , 2011, 306, 627-36.	3.8	213
60	Screening to detect Lynch syndrome and prevent hereditary cancers in relatives. <i>Journal of Medical Screening</i> , 2011, 18, 167-168.	1.1	2
61	An Introduction to Assessing Genomic Screening and Diagnostic Tests. <i>Nutrition Today</i> , 2011, 46, 162-168.	0.6	6
62	Use of genomic profiling to assess risk for cardiovascular disease and identify individualized prevention strategies—A targeted evidence-based review. <i>Genetics in Medicine</i> , 2010, 12, 772-784.	1.1	32
63	Examination of the pregnancy-associated plasma protein-A assay on the Beckman Coulter AccessA [®] platform: suitability for use in first trimester Down's syndrome screening. <i>Journal of Medical Screening</i> , 2010, 17, 109-113.	1.1	5
64	Association Between 9p21 Genomic Markers and Heart Disease. <i>JAMA - Journal of the American Medical Association</i> , 2010, 303, 648.	3.8	141
65	Four Years' Experience With an Interlaboratory Comparison Program Involving First-Trimester Markers of Down Syndrome. <i>Archives of Pathology and Laboratory Medicine</i> , 2010, 134, 1685-1691.	1.2	6
66	Can UGT1A1 genotyping reduce morbidity and mortality in patients with metastatic colorectal cancer treated with irinotecan? An evidence-based review. <i>Genetics in Medicine</i> , 2009, 11, 21-34.	1.1	135
67	EGAPP supplementary evidence review: DNA testing strategies aimed at reducing morbidity and mortality from Lynch syndrome. <i>Genetics in Medicine</i> , 2009, 11, 42-65.	1.1	431
68	Identifying Lynch syndrome. <i>International Journal of Cancer</i> , 2009, 125, 1492-1493.	2.3	32
69	Early onset preeclampsia and second trimester serum markers. <i>Prenatal Diagnosis</i> , 2009, 29, 1109-1117.	1.1	14
70	Technical standards and guidelines: Prenatal screening for Down syndrome that includes first-trimester biochemistry and/or ultrasound measurements. <i>Genetics in Medicine</i> , 2009, 11, 669-681.	1.1	42
71	The Evaluation of Genomic Applications in Practice and Prevention (EGAPP) initiative: methods of the EGAPP Working Group. <i>Genetics in Medicine</i> , 2009, 11, 3-14.	1.1	584
72	Quality assessment of routine nuchal translucency measurements: a North American laboratory perspective. <i>Genetics in Medicine</i> , 2008, 10, 131-138.	1.1	47

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73	A Summary Analysis of Down Syndrome Markers in the Late First Trimester. <i>Advances in Clinical Chemistry</i> , 2007, 43, 177-210.	1.8	30
74	Estimating first-trimester combined screening performance for Down syndrome in dried blood spots versus fresh sera. <i>Genetics in Medicine</i> , 2007, 9, 458-463.	1.1	6
75	Hyperglycosylated-hCG (h-hCG) and Down syndrome screening in the first and second trimesters of pregnancy. <i>Prenatal Diagnosis</i> , 2007, 27, 808-813.	1.1	16
76	A summary analysis of Down syndrome markers in the late first trimester. <i>Advances in Clinical Chemistry</i> , 2007, 43, 177-210.	1.8	5
77	Comparing Three Screening Strategies for Combining First- and Second-Trimester Down Syndrome Markers. <i>Obstetrics and Gynecology</i> , 2006, 107, 367-375.	1.2	48
78	Comparison of Serum Markers in First-Trimester Down Syndrome Screening. <i>Obstetrics and Gynecology</i> , 2006, 108, 1192-1199.	1.2	44
79	Stability of first- and second-trimester serum markers after storage and shipment. <i>Prenatal Diagnosis</i> , 2006, 26, 17-21.	1.1	21
80	Repeated measurement of pregnancy-associated plasma protein-A (PAPP-A) in Down syndrome screening: A validation study. <i>Prenatal Diagnosis</i> , 2006, 26, 730-739.	1.1	23
81	First-Trimester Down Syndrome Screening: Reply. <i>Clinical Chemistry</i> , 2006, 52, 161-161.	1.5	1
82	Adjusting the estimated proportion of breast cancer cases associated with BRCA1 and BRCA2 mutations: Public health implications. <i>Genetics in Medicine</i> , 2005, 7, 28-33.	1.1	70
83	An evaluation of BRCA1 and BRCA2 founder mutations penetrance estimates for breast cancer among Ashkenazi Jewish women. <i>Genetics in Medicine</i> , 2005, 7, 34-39.	1.1	17
84	Patient and Health Professional Acceptance of Integrated Serum Screening for Down Syndrome. <i>Seminars in Perinatology</i> , 2005, 29, 247-251.	1.1	9
85	Integrated serum screening for Down syndrome in primary obstetric practice. <i>Prenatal Diagnosis</i> , 2005, 25, 1162-1167.	1.1	29
86	Maternal Serum Invasive Trophoblast Antigen and First-Trimester Down Syndrome Screening. <i>Clinical Chemistry</i> , 2005, 51, 1499-1504.	1.5	23
87	Technical standards and guidelines: Prenatal screening for Down syndrome: This new section on "Prenatal Screening for Down Syndrome," together with the new section on "Prenatal Screening for Open Neural Tube Defects," replaces the previous Section H of the American College of Medical Genetics Standards and Guidelines for Clinical Genetics Laboratories*. <i>Genetics in Medicine</i> , 2005, 7, 344-354.	1.1	16
88	Second-Trimester Maternal Serum Invasive Trophoblast Antigen: A Marker for Down Syndrome Screening. <i>Clinical Chemistry</i> , 2004, 50, 1433-1435.	1.5	18
89	Maternal Serum Invasive Trophoblast Antigen (Hyperglycosylated hCG) as a Screening Marker for Down Syndrome during the Second Trimester. <i>Clinical Chemistry</i> , 2004, 50, 1804-1808.	1.5	25
90	Epidemiologic monitoring of prenatal screening for neural tube defects and Down syndrome. <i>Clinics in Laboratory Medicine</i> , 2003, 23, 531-551.	0.7	25

