



Analys av foster-DNA i kvinnans blod: icke-invasiv fosterdiagnostik (NIPT) för trisomi 13, 18 och 21, Alertrapport 2015-03 (2015)

### Bilaga 3 Studier med låg kvalitet

Artikel	Bedömning/orsak
Beamon CJ,Hardisty EE,Harris SC, Vora NL. A single center's experience with noninvasive prenatal testing. <i>Genet Med</i> , 2014.	Low quality
Dar P,Curnow KJ,Gross SJ,Hall MP,Stosic M,Demko Z, et al. Clinical experience and follow-up with large scale single-nucleotide polymorphism-based noninvasive prenatal aneuploidy testing. <i>Am J Obstet Gynecol</i> , 2014; 211 (5): 527.e1-27.e17.	Low quality
Deng YH, Yin AH, He Q, Chen JC, He YS, Wang HQ, et al. Non-invasive prenatal diagnosis of trisomy 21 by reverse transcriptase multiplex ligation-dependent probe amplification. <i>Clinical chemistry and laboratory medicine: CCLM / FESCC</i> . 2011 Apr;49(4):641-6.	Low quality
Korostelev S, Totchiev G,Kanivets I,Gnetetskaya V,Author A,First Moscow Medical U, et al. Association of non-invasive prenatal testing and chromosomal microarray analysis for prenatal diagnostics. <i>Gynecological Endocrinology</i> , 2014; 30 Supplement113-16.	Low quality
Lambert-Messerlian G, Kloza EM,Williams J, 3rd,Loucky J,O'Brien B,Wilkins-Haug L, et al. Maternal plasma DNA testing for aneuploidy in pregnancies achieved by assisted reproductive technologies. <i>Genet Med</i> , 2014; 16 (5): 419-22.	Low quality
Lau TK, Chan MK, Lo PS, Chan HY, Chan WS, Koo TY, et al. Clinical utility of noninvasive fetal trisomy (NIFTY) test-early experience. <i>The journal of maternal-fetal &amp; 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</i> . 2012 Oct;25(10):1856-9.	Low quality
Li PQ, Zhang J, Fan JH, Zhang YZ, Hou HY. Development of noninvasive prenatal diagnosis of trisomy 21 by RT-MLPA with a new set of SNP markers. <i>Archives of gynecology and obstetrics</i> . 2013 Jul 5.	Low quality
Liao C,Yin AH,Peng CF,Fu F,Yang JX,Li R, et al. Noninvasive prenatal diagnosis of common aneuploidies by semiconductor sequencing. <i>Proc Natl Acad Sci U S A</i> , 2014.	Low quality
Lim JH, Kim SY, Park SY, Lee SY, Kim MJ, Han YJ, et al. Non-invasive epigenetic detection of fetal trisomy 21 in first trimester maternal plasma. <i>PloS one</i> . 2011;6(11):e27709.	Low quality

Manegold-Brauer G, Berg C, Flock A, Ruland A, Gembruch U, Geipel A. Uptake of non-invasive prenatal testing (NIPT) and impact on invasive procedures in a tertiary referral center. <i>Arch Gynecol Obstet</i> , 2015.	Low quality
McCullough RM, Almasri EA, Guan X, Geis JA, Hicks SC, Mazloom AR, et al. Non-invasive prenatal chromosomal aneuploidy testing--clinical experience: 100,000 clinical samples. <i>PLoS One</i> , 2014; 9 (10): e109173.	Low quality
Meck JM, Kramer Dugan E, Matyakhina L, Aviram A, Trunca C