

# Rossa W K Chiu

## List of Publications by Year in descending order

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97  
papers

11,913  
citations

50170

46  
h-index

40881

93  
g-index

100  
all docs

100  
docs citations

100  
times ranked

9035  
citing authors

#	ARTICLE	IF	CITATIONS
1	Maternal Plasma DNA Sequencing Reveals the Genome-Wide Genetic and Mutational Profile of the Fetus. <i>Science Translational Medicine</i> , 2010, 2, 61ra91.	5.8	878
2	Noninvasive prenatal diagnosis of fetal chromosomal aneuploidy by massively parallel genomic sequencing of DNA in maternal plasma. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2008, 105, 20458-20463.	3.3	809
3	Non-invasive prenatal assessment of trisomy 21 by multiplexed maternal plasma DNA sequencing: large scale validity study. <i>BMJ: British Medical Journal</i> , 2011, 342, c7401-c7401.	2.4	641
4	Plasma DNA tissue mapping by genome-wide methylation sequencing for noninvasive prenatal, cancer, and transplantation assessments. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, E5503-12.	3.3	579
5	Lengthening and shortening of plasma DNA in hepatocellular carcinoma patients. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, E1317-25.	3.3	543
6	Analysis of Plasma Epstein-Barr Virus DNA to Screen for Nasopharyngeal Cancer. <i>New England Journal of Medicine</i> , 2017, 377, 513-522.	13.9	531
7	Predominant Hematopoietic Origin of Cell-free DNA in Plasma and Serum after Sex-mismatched Bone Marrow Transplantation. <i>Clinical Chemistry</i> , 2002, 48, 421-427.	1.5	483
8	Cancer Genome Scanning in Plasma: Detection of Tumor-Associated Copy Number Aberrations, Single-Nucleotide Variants, and Tumoral Heterogeneity by Massively Parallel Sequencing. <i>Clinical Chemistry</i> , 2013, 59, 211-224.	1.5	447
9	Microfluidics Digital PCR Reveals a Higher than Expected Fraction of Fetal DNA in Maternal Plasma. <i>Clinical Chemistry</i> , 2008, 54, 1664-1672.	1.5	396
10	mRNA of placental origin is readily detectable in maternal plasma. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2003, 100, 4748-4753.	3.3	363
11	Noninvasive detection of cancer-associated genome-wide hypomethylation and copy number aberrations by plasma DNA bisulfite sequencing. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 18761-18768.	3.3	363
12	Hypermethylated RASSF1A in Maternal Plasma: A Universal Fetal DNA Marker that Improves the Reliability of Noninvasive Prenatal Diagnosis. <i>Clinical Chemistry</i> , 2006, 52, 2211-2218.	1.5	319
13	Noninvasive prenatal diagnosis of monogenic diseases by digital size selection and relative mutation dosage on DNA in maternal plasma. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2008, 105, 19920-19925.	3.3	310
14	Detection of the placental epigenetic signature of the maspin gene in maternal plasma. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2005, 102, 14753-14758.	3.3	307
15	Epigenetics, fragmentomics, and topology of cell-free DNA in liquid biopsies. <i>Science</i> , 2021, 372, .	6.0	263
16	Position statement from the Chromosome Abnormality Screening Committee on behalf of the Board of the International Society for Prenatal Diagnosis. <i>Prenatal Diagnosis</i> , 2015, 35, 725-734.	1.1	243
17	Integrative single-cell and cell-free plasma RNA transcriptomics elucidates placental cellular dynamics. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2017, 114, E7786-E7795.	3.3	242
18	Size-based molecular diagnostics using plasma DNA for noninvasive prenatal testing. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, 8583-8588.	3.3	233

