Glenn E Palomaki

List of Publications by Year
in descending order

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	Maternal serum screening for Down's syndrome in early pregnancy.. BMJ: British Medical Journal, 1988, 297, 883-887.	2.4	866
2	DNA sequencing of maternal plasma to detect Down syndrome: An international clinical validation study. Genetics in Medicine, 2011, 13, 913-920.	1.1	809
3	The Evaluation of Genomic Applications in Practice and Prevention (EGAPP) initiative: methods of the EGAPP Working Group. Genetics in Medicine, 2009, 11, 3-14.	1.1	584
4	DNA sequencing of maternal plasma reliably identifies trisomy 18 and trisomy 13 as well as Down syndrome: an international collaborative study. Genetics in Medicine, 2012, 14, 296-305.	1.1	471
5	Prenatal Screening for Down's Syndrome with Use of Maternal Serum Markers. New England Journal of Medicine, 1992, 327, 588-593.	13.9	450
6	EGAPP supplementary evidence review: DNA testing strategies aimed at reducing morbidity and mortality from Lynch syndrome. Genetics in Medicine, 2009, 11, 42-65.	1.1	431
7	The impact of maternal plasma DNA fetal fraction on next generation sequencing tests for common fetal aneuploidies. Prenatal Diagnosis, 2013, 33, 667-674.	1.1	310
8	Low second trimester maternal serum unconjugated oestriol in pregnancies with Down's syndrome. BJOG: an International Journal of Obstetrics and Gynaecology, 1988, 95, 330-333.	1.1	276
9	Screening of Maternal Serum for Fetal Down's Syndrome in the First Trimester. New England Journal of Medicine, 1998, 338, 955-962.	13.9	242
10	Noninvasive Fetal Sex Determination Using Cell-Free Fetal DNA. JAMA - Journal of the American Medical Association, 2011, 306, 627-36.	3.8	213
11	Reducing the Need for Amniocentesis in Women 35 Years of Age or Older with Serum Markers for Screening. New England Journal of Medicine, 1994, 330, 1114-1118.	13.9	209
12	Noninvasive prenatal detection of sex chromosomal aneuploidies by sequencing circulating cellâ€free DNA from maternal plasma. Prenatal Diagnosis, 2013, 33, 591-597.	1.1	173
13	DNA sequencing of maternal plasma to identify Down syndrome and other trisomies in multiple gestations. Prenatal Diagnosis, 2012, 32, 730-734.	1.1	153
14	Association Between 9p21 Genomic Markers and Heart Disease. JAMA - Journal of the American Medical Association, 2010, 303, 648.	3.8	141
15	Can UGT1A1 genotyping reduce morbidity and mortality in patients with metastatic colorectal cancer treated with irinotecan? An evidence-based review. Genetics in Medicine, 2009, 11, 21-34.	1.1	135
16	REFINEMENTS IN MANAGING MATERNAL WEIGHT ADJUSTMENT FOR INTERPRETING PRENATAL SCREENING RESULTS. , 1996, 16, 1115-1119.		124
17	Maternal serum Î±-fetoprotein, age, and Down syndrome risk. American Journal of Obstetrics and Gynecology, 1987, 156, 460-463.	0.7	120
18	Maternal serum screening for Down syndrome in the United States: A 1995 survey. American Journal of Obstetrics and Gynecology, 1997, 176, 1046-1051.	0.7	111

