



Bilaga 4 Exkluderade studier

Artikel	Bedömning/orsak
Allyse M, Minear MA, Berson E, Sridhar S, Rote M, Hung A, et al. Non-invasive prenatal testing: A review of international implementation and challenges. International Journal of Women's Health, 2015; 7113-26.	Not primary study
Allyse M, Sayres LC, Goodspeed TA, Cho MK. Attitudes towards non-invasive prenatal testing for aneuploidy among US adults of reproductive age. Case Rep Genet, 2014.	Not primary study
Al-Mufti R, Hambley H, Farzaneh F, Nicolaidis KH. Investigation of maternal blood enriched for fetal cells: role in screening and diagnosis of fetal trisomies. American journal of medical genetics. 1999 Jul 2;85(1):66-75.	Method paper
Ashoor G, Poon L, Syngelaki A, Mosimann B, Nicolaidis KH. Fetal fraction in maternal plasma cell-free DNA at 11-13 weeks' gestation: effect of maternal and fetal factors. Fetal diagnosis and therapy. 2012;31(4):237-43.	Method paper
Ashoor G, Syngelaki A, Poon LC, Rezende JC, Nicolaidis KH. Fetal fraction in maternal plasma cell-free DNA at 11-13 weeks' gestation: relation to maternal and fetal characteristics. Ultrasound in obstetrics & gynecology : the official journal of the International Society of Ultrasound in Obstetrics and Gynecology. 2013 Jan;41(1):26-32.	Method paper
Avent ND, Madgett TE, Maddocks DG, Soothill PW. Cell-free fetal DNA in the maternal serum and plasma: current and evolving applications. Current opinion in obstetrics & gynecology. 2009 Apr;21(2):175-9.	Not primary study
Avent ND. Maternal plasma biomarkers for down syndrome: present and future. Drugs of today (Barcelona, Spain : 1998). 2013 Feb;49(2):145-52.	Not primary study
Avent ND. Refining noninvasive prenatal diagnosis with single-molecule next-generation sequencing. Clinical chemistry. 2012 Apr;58(4):657-8.	Method paper
Aymerich MACEDSEMM. The gap between systematic reviews and clinical practice: the case of prenatal diagnosis of Down syndrome. The Challenge of Collaboration 18th Annual Meeting of the International Society of Technology Assessment in Health Care, June 9-12, 2002 in Berlin, Germany [serial on the Internet]. 2002: Available from: http://onlinelibrary.wiley.com/o/cochrane/clcmr/articles/CMR-5008/frame.html .	Wrong intervention
Ayres AC, Whitty JA, Ellwood DA. A cost-effectiveness analysis comparing different strategies to implement noninvasive prenatal testing into a Down syndrome screening program. Aust N Z J Obstet Gynaecol, 2014; 54 (5): 412-7.	Not primary study

Bassiouny YA, Bayoumi YA, Gouda HM, Hassan AA. Is intracytoplasmic sperm injection (ICSI) associated with higher incidence of congenital anomalies? A single center prospective controlled study in Egypt. <i>Journal of maternal-fetal & neonatal medicine</i> , 2014; 27 (3): 279-82.	Wrong intervention
Bayindir B, Dehaspe L, Brison N, Brady P, Ardui S, Kammoun M, et al. Noninvasive prenatal testing using a novel analysis pipeline to screen for all autosomal fetal aneuploidies improves pregnancy management. <i>Eur J Hum Genet</i> , 2015.	Method paper
Beazoglou T, Heffley D, Kyriopoulos J, Vintzileos A, Benn P. Economic evaluation of prenatal screening for Down syndrome in the USA (Structured abstract). <i>Prenatal diagnosis</i> [serial on the Internet]. 1998; 18(12): Available from: http://onlinelibrary.wiley.com/o/cochrane/cleed/articles/NHSEED-21999000144/frame.html . <a href="http://onlinelibrary.wiley.com/doi/10.1002/(SICI)1097-0223(199812)18:12<1241::AID-PD440>3.0.CO;2-M/abstract">http://onlinelibrary.wiley.com/doi/10.1002/(SICI)1097-0223(199812)18:12<1241::AID-PD440>3.0.CO;2-M/abstract .	Not primary study