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19	High-Resolution Profiling of Fetal DNA Clearance from Maternal Plasma by Massively Parallel Sequencing. <i>Clinical Chemistry</i> , 2013, 59, 1228-1237.	1.5	202
20	Quantitative Analysis of Circulating Mitochondrial DNA in Plasma. <i>Clinical Chemistry</i> , 2003, 49, 719-726.	1.5	181
21	Orientation-aware plasma cell-free DNA fragmentation analysis in open chromatin regions informs tissue of origin. <i>Genome Research</i> , 2019, 29, 418-427.	2.4	159
22	Plasma DNA End-Motif Profiling as a Fragmentomic Marker in Cancer, Pregnancy, and Transplantation. <i>Cancer Discovery</i> , 2020, 10, 664-673.	7.7	152
23	Second generation noninvasive fetal genome analysis reveals de novo mutations, single-base parental inheritance, and preferred DNA ends. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, E8159-E8168.	3.3	142
24	Preferred end coordinates and somatic variants as signatures of circulating tumor DNA associated with hepatocellular carcinoma. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, E10925-E10933.	3.3	140
25	<i>Dnase1l3</i> deletion causes aberrations in length and end-motif frequencies in plasma DNA. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2019, 116, 641-649.	3.3	134
26	Noninvasive Prenatal Methylomic Analysis by Genomewide Bisulfite Sequencing of Maternal Plasma DNA. <i>Clinical Chemistry</i> , 2013, 59, 1583-1594.	1.5	131
27	Hypermethylation of RASSF1A in Human and Rhesus Placentas. <i>American Journal of Pathology</i> , 2007, 170, 941-950.	1.9	128
28	The Biology of Cell-free DNA Fragmentation and the Roles of DNASE1, DNASE1L3, and DFFB. <i>American Journal of Human Genetics</i> , 2020, 106, 202-214.	2.6	127
29	Maternal Plasma DNA Analysis with Massively Parallel Sequencing by Ligation for Noninvasive Prenatal Diagnosis of Trisomy 21. <i>Clinical Chemistry</i> , 2010, 56, 459-463.	1.5	125
30	Systematic Search for Placental DNA-Methylation Markers on Chromosome 21: Toward a Maternal Plasma-Based Epigenetic Test for Fetal Trisomy 21. <i>Clinical Chemistry</i> , 2008, 54, 500-511.	1.5	123
31	Noninvasive Prenatal Detection of Trisomy 21 by an Epigenetic Genetic Chromosome-Dosage Approach. <i>Clinical Chemistry</i> , 2010, 56, 90-98.	1.5	115
32	Sequencing-based counting and size profiling of plasma Epstein-Barr virus DNA enhance population screening of nasopharyngeal carcinoma. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, E5115-E5124.	3.3	114
33	Size-tagged preferred ends in maternal plasma DNA shed light on the production mechanism and show utility in noninvasive prenatal testing. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, E5106-E5114.	3.3	107
34	Identification and characterization of extrachromosomal circular DNA in maternal plasma. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2020, 117, 1658-1665.	3.3	106
35	Plasma DNA aberrations in systemic lupus erythematosus revealed by genomic and methylomic sequencing. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, E5302-11.	3.3	105
36	Nonhematopoietically Derived DNA Is Shorter than Hematopoietically Derived DNA in Plasma: A Transplantation Model. <i>Clinical Chemistry</i> , 2012, 58, 549-558.	1.5	103

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37	Maternal Plasma Fetal DNA Fractions in Pregnancies with Low and High Risks for Fetal Chromosomal Aneuploidies. PLoS ONE, 2014, 9, e88484.	1.1	92
38	Universal Haplotype-Based Noninvasive Prenatal Testing for Single Gene Diseases. Clinical Chemistry, 2017, 63, 513-524.	1.5	89
39	High Resolution Size Analysis of Fetal DNA in the Urine of Pregnant Women by Paired-End Massively Parallel Sequencing. PLoS ONE, 2012, 7, e48319.	1.1	86
40	Noninvasive twin zygosity assessment and aneuploidy detection by maternal plasma DNA sequencing. Prenatal Diagnosis, 2013, 33, 675-681.	1.1	75
41	Genome-wide detection of cytosine methylation by single molecule real-time sequencing. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	65
42	DNA of Erythroid Origin Is Present in Human Plasma and Informs the Types of Anemia. Clinical Chemistry, 2017, 63, 1614-1623.	1.5	63
43	Detection and characterization of jagged ends of double-stranded DNA in plasma. Genome Research, 2020, 30, 1144-1153.	2.4	61
44	Liver- and Colon-Specific DNA Methylation Markers in Plasma for Investigation of Colorectal Cancers with or without Liver Metastases. Clinical Chemistry, 2018, 64, 1239-1249.	1.5	60
45	Methy-Pipe: An Integrated Bioinformatics Pipeline for Whole Genome Bisulfite Sequencing Data Analysis. PLoS ONE, 2014, 9, e100360.	1.1	54
46	Methylation analysis of plasma DNA informs etiologies of Epstein-Barr virus-associated diseases. Nature Communications, 2019, 10, 3256.	5.8	52
47	Noninvasive Prenatal Testing by Nanopore Sequencing of Maternal Plasma DNA: Feasibility Assessment. Clinical Chemistry, 2015, 61, 1305-1306.	1.5	44
48	Single-molecule sequencing reveals a large population of long cell-free DNA molecules in maternal plasma. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	43
49	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
50	Plasma DNA Profile Associated with DNASE1L3 Gene Mutations: Clinical Observations, Relationships to Nuclease Substrate Preference, and In Vivo Correction. American Journal of Human Genetics, 2020, 107, 882-894.	2.6	37
51	DNase1 Does Not Appear to Play a Major Role in the Fragmentation of Plasma DNA in a Knockout Mouse Model. Clinical Chemistry, 2018, 64, 406-408.	1.5	34
52	Noninvasive Detection of Bladder Cancer by Shallow-Depth Genome-Wide Bisulfite Sequencing of Urinary Cell-Free DNA for Methylation and Copy Number Profiling. Clinical Chemistry, 2019, 65, 927-936.	1.5	34
53	FetalQuantSD: accurate quantification of fetal DNA fraction by shallow-depth sequencing of maternal plasma DNA. Npj Genomic Medicine, 2016, 1, 16013.	1.7	31
54	Application of fetal DNA in maternal plasma for noninvasive prenatal diagnosis. Expert Review of Molecular Diagnostics, 2002, 2, 32-40.	1.5	29