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19	High-Throughput Massively Parallel Sequencing for Fetal Aneuploidy Detection from Maternal Plasma. PLoS ONE, 2013, 8, e57381.	1.1	86
20	Circulating cell free DNA testing: are some test failures informative?. Prenatal Diagnosis, 2015, 35, 289-293.	1.1	79
21	Adjusting the estimated proportion of breast cancer cases associated with BRCA1 and BRCA2 mutations: Public health implications. Genetics in Medicine, 2005, 7, 28-33.	1.1	70
22	Second trimester screening for Down's syndrome using maternal serum dimeric inhibin A. Journal of Medical Screening, 1998, 5, 115-119.	1.1	65
23	Maternal plasma DNA: A major step forward in prenatal testing. Journal of Medical Screening, 2012, 19, 57-59.	1.1	63
24	The effect of smoking in pregnancy on maternal serum alpha-fetoprotein, unconjugated oestriol, human chorionic gonadotrophin, progesterone and dehydroepiandrosterone sulphate levels. BJOG: an International Journal of Obstetrics and Gynaecology, 1990, 97, 272-274.	1.1	56
25	Comparing Three Screening Strategies for Combining First- and Second-Trimester Down Syndrome Markers. Obstetrics and Gynecology, 2006, 107, 367-375.	1.2	48
26	Quality assessment of routine nuchal translucency measurements: a North American laboratory perspective. Genetics in Medicine, 2008, 10, 131-138.	1.1	47
27	Maternal serum screening for fetal down syndrome in the United States: A 1992 survey. American Journal of Obstetrics and Gynecology, 1993, 169, 1558-1562.	0.7	45
28	Cigarette smoking and levels of maternal serum alpha-fetoprotein, unconjugated estriol, and hCG: impact on Down syndrome screening. Obstetrics and Gynecology, 1993, 81, 675-8.	1.2	45
29	Comparison of Serum Markers in First-Trimester Down Syndrome Screening. Obstetrics and Gynecology, 2006, 108, 1192-1199.	1.2	44
30	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
31	Technical standards and guidelines: Prenatal screening for Down syndrome that includes first-trimester biochemistry and/or ultrasound measurements. Genetics in Medicine, 2009, 11, 669-681.	1.1	42
32	International Society for Prenatal Diagnosis Position Statement: cell free (cf)<scp>DNA</scp> screening for Down syndrome in multiple pregnancies. Prenatal Diagnosis, 2021, 41, 1222-1232.	1.1	41
33	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
34	The clinical utility of DNA-based screening for fetal aneuploidy by primary obstetrical care providers in the general pregnancy population. Genetics in Medicine, 2017, 19, 778-786.	1.1	36
35	hCG and the free Î²-subunit as screening tests for Down syndrome. Prenatal Diagnosis, 1998, 18, 235-245.	1.1	35
36	COUPLE-BASED PRENATAL SCREENING FOR CYSTIC FIBROSIS IN PRIMARY CARE SETTINGS. , 1996, 16, 397-404.		33

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37	Identifying Lynch syndrome. International Journal of Cancer, 2009, 125, 1492-1493.	2.3	32
38	Use of genomic profiling to assess risk for cardiovascular disease and identify individualized prevention strategiesâ€"A targeted evidence-based review. Genetics in Medicine, 2010, 12, 772-784.	1.1	32
39	A Summary Analysis of Down Syndrome Markers in the Late First Trimester. Advances in Clinical Chemistry, 2007, 43, 177-210.	1.8	30
40	Screening for Down Syndrome in the United States: Results of Surveys in 2011 and 2012. Archives of Pathology and Laboratory Medicine, 2013, 137, 921-926.	1.2	30
41	Integrated serum screening for Down syndrome in primary obstetric practice. Prenatal Diagnosis, 2005, 25, 1162-1167.	1.1	29
42	Modeling risk for severe adverse outcomes using angiogenic factor measurements in women with suspected preterm preeclampsia. Prenatal Diagnosis, 2015, 35, 386-393.	1.1	28
43	Serum Progesterone Levels in Pregnant Women with Obstructive Sleep Apnea: A Case Control Study. Journal of Women's Health, 2017, 26, 259-265.	1.5	28
44	Pregnancy associated plasma protein A as a marker for Down syndrome in the second trimester of pregnancy. Prenatal Diagnosis, 1993, 13, 222-223.	1.1	27
45	Biparietal diameter and crown-rump length in fetuses with Down's syndrome: implications for antenatal serum screening for Down's syndrome. BJOG: an International Journal of Obstetrics and Gynaecology, 1993, 100, 430-435.	1.1	27
46	Epidemiologic monitoring of prenatal screening for neural tube defects and Down syndrome. Clinics in Laboratory Medicine, 2003, 23, 531-551.	0.7	25
47	Analytic validity of cystic fibrosis testing: A preliminary estimate. Genetics in Medicine, 2003, 5, 15-20.	1.1	25
48	Maternal Serum Invasive Trophoblast Antigen (Hyperglycosylated hCG) as a Screening Marker for Down Syndrome during the Second Trimester. Clinical Chemistry, 2004, 50, 1804-1808.	1.5	25
49	Impact of smoking on maternal serum markers and prenatal screening in the first and second trimesters. Prenatal Diagnosis, 2011, 31, 583-588.	1.1	24
50	Evaluation of Patient Education Materials: The Example of Circulating cell free DNA Testing for Aneuploidy. Journal of Genetic Counseling, 2015, 24, 259-266.	0.9	24
51	Maternal Serum Invasive Trophoblast Antigen and First-Trimester Down Syndrome Screening. Clinical Chemistry, 2005, 51, 1499-1504.	1.5	23
52	Repeated measurement of pregnancy-associated plasma protein-A (PAPP-A) in Down syndrome screening: A validation study. Prenatal Diagnosis, 2006, 26, 730-739.	1.1	23
53	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
54	Estimated analytic validity of HFE C282Y mutation testing in population screening: The potential value of confirmatory testing. Genetics in Medicine, 2003, 5, 440-443.	1.1	21