Bekker HL, Hewison J, Thornton JG. Applying decision analysis to facilitate informed decision making about prenatal diagnosis for Down syndrome: a randomised controlled trial. <i>Prenatal diagnosis</i> [serial on the Internet]. 2004; 24(4): Available from: http://onlinelibrary.wiley.com/o/cochrane/clcentral/articles/629/CN-00467629/frame.html	Wrong intervention
Benachi A, Costa JM. Non-invasive prenatal diagnosis of fetal aneuploidies. <i>Lancet</i> . 2007 Feb 10;369(9560):440-2.	Not primary study
Benn P, Borrell A, Cuckle H, Dugoff L, Gross S, Johnson JA, et al. Prenatal Detection of Down Syndrome using Massively Parallel Sequencing (MPS): A rapid response statement from a committee on behalf of the Board of the International Society for Prenatal Diagnosis, 24 October 2011. <i>Prenatal diagnosis</i> . 2012 January;32(1):1-2.	Not primary study
Benn P, Cuckle H, Pergament E. Non-invasive prenatal diagnosis for Down syndrome: the paradigm will shift, but slowly. <i>Ultrasound in obstetrics & gynecology : the official journal of the International Society of Ultrasound in Obstetrics and Gynecology</i> . 2012 Feb;39(2):127-30.	Not primary study
Benn P, Cuckle H. Theoretical performance of non-invasive prenatal testing for chromosome imbalances using counting of cell-free DNA fragments in maternal plasma. <i>Prenat Diagn</i> , 2014.	Method paper
Benn PA, Chapman AR. Practical and ethical considerations of noninvasive prenatal diagnosis. <i>JAMA - Journal of the American Medical Association</i> . 2009 27;301(20):2154-6.	Not primary study
Beolchi S, Fadin M, Paganelli A, Brambilla CI, Messina C, Morandi C. [The biochemical screening of Down's syndrome]. <i>Minerva ginecologica</i> . 1995 Jul-Aug;47(7-8):315-8.	Wrong language
Beulen L, Grutters JP, Faas BH, Feenstra I, van Vugt JM, Bekker MN. The consequences of implementing non-invasive prenatal testing in Dutch national health care: a cost-effectiveness analysis. <i>Eur J Obstet Gynecol Reprod Biol</i> , 2014; 18253-61.	Not primary study

Bevilacqua E, Gil MM, Nicolaides KH, Ordonez E, Cirigliano V, Dierickx H, et al. Performance of screening for aneuploidies by cell-free DNA analysis of maternal blood in twin pregnancies. <i>Ultrasound Obstet Gynecol</i> , 2015; 45 (1): 61-6.	Twin study
Bianchi DW, Maron JL, Johnson KL. Insights into fetal and neonatal development through analysis of cell-free RNA in body fluids. <i>Early human development</i> . 2010 Nov;86(11):747-52.	Not primary study
Bianchi DW, Oepkes D, Ghidini A. Current controversies in prenatal diagnosis 1: should noninvasive DNA testing be the standard screening test for Down syndrome in all pregnant women? <i>Prenatal diagnosis</i> . 2013 Sep 3:1-6.	Not primary study
Bianchi DW, Prosen T, Platt LD, Goldberg JD, Abuhamad AZ, Rava RP, et al. Massively parallel sequencing of maternal plasma DNA in 113 cases of fetal nuchal cystic hygroma. <i>Obstetrics and gynecology</i> . 2013 May;121(5):1057-62.	Method paper
Bianchi DW, Williams JM, Sullivan LM, Hanson FW, Klinger KW, Shuber AP. PCR quantitation of fetal cells in maternal blood in normal and aneuploid pregnancies. <i>American journal of human genetics</i> . 1997 Oct;61(4):822-9.	Method paper