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55	The Biology and Diagnostic Applications of Plasma RNA. <i>Annals of the New York Academy of Sciences</i> , 2004, 1022, 135-139.	1.8	27
56	Noninvasive prenatal diagnosis empowered by high-throughput sequencing. <i>Prenatal Diagnosis</i> , 2012, 32, 401-406.	1.1	27
57	Racing Towards the Development of Diagnostics for a Novel Coronavirus (2019-nCoV). <i>Clinical Chemistry</i> , 2020, 66, 503-504.	1.5	26
58	Characteristics of Fetal Extrachromosomal Circular DNA in Maternal Plasma: Methylation Status and Clearance. <i>Clinical Chemistry</i> , 2021, 67, 788-796.	1.5	26
59	Noninvasive prenatal exclusion of congenital adrenal hyperplasia by maternal plasma analysis: a feasibility study. <i>Clinical Chemistry</i> , 2002, 48, 778-80.	1.5	25
60	Single-Stranded DNA Library Preparation Preferentially Enriches Short Maternal DNA in Maternal Plasma. <i>Clinical Chemistry</i> , 2017, 63, 1031-1037.	1.5	24
61	Jagged Ends of Urinary Cell-Free DNA: Characterization and Feasibility Assessment in Bladder Cancer Detection. <i>Clinical Chemistry</i> , 2021, 67, 621-630.	1.5	24
62	Proteomic analysis reveals platelet factor 4 and beta-thromboglobulin as prognostic markers in severe acute respiratory syndrome. <i>Electrophoresis</i> , 2012, 33, 1894-1900.	1.3	23
63	Single-Molecule Sequencing Enables Long Cell-Free DNA Detection and Direct Methylation Analysis for Cancer Patients. <i>Clinical Chemistry</i> , 2022, 68, 1151-1163.	1.5	22
64	Mass Spectrometry-Based Detection of Hemoglobin E Mutation by Allele-Specific Base Extension Reaction. <i>Clinical Chemistry</i> , 2007, 53, 2205-2209.	1.5	21
65	Molecular Diagnostics: A Revolution in Progress. <i>Clinical Chemistry</i> , 2015, 61, 1-3.	1.5	21
66	Early Detection of Cancer: Evaluation of MR Imaging Grading Systems in Patients with Suspected Nasopharyngeal Carcinoma. <i>American Journal of Neuroradiology</i> , 2020, 41, 515-521.	1.2	20
67	Topologic Analysis of Plasma Mitochondrial DNA Reveals the Coexistence of Both Linear and Circular Molecules. <i>Clinical Chemistry</i> , 2019, 65, 1161-1170.	1.5	19
68	Applications of genetic-epigenetic tissue mapping for plasma DNA in prenatal testing, transplantation and oncology. <i>ELife</i> , 2021, 10, .	2.8	19
69	Cell-Free DNA Fragmentomics: The New "Omics" on the Block. <i>Clinical Chemistry</i> , 2020, 66, 1480-1484.	1.5	18
70	COFFEE: control-free noninvasive fetal chromosomal examination using maternal plasma DNA. <i>Prenatal Diagnosis</i> , 2017, 37, 336-340.	1.1	17
71	Combined Count- and Size-Based Analysis of Maternal Plasma DNA for Noninvasive Prenatal Detection of Fetal Subchromosomal Aberrations Facilitates Elucidation of the Fetal and/or Maternal Origin of the Aberrations. <i>Clinical Chemistry</i> , 2017, 63, 495-502.	1.5	16
72	Noninvasive reconstruction of placental methylome from maternal plasma DNA: Potential for prenatal testing and monitoring. <i>Prenatal Diagnosis</i> , 2018, 38, 196-203.	1.1	16