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55	Stability of first- and second-trimester serum markers after storage and shipment. Prenatal Diagnosis, 2006, 26, 17-21.	1.1	21
56	Three-year experience of a CAP/ACMG methods-based external proficiency testing program for laboratories offering DNA sequencing for rare inherited disorders. Genetics in Medicine, 2014, 16, 25-32.	1.1	21
57	Clinical Articles Maternal serum Î±-fetoprotein screening for fetal Down syndrome in the United States: Results of a survey. American Journal of Obstetrics and Gynecology, 1990, 162, 317-321.	0.7	19
58	Assessing the analytic validity of molecular testing for Huntington disease using data from an external proficiency testing survey. Genetics in Medicine, 2012, 14, 69-75.	1.1	19
59	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. Prenatal Diagnosis, 2015, 35, 789-796.	1.1	19
60	Second-Trimester Maternal Serum Invasive Trophoblast Antigen: A Marker for Down Syndrome Screening. Clinical Chemistry, 2004, 50, 1433-1435.	1.5	18
61	An evaluation of BRCA1 and BRCA2 founder mutations penetrance estimates for breast cancer among Ashkenazi Jewish women. Genetics in Medicine, 2005, 7, 34-39.	1.1	17
62	Technical standards and guidelines: Prenatal screening for Down syndrome: This new section on â€œPrenatal Screening for Down Syndrome,â€•together with the new section on â€œPrenatal Screening for Open Neural Tube Defects,â€•replaces the previous Section H of the American College of Medical Genetics Standards and Guidelines for Clinical Genetics Laboratories*. Genetics in Medicine, 2005, 7, 344-354.	1.1	16
63	Hyperglycosylated-hCG (h-hCG) and Down syndrome screening in the first and second trimesters of pregnancy. Prenatal Diagnosis, 2007, 27, 808-813.	1.1	16
64	Is it time for BRCA1/2 mutation screening in the general adult population?: impact of population characteristics. Genetics in Medicine, 2015, 17, 24-26.	1.1	16
65	Molecular testing for the BRCA1 and BRCA2 Ashkenazi Jewish founder mutations: a report on the College of American Pathologists proficiency testing surveys. Genetics in Medicine, 2015, 17, 58-62.	1.1	16
66	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. Genetics in Medicine, 2014, 16, 695-702.	1.1	15
67	Early onset preeclampsia and second trimester serum markers. Prenatal Diagnosis, 2009, 29, 1109-1117.	1.1	14
68	Use of first or second trimester serum markers, or both, to predict preeclampsia. Pregnancy Hypertension, 2014, 4, 271-278.	0.6	14
69	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
70	Molecular genetic testing for cystic fibrosis: laboratory performance on the College of American Pathologists external proficiency surveys. Genetics in Medicine, 2015, 17, 219-225.	1.1	13
71	Where have all the trisomies gone?. American Journal of Obstetrics and Gynecology, 2016, 215, 583-587.e1.	0.7	13
72	Use of genomic panels to determine risk of developing type 2 diabetes in the general population: a targeted evidence-based review. Genetics in Medicine, 2013, 15, 600-611.	1.1	12