Bianchi DW, Oepkes D, Ghidini A. Current controversies in prenatal diagnosis 1: should noninvasive DNA testing be the standard screening test for Down syndrome in all pregnant women? <i>Prenat Diagn</i> , 2014; 34 (1): 6-11.	Not primary study
Bianchi DW. Circulating fetal DNA: its origin and diagnostic potential-a review. <i>Placenta</i> . 2004 Apr;25 Suppl A:S93-s101.	Not primary study
Bianchi DW. Current knowledge about fetal blood cells in the maternal circulation. <i>Journal of perinatal medicine</i> . 1998;26(3):175-85.	Not primary study
Bianchi DW. Fetal cells in the mother: from genetic diagnosis to diseases associated with fetal cell microchimerism. <i>European journal of obstetrics, gynecology, and reproductive biology</i> . 2000 Sep;92(1):103-8.	Not primary study
Bianchi DW. Prenatal diagnosis by analysis of fetal cells in maternal blood. <i>The Journal of pediatrics</i> . 1995 Dec;127(6):847-56.	Not primary study
Bianchi DW. The 2010 ISPD meeting issue: World class science, World Cup football. <i>Prenatal diagnosis</i> . 2011 March;31(3):225-7.	Not primary study
Bischoff FZ, Lewis DE, Simpson JL. Cell-free fetal DNA in maternal blood: Kinetics, source and structure. <i>Human reproduction update</i> . 2005 January/February;11(1):59-67.	Not primary study

Bischoff FZ, Sinacori MK, Dang DD, Marquez-Do D, Horne C, Lewis DE, et al. Cell-free fetal DNA and intact fetal cells in maternal blood circulation: implications for first and second trimester non-invasive prenatal diagnosis. <i>Human reproduction update</i> . 2002 Nov-Dec;8(6):493-500.	Method paper
Brajenovic-Milic B, Babic I, Ristic S, Vranekovic J, Brumini G, Kapovic M. Pregnant women's attitudes toward amniocentesis before receiving Down syndrome screening results. <i>Women's health issues: official publication of the Jacobs Institute of Women's Health [serial on the Internet]</i> . 2008; 18(2): Available from: http://onlinelibrary.wiley.com/doi/10.1016/j.whi.2008.06.006	Wrong intervention
Buchanan A, Sachs A, Toler T, Tsipis J, Author A, Brandeis, et al. NIPT: Current utilization and implications for the future of prenatal genetic counseling. <i>Prenatal Diagnosis</i> , 2014; 34 (9): 850-57.	Not primary study
Bunce K, Chu T, Surti U, Hogge WA, Peters DG. Discovery of epigenetic biomarkers for the noninvasive diagnosis of fetal disease. <i>Prenatal diagnosis</i> . 2012 Jun;32(6):542-9.	Method paper
Bussani C, Cioni R, Mattei A, Fambrini M, Marchionni M, Scarselli G. Prenatal diagnosis of common aneuploidies in transcervical samples using quantitative fluorescent-PCR analysis. <i>Molecular diagnosis & therapy</i> . 2007;11(2):117-21.	Method paper
Bustamante-Aragones A, Rodriguez de Alba M, Perlado S, Trujillo-Tiebas MJ, Arranz JP, Diaz-Recasens J, et al. Non-invasive prenatal diagnosis of single-gene disorders from maternal blood. <i>Gene</i> . 2012 Aug 1;504(1):144-9.	Not primary study
Buysse K, Beulen L, Gomes I, Gilissen C, Keesmaat C, Janssen IM, et al. Reliable noninvasive prenatal testing by massively parallel sequencing of circulating cell-free DNA from maternal plasma processed up to 24h after venipuncture. <i>Clinical biochemistry</i> . 2013 Aug 8.	Method paper
Calabrese G, Baldi M, Fantasia D, Sessa MT, Kalantar M, Holzhauer C, et al. Detection of chromosomal aneuploidies in fetal cells isolated from maternal blood using single-chromosome dual-probe FISH analysis. <i>Clinical genetics</i> . 2012 Aug;82(2):131-9.	Method paper