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91	Analytic validity of cystic fibrosis testing: A preliminary estimate. <i>Genetics in Medicine</i> , 2003, 5, 15-20.	1.1	25
92	Estimated analytic validity of HFE C282Y mutation testing in population screening: The potential value of confirmatory testing. <i>Genetics in Medicine</i> , 2003, 5, 440-443.	1.1	21
93	hCG and the free β -subunit as screening tests for Down syndrome. <i>Prenatal Diagnosis</i> , 1998, 18, 235-245.	1.1	35
94	Screening of Maternal Serum for Fetal Down's Syndrome in the First Trimester. <i>New England Journal of Medicine</i> , 1998, 338, 955-962.	13.9	242
95	Second trimester screening for Down's syndrome using maternal serum dimeric inhibin A. <i>Journal of Medical Screening</i> , 1998, 5, 115-119.	1.1	65
96	Maternal serum screening for Down syndrome in the United States: A 1995 survey. <i>American Journal of Obstetrics and Gynecology</i> , 1997, 176, 1046-1051.	0.7	111
97	COUPLE-BASED PRENATAL SCREENING FOR CYSTIC FIBROSIS IN PRIMARY CARE SETTINGS. , 1996, 16, 397-404.		33
98	REFINEMENTS IN MANAGING MATERNAL WEIGHT ADJUSTMENT FOR INTERPRETING PRENATAL SCREENING RESULTS. , 1996, 16, 1115-1119.		124
99	Reducing the Need for Amniocentesis in Women 35 Years of Age or Older with Serum Markers for Screening. <i>New England Journal of Medicine</i> , 1994, 330, 1114-1118.	13.9	209
100	Pregnancy associated plasma protein A as a marker for Down syndrome in the second trimester of pregnancy. <i>Prenatal Diagnosis</i> , 1993, 13, 222-223.	1.1	27
101	Biparietal diameter and crown-rump length in fetuses with Down's syndrome: implications for antenatal serum screening for Down's syndrome. <i>BJOG: an International Journal of Obstetrics and Gynaecology</i> , 1993, 100, 430-435.	1.1	27
102	Maternal serum screening for fetal down syndrome in the United States: A 1992 survey. <i>American Journal of Obstetrics and Gynecology</i> , 1993, 169, 1558-1562.	0.7	45
103	Cigarette smoking and levels of maternal serum alpha-fetoprotein, unconjugated estriol, and hCG: impact on Down syndrome screening. <i>Obstetrics and Gynecology</i> , 1993, 81, 675-8.	1.2	45
104	Prenatal Screening for Down's Syndrome with Use of Maternal Serum Markers. <i>New England Journal of Medicine</i> , 1992, 327, 588-593.	13.9	450
105	The effect of smoking in pregnancy on maternal serum alpha-fetoprotein, unconjugated oestriol, human chorionic gonadotrophin, progesterone and dehydroepiandrosterone sulphate levels. <i>BJOG: an International Journal of Obstetrics and Gynaecology</i> , 1990, 97, 272-274.	1.1	56
106	Clinical Articles Maternal serum β -fetoprotein screening for fetal Down syndrome in the United States: Results of a survey. <i>American Journal of Obstetrics and Gynecology</i> , 1990, 162, 317-321.	0.7	19
107	Low second trimester maternal serum unconjugated oestriol in pregnancies with Down's syndrome. <i>BJOG: an International Journal of Obstetrics and Gynaecology</i> , 1988, 95, 330-333.	1.1	276
108	Maternal serum screening for Down's syndrome in early pregnancy.. <i>BMJ: British Medical Journal</i> , 1988, 297, 883-887.	2.4	866

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109	Maternal serum α -fetoprotein, age, and Down syndrome risk. American Journal of Obstetrics and Gynecology, 1987, 156, 460-463.	0.7	120