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73	Maternal plasma DNA testing for aneuploidy in pregnancies achieved by assisted reproductive technologies. Genetics in Medicine, 2014, 16, 419-422.	1.1	12
74	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
75	Patient and Health Professional Acceptance of Integrated Serum Screening for Down Syndrome. Seminars in Perinatology, 2005, 29, 247-251.	1.1	9
76	Down syndrome screening: Suitability of a WHO 5 standardized total hCG assay. Clinical Biochemistry, 2014, 47, 629-631.	0.8	9
77	Offering Prenatal Screening in the Age of Genomic Medicine: A Practical Guide. Journal of Women's Health, 2017, 26, 755-761.	1.5	9
78	Emerging Considerations for Noninvasive Prenatal Testing. Clinical Chemistry, 2017, 63, 946-953.	1.5	9
79	Snoring and markers of fetal and placental wellbeing. Clinica Chimica Acta, 2018, 485, 139-143.	0.5	9
80	Nuchal translucency measurement in the era of prenatal screening for aneuploidy using cell free (cf)DNA. Prenatal Diagnosis, 2017, 37, 303-305.	1.1	7
81	CAP/ACMG proficiency testing for biochemical genetics laboratories: a summary of performance. Genetics in Medicine, 2018, 20, 83-90.	1.1	7
82	Adjusting antimÃ¼llerian 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
83	Estimating first-trimester combined screening performance for Down syndrome in dried blood spots versus fresh sera. Genetics in Medicine, 2007, 9, 458-463.	1.1	6
84	An Introduction to Assessing Genomic Screening and Diagnostic Tests. Nutrition Today, 2011, 46, 162-168.	0.6	6
85	Results from an external proficiency testing program: 11 years of molecular genetics testing for myotonic dystrophy type 1. Genetics in Medicine, 2016, 18, 1290-1294.	1.1	6
86	Relaxin-2 connecting peptide (pro-RLX2) levels in second trimester serum samples to predict preeclampsia. Pregnancy Hypertension, 2018, 11, 124-128.	0.6	6
87	Four Years' Experience With an Interlaboratory Comparison Program Involving First-Trimester Markers of Down Syndrome. Archives of Pathology and Laboratory Medicine, 2010, 134, 1685-1691.	1.2	6
88	Examination of the pregnancy-associated plasma protein-A assay on the Beckman Coulter AccessÂ® platform: suitability for use in first trimester Down's syndrome screening. Journal of Medical Screening, 2010, 17, 109-113.	1.1	5
89	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
90	A summary analysis of Down syndrome markers in the late first trimester. Advances in Clinical Chemistry, 2007, 43, 177-210.	1.8	5

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91	Is maternal plasma DNA testing impacting serum-based screening for aneuploidy in the United States?. Genetics in Medicine, 2015, 17, 897-900.	1.1	4
92	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
93	Sequencing Cell-Free DNA in theÂMaternal Circulation to Screen for Down Syndrome, Other Common Trisomies, and Selected Genetic Disorders. , 2019, , 561-582.		3
94	An Educational Assessment of Evidence Used for Variant Classification. Journal of Molecular Diagnostics, 2022, 24, 555-565.	1.2	3
95	Screening to detect Lynch syndrome and prevent hereditary cancers in relatives. Journal of Medical Screening, 2011, 18, 167-168.	1.1	2
96	Maternal Plasma DNA Testing: Experience of Women Counseled at a Prenatal Diagnosis Center. Genetic Testing and Molecular Biomarkers, 2014, 18, 665-669.	0.3	2
97	A flawed challenge but valid recommendation: a response to Takoudes and Hamar. Ultrasound in Obstetrics and Gynecology, 2015, 45, 117-117.	0.9	2
98	Measuring maternal serum screening markers for Downâ€™s syndrome in plasma collected for cell-free DNA testing. Journal of Medical Screening, 2017, 24, 113-119.	1.1	2
99	First-Trimester Down Syndrome Screening: Reply. Clinical Chemistry, 2006, 52, 161-161.	1.5	1
100	Feasibility of Using Plasma Rather Than Serum in First and Second Trimester Multiple Marker Down's Syndrome Screening. Journal of Medical Screening, 2012, 19, 164-170.	1.1	1
101	Screening for breast cancer by molecular testing for three founder mutations in the BRCA1 and BRCA2 genes among women of Ashkenazi Jewish heritage. Journal of Medical Screening, 2015, 22, 109-111.	1.1	1
102	Confusion between analytic validity and clinicalÂvalidity. American Journal of Obstetrics and Gynecology, 2016, 215, 533-534.	0.7	1
103	Levels of angiogenic markers in second-trimester maternal serum from inÂvitro fertilization pregnancies with oocyte donation. Fertility and Sterility, 2019, 112, 1112-1117.	0.5	1
104	Fewer women aged 35 and older choose serum screening for Downâ€™s syndrome: Impact and implications. Journal of Medical Screening, 2019, 26, 59-66.	1.1	1
105	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
106	Prenatal serum screening markers may not require adjustment in former smokers. Prenatal Diagnosis, 2015, 35, 1371-1373.	1.1	0
107	Comment on â€œExpanded carrier screening for autosomal recessive conditions in health care: Arguments for a coupleâ€based approach and examination of couples' viewsâ€. Prenatal Diagnosis, 2019, 39, 1038-1038.	1.1	0
108	Sequencing Cell Free DNA in the Maternal Circulation to Screen for Down Syndrome and Other Common Aneuploidies. , 2015, , 563-580.		0

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109	Firstâ€trimester screening for preâ€eclampsia: estimated vs measured mean arterial pressure. Ultrasound in Obstetrics and Gynecology, 2022, 59, 692-693.	0.9	0