Campagnoli C, Mulhaupt HA, Ludomirski A, Haut MJ, Warhol MJ. Noninvasive prenatal diagnosis. Use of density gradient centrifugation, magnetically activated cell sorting and in situ hybridization. <i>The Journal of reproductive medicine</i> . 1997 Apr;42(4):193-9.	Method paper
Canick JA, Kloza EM, Lambert-Messerlian GM, Haddow JE, Ehrich M, van den Boom D, et al. DNA sequencing of maternal plasma to identify Down syndrome and other trisomies in multiple gestations. <i>Prenatal diagnosis</i> . 2012 Aug;32(8):730-4.	Method paper
Canick JA, Palomaki GE, Kloza EM, Lambert-Messerlian GM, Haddow JE. The impact of maternal plasma DNA fetal fraction on next generation sequencing tests for common fetal aneuploidies. <i>Prenatal diagnosis</i> . 2013 Jul;33(7):667-74.	Not primary study

Canick JA, Saller DN, Jr., Lambert-Messerlian GM. Prenatal screening for Down syndrome: current and future methods. Clinics in laboratory medicine. 2003 Jun;23(2):395-411.	Not primary study
Carvalho Rodrigues L. False positive results of trisomy 21 prenatal screening as a surrogate marker for adverse pregnancy outcome. Diagnostico Prenatal, 2013; 24 (4): 135-40.	Wrong intervention
Cell-free fetal DNA tests for trisomy show promise in women at lower risk of affected pregnancies: Lower rates of false-positive returns, higher positive predictive value are associated with cfDNA tests versus standard screening panels, say experts AUTHOR ADDRESSES SOURCE American Journal of Medical Genetics, Part A (2014) 164:6 (8-9). Date of Publication: June 2014.	Not primary study
Cha DH, Farina A, Bianchi DW, Johnson KL. ROC analysis of an erythroblast morphologic scoring system to improve identification of fetal cells in maternal blood. Prenatal diagnosis. 2004 Feb;24(2):117-20.	Method paper
Cha DH, Khosrotehrani K, Bianchi DW, Johnson KL. The utility of an erythroblast scoring system and gender-independent short tandem repeat (STR) analysis for the detection of aneuploid fetal cells in maternal blood. Prenatal diagnosis. 2005 Jul;25(7):586-91.	Method paper
Chackin CJ. What potent blood: Non-invasive prenatal genetic diagnosis and the transformation of modern prenatal care. American Journal of Law and Medicine. 2007;33(1):9-53.	Not primary study
Chan YM, Leung TY, Chan OKC, Cheng YKY, Sahota DS, Author A, et al. Patient's choice between a non-invasive prenatal test and invasive prenatal diagnosis based on test accuracy. Fetal Diagnosis and Therapy, 2014; 35 (3): 193-98.	Not primary study
Chan YM, Leung WC, Chan WP, Leung TY, Cheng YK, Sahota DS. Women's uptake of non-invasive DNA testing following a high-risk screening test for trisomy 21 within a publicly funded healthcare system: findings from a retrospective review. Prenat Diagn, 2014.	Not primary study
Chandrananda D, Thorne NP, Ganesamoorthy D, Bruno DL, Benjamini Y, Speed TP, et al. Investigating and correcting plasma DNA sequencing coverage bias to enhance aneuploidy discovery. PLoS One, 2014; 9 (1): e86993.	Method paper
Chen C, Cram DS, Xie F, Wang P, Xu X, Li H, et al. A pregnancy with discordant fetal and placental chromosome 18 aneuploidies revealed by invasive and noninvasive prenatal diagnosis. Reproductive BioMedicine Online, 2014; 29 (1): 136-39.	Less than 100 samples
Chen M, Lee CP, Lam YH, Tang RY, Chan BC, Wong SF, et al. Comparison of nuchal and detailed morphology ultrasound examinations in early pregnancy for fetal structural abnormality screening: a randomized controlled trial. Ultrasound in obstetrics & gynecology : the official journal of the International Society of Ultrasound in Obstetrics and Gynecology [serial on the Internet]. 2008; 31(2): Available from: http://onlinelibrary.wiley.com/o/cochrane/clcentral/articles/640/CN-00647640/frame.html	Wrong intervention

Chen X,Xiong L,Zeng T, Xiao K, Huang Y,Guo H, et al. Hypermethylated ERG as a cell-free fetal DNA biomarker for non-invasive prenatal testing of Down syndrome. Clin Chim Acta, 2015; 444289-92.	Method paper
Chervenak FA,McCullough LB,Author A,Department of Obstetrics a,Gynecology NYPHWCMC,East 68th Street MPOBNYNYUS, et al. Ethical dimensions of first-trimester fetal aneuploidy screening. Clinical Obstetrics and Gynecology, 2014; 57 (1): 226-31.	Not primary study
Cheung SW, Patel A, Leung TY. Accurate Description of DNA-Based Noninvasive Prenatal Screening. N Engl J Med, 2015.	Not primary study
Chim SS, Jin S, Lee TY, Lun FM, Lee WS, Chan LY, et al. Systematic search for placental DNA-methylation markers on chromosome 21: toward a maternal plasma-based epigenetic test for fetal trisomy 21. Clinical chemistry. 2008 Mar;54(3):500-11.	Not primary study
Chitayat D, Langlois S, Wilson RD. Prenatal screening for fetal aneuploidy in singleton pregnancies. Journal of obstetrics and gynaecology Canada: JOGC = Journal d'obstetrique et gynecologie du Canada : JOGC. 2011 Jul;33(7):736-50.	Not primary study
Chitty LS, Finning K, Wade A,Soothill P,Martin B,Oxenford K, et al. Diagnostic accuracy of routine antenatal determination of fetal RHD status across gestation: population based cohort study. Bmj, 2014; 349g5243.	Not trisomy
Chitty LS, Hill M, White H, Wright D, Morris S. Noninvasive prenatal testing for aneuploidy-ready for prime time? American journal of obstetrics and gynecology. 2012 Apr;206(4):269-75.	Not primary study
Chitty LS, Kistler J, Akolekar R, Liddle S, Nicolaidis K, Levett L. Multiplex ligation-dependent probe amplification (MLPA): a reliable alternative for fetal chromosome analysis? The journal of maternal-fetal & neonatal medicine: the official journal of the European Association of Perinatal Medicine, the Federation of Asia and Oceania Perinatal Societies, the International Society of Perinatal Obstet. 2012 Aug;25(8):1383-6.	Wrong intervention
Chiu RW, Chan KC, Gao Y, Lau VY, Zheng W, Leung TY, et al. Noninvasive prenatal diagnosis of fetal chromosomal aneuploidy by massively parallel genomic sequencing of DNA in maternal plasma. Proceedings of the National Academy of Sciences of the United States of America. 2008 Dec 23;105(51):20458-63.	Less than 100 samples
Chiu RW, Lo YM. Non-invasive prenatal diagnosis by fetal nucleic acid analysis in maternal plasma: the coming of age. Seminars in fetal & neonatal medicine. 2011 Apr;16(2):88-93.	Not primary study
Chiu RW, Lo YM. Noninvasive prenatal diagnosis empowered by high-throughput sequencing. Prenatal diagnosis. 2012 Apr;32(4):401-6.	Method paper
Chiu RW, Sun H, Akolekar R, Clouser C, Lee C, McKernan K, et al. Maternal plasma DNA analysis with massively parallel sequencing by ligation for	Less than 100 samples

noninvasive prenatal diagnosis of trisomy 21. <i>Clinical chemistry</i> . 2010 Mar;56(3):459-63.	
Choi H, Lau TK, Jiang FM, Chan MK, Zhang HY, Lo PS, et al. Fetal aneuploidy screening by maternal plasma DNA sequencing: 'false positive' due to confined placental mosaicism. <i>Prenatal diagnosis</i> . 2013 Feb;33(2):198-200.	Method paper
Choolani M, Mahyuddin AP, Hahn S. The promise of fetal cells in maternal blood. <i>Best Practice and Research: Clinical Obstetrics and Gynaecology</i> . 2012 October;26(5):655-67.	Not primary study
Chu T, Burke B, Bunce K, Surti U, Allen Hogge W, Peters DG. A microarray-based approach for the identification of epigenetic biomarkers for the noninvasive diagnosis of fetal disease. <i>Prenatal diagnosis</i> . 2009 Nov;29(11):1020-30.	Method paper
Chueh J, Golbus MS. Prenatal diagnosis using fetal cells from the maternal circulation. <i>The Western journal of medicine</i> . 1993 Sep;159(3):308-11.	Not primary study
Clark-Ganheart CA, Iqbal SN, Brown DL, Black S, Fries MH. Understanding the Limitations of Circulating Cell Free Fetal DNA: An Example of Two Unique Cases. <i>J Clin Gynecol Obstet</i> , 2014; 3 (2): 38-70.	Less than 100 samples
Cole R, Jones G. Testing times: do new prenatal tests signal the end of Down syndrome? <i>The New Zealand medical journal</i> . 2013 Mar 1;126(1370):96-102.	Not primary study
Conner P, Gustafsson S, Kublickas M. First trimester contingent testing with either nuchal translucency or cell-free DNA, cost efficiency and the role of ultrasound dating. <i>Acta Obstet Gynecol Scand</i> , 2015; 94 (4): 368-75.	Not primary study
Conner SN, Longman RE, Cahill AG, Author A, Department of Obstetrics a, Gynecology DoMFMWU, et al. The role of ultrasound in the diagnosis of fetal genetic syndromes. <i>Best Practice and Research: Clinical Obstetrics and Gynaecology</i> , 2014; 28 (3): 417-28.	Not primary study
Cuckle H, Benn P, Pergament E. Maternal cfDNA screening for Down syndrome--a cost sensitivity analysis. <i>Prenatal diagnosis</i> . 2013 Jul;33(7):636-42.	Not primary study
Davis C, Cuckle H, Yaron Y. Screening for Down syndrome--incidental diagnosis of other aneuploidies. <i>Prenat Diagn</i> , 2014; 34 (11): 1044-8.	Wrong intervention
Davis DS. Opportunistic testing: the death of informed consent? <i>Health matrix (Cleveland, Ohio : 1991)</i> . 2013 Spring;23(1):35-54.	Not primary study
de Jong A, de Wert GM. Prenatal screening: an ethical agenda for the near future. <i>Bioethics</i> , 2015; 29 (1): 46-55.	Not primary study

De Jong A, Dondorp WJ, De Die-Smulders CEM, Frints SGM, De Wert GMWR. Non-invasive prenatal testing: Ethical issues explored. <i>European Journal of Human Genetics</i> . 2010 March;18(3):272-7.	Not primary study
de Jong A, Dondorp WJ, Frints SG, de Die-Smulders CE, de Wert GM. Non-invasive prenatal diagnosis for aneuploidy: toward an integral ethical assessment. <i>Human reproduction (Oxford, England)</i> . 2011 Nov;26(11):2915-7.	Not primary study
Deans Z, Newson AJ. Ethical considerations for choosing between possible models for using NIPD for aneuploidy detection. <i>Journal of medical ethics</i> . 2012 Oct;38(10):614-8.	Not primary study
Dhallan R, Guo X, Emche S, Damewood M, Bayliss P, Cronin M, et al. A non-invasive test for prenatal diagnosis based on fetal DNA present in maternal blood: a preliminary study. <i>Lancet</i> . 2007 Feb 10;369(9560):474-81.	Less than 100 samples
Dharajiya N, Zwiefelhofer T, Guan X, Angkachatchai V, Saldivar JS. Noninvasive prenatal testing using cell-free fetal DNA in maternal plasma. <i>Curr Protoc Hum Genet</i> , 2015;848.15.1-8.15.20.	Not primary study
Du Y, Zhang J, Wang H, Yan X, Yang Y, Yang L, et al. Hypomethylated DSCR4 is a placenta-derived epigenetic marker for trisomy 21. <i>Prenatal diagnosis</i> . 2011 Feb;31(2):207-14.	Method paper
Evans MI, Wright DA, Pergament E, Cuckle HS, Nicolaides KH. Digital PCR for noninvasive detection of aneuploidy: power analysis equations for feasibility. <i>Fetal diagnosis and therapy</i> . 2012;31(4):244-7.	Method paper
Evans MI, Sonek JD, Hallahan TW, Krantz DA. Cell-free fetal DNA screening in the USA: a cost analysis of screening strategies. <i>Ultrasound Obstet Gynecol</i> , 2015; 45 (1): 74-83.	Not primary study
Faas BH, Cirigliano V, Bui TH. Rapid methods for targeted prenatal diagnosis of common chromosome aneuploidies. <i>Seminars in fetal & neonatal medicine</i> . 2011 Apr;16(2):81-7.	Not primary study
Fairbrother G, Johnson S, Musci TJ, Song K. Clinical experience of noninvasive prenatal testing with cell-free DNA for fetal trisomies 21, 18, and 13, in a general screening population. <i>Prenatal diagnosis</i> . 2013 Jun;33(6):580-3.	No reference test
Fan HC, Blumenfeld YJ, Chitkara U, Hudgins L, Quake SR. Analysis of the size distributions of fetal and maternal cell-free DNA by paired-end sequencing. <i>Clinical chemistry</i> . 2010 Aug;56(8):1279-86.	Method paper
Fan HC, Blumenfeld YJ, Chitkara U, Hudgins L, Quake SR. Noninvasive diagnosis of fetal aneuploidy by shotgun sequencing DNA from maternal blood. <i>Proceedings of the National Academy of Sciences of the United States of America</i> . 2008 Oct 21;105(42):16266-71.	Less than 100 samples
Farina A, Bianchi DW. Fetal cells in maternal blood as a second non-invasive step for fetal Down syndrome screening. <i>Prenatal diagnosis</i> . 1998 Sep;18(9):983-4.	Not primary study

Farina A, Caramelli E, Concu M, Sekizawa A, Ruggeri R, Bovicelli L, et al. Testing normality of fetal DNA concentration in maternal plasma at 10-12 completed weeks' gestation: a preliminary approach to a new marker for genetic screening. <i>Prenatal diagnosis</i> . 2002 Feb;22(2):148-52.	Method paper
Favre R, Moutel G, Duchange N, Vayssiere C, Kohler M, Bouffet N, et al. What about informed consent in first-trimester ultrasound screening for Down syndrome? <i>Fetal diagnosis and therapy</i> . 2008;23(3):173-84.	Wrong intervention
Fiegler H, Geigl JB, Langer S, Rigler D, Porter K, Unger K, et al. High resolution array-CGH analysis of single cells. <i>Nucleic acids research</i> . 2007;35(3):e15.	Wrong intervention
Fuchs KM, Peipert JF. First trimester Down syndrome screening: public health implications. <i>Seminars in perinatology</i> . 2005 Aug;29(4):267-71.	Not primary study
Futch T, Spinosa J, Bhatt S, de Feo E, Rava RP, Sehnert AJ. Initial clinical laboratory experience in noninvasive prenatal testing for fetal aneuploidy from maternal plasma DNA samples. <i>Prenatal diagnosis</i> . 2013 Jun;33(6):569-74.	No reference test
Gagnon A, Audibert F, Author A, University of British Columbia V, Bc C, Universite de Montreal MQCC, et al. Prenatal screening and diagnosis of aneuploidy in multiple pregnancies. <i>Best Practice and Research: Clinical Obstetrics and Gynaecology</i> , 2014; 28 (2): 285-94.	Not primary study
Gao Y, Stejskal D, Jiang F, Wang W. False-negative trisomy 18 non-invasive prenatal test result due to 48,XXX,+18 placental mosaicism. <i>Ultrasound Obstet Gynecol</i> , 2014; 43 (4): 477-8.	Not primary study
Gekas J, Durand A, Bujold E, Vallee M, Forest JC, Rousseau F, et al. Cost-effectiveness and accuracy of prenatal Down syndrome screening strategies: should the combined test continue to be widely used? (Structured abstract). <i>American journal of obstetrics and gynecology [serial on the Internet]</i> . 2011; 204(2): Available from: http://onlinelibrary.wiley.com/o/cochrane/cleed/articles/NHSEED-22011000422/frame.html .	Wrong intervention
Georgsson Öhman S, Waldenström U. Effect of first-trimester ultrasound screening for Down syndrome on maternal-fetal attachment--a randomized controlled trial. <i>Sexual & reproductive healthcare : official journal of the Swedish Association of Midwives [serial on the Internet]</i> . 2010; 1(3): Available from: http://onlinelibrary.wiley.com/o/cochrane/clcentral/articles/979/CN-00786979/frame.html .	Not primary study
Gerovassili A, Garner C, Nicolaidis KH, Thein SL, Rees DC. Free fetal DNA in maternal circulation: a potential prognostic marker for chromosomal abnormalities? <i>Prenatal diagnosis</i> . 2007 Feb;27(2):104-10.	Method paper
Ghanta S, Mitchell ME, Ames M, Hidestrand M, Simpson P, Goetsch M, et al. Non-invasive prenatal detection of trisomy 21 using tandem single nucleotide polymorphisms. <i>PloS one</i> . 2010;5(10):e13184.	Less than 100 samples

Gil MM, Quezada MS, Bregant B, Ferraro M, Nicolaidis KH. Implementation of maternal blood cell-free DNA testing in early screening for aneuploidies. <i>Ultrasound in obstetrics & gynecology: the official journal of the International Society of Ultrasound in Obstetrics and Gynecology</i> . 2013 Jul;42(1):34-40.	No reference test
Gil MM, Akolekar R, Quezada MS, Bregant B, Nicolaidis KH. Analysis of Cell-Free DNA in Maternal Blood in Screening for Aneuploidies: Meta-Analysis. <i>Fetal Diagn Ther</i> , 2014.	Metaanalysis
Gil MM, Giunta G, Macalli EA, Poon LC, Nicolaidis KH. UK NHS pilot study on cell-free DNA testing in screening for fetal trisomies: factors affecting uptake. <i>Ultrasound Obstet Gynecol</i> , 2015; 45 (1): 67-73.	Not primary study
Gil MM, Quezada MS, Revello R, Akolekar R, Nicolaidis KH. Analysis of cell-free DNA in maternal blood in screening for fetal aneuploidies: updated meta-analysis. <i>Ultrasound Obstet Gynecol</i> , 2015; 45 (3): 249-66.	Metaanalysis
Go AT, van Vugt JM, Oudejans CB. Non-invasive aneuploidy detection using free fetal DNA and RNA in maternal plasma: recent progress and future possibilities. <i>Human reproduction update</i> . 2011 May-Jun;17(3):372-82.	Not primary study
Go AT, Visser A, Betsalel OT, van Vugt JM, Blankenstein MA, Oudejans CB. Measurement of allelic-expression ratios in trisomy 21 placentas by quencher extension of heterozygous samples identified by partially denaturing HPLC. <i>Clinical chemistry</i> . 2008 Feb;54(2):437-40.	Method paper
Go AT, Visser A, Mulders MA, Blankenstein MA, van Vugt JM, Oudejans CB. 44 single-nucleotide polymorphisms expressed by placental RNA: assessment for use in noninvasive prenatal diagnosis of trisomy 21. <i>Clinical chemistry</i> . 2007 Dec;53(12):2223-4.	Method paper
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