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73	Cell-free fetal DNA coming in all sizes and shapes. <i>Prenatal Diagnosis</i> , 2021, 41, 1193-1201.	1.1	16
74	Epigenetic Analysis of RASSF1A Gene in Cell-Free DNA in Amniotic Fluid. <i>Clinical Chemistry</i> , 2007, 53, 796-798.	1.5	15
75	Circulating Nucleic Acids in Plasma/Serum III and Serum Proteomics Recent Developments in Fetal DNA in Maternal Plasma. <i>Annals of the New York Academy of Sciences</i> , 2004, 1022, 100-104.	1.8	14
76	Gestational Age Assessment by Methylation and Size Profiling of Maternal Plasma DNA: A Feasibility Study. <i>Clinical Chemistry</i> , 2017, 63, 606-608.	1.5	14
77	Effects of nucleases on cell-free extrachromosomal circular DNA. <i>JCI Insight</i> , 2022, 7, .	2.3	12
78	Fetal mitochondrial <scp>DNA</scp> in maternal plasma in surrogate pregnancies: Detection and topology. <i>Prenatal Diagnosis</i> , 2021, 41, 368-375.	1.1	11
79	Technical Optimization of RhD Zygosity Determination by Real-Time Quantitative Polymerase Chain Reaction. <i>Annals of the New York Academy of Sciences</i> , 2006, 945, 156-160.	1.8	10
80	Sequencing Analysis of Plasma Epstein-Barr Virus DNA Reveals Nasopharyngeal Carcinoma-Associated Single Nucleotide Variant Profiles. <i>Clinical Chemistry</i> , 2020, 66, 598-605.	1.5	10
81	Noninvasive Prenatal Diagnosis by Analysis of Fetal DNA in Maternal Plasma. , 2006, 336, 101-110.		9
82	Emerging Considerations for Noninvasive Prenatal Testing. <i>Clinical Chemistry</i> , 2017, 63, 946-953.	1.5	9
83	Single Cell and Plasma RNA Sequencing for RNA Liquid Biopsy for Hepatocellular Carcinoma. <i>Clinical Chemistry</i> , 2021, 67, 1492-1502.	1.5	9
84	The Next Frontier in Noninvasive Prenatal Diagnostics: Cell-Free Fetal DNA Analysis for Monogenic Disease Assessment. <i>Annual Review of Genomics and Human Genetics</i> , 2022, 23, 413-425.	2.5	9
85	Enrichment of fetal and maternal long cell-free DNA fragments from maternal plasma following DNA repair. <i>Prenatal Diagnosis</i> , 2019, 39, 88-99.	1.1	8
86	Jagged Ends on Multinucleosomal Cell-Free DNA Serve as a Biomarker for Nuclease Activity and Systemic Lupus Erythematosus. <i>Clinical Chemistry</i> , 2022, 68, 917-926.	1.5	7
87	Fetal Rhesus D mRNA Is Not Detectable in Maternal Plasma. <i>Clinical Chemistry</i> , 2005, 51, 2210-2211.	1.5	4
88	Nuclease deficiencies alter plasma cell-free DNA methylation profiles. <i>Genome Research</i> , 2021, 31, 2008-2021.	2.4	4
89	High-resolution analysis for urinary DNA jagged ends. <i>Npj Genomic Medicine</i> , 2022, 7, 14.	1.7	4
90	Noninvasive prenatal testing by maternal plasma DNA analysis: Current practice and future applications. <i>Scandinavian Journal of Clinical and Laboratory Investigation</i> , 2014, 74, 48-53.	0.6	3

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91	Molecular Diagnostics: Going from Strength to Strength. <i>Clinical Chemistry</i> , 2020, 66, 1-2.	1.5	2
92	Dissection of PIK3CA Aberration for Cervical Adenocarcinoma Outcomes. <i>Cancers</i> , 2021, 13, 3218.	1.7	2
93	Methy-Pipe: An integrated bioinformatics data analysis pipeline for whole genome methylome analysis. , 2010, , .		1
94	Current controversies in prenatal diagnosis 2: prediction and prevention of adverse pregnancy outcomes requires a genomic rather than proteomic solution. <i>Prenatal Diagnosis</i> , 2015, 35, 15-18.	1.1	1
95	Circulating microRNAs: In a Class of Their Own. <i>Clinical Chemistry</i> , 2020, 66, 257-258.	1.5	1
96	Nicht-invasive pränatale Diagnostik fetaler chromosomaler Aneuploidien mittels Nukleinsäureanalyse des mütterlichen Plasmas / Noninvasive prenatal diagnosis of fetal chromosomal aneuploidies by	0.1	0
97	Fastidious Detection of Circulating Tumor DNA Mutations in Residual Breast Cancer Disease for Ultimate Analytical Sensitivity and Specificity. <i>Clinical Chemistry</i> , 2020, 66, 866-867.	1.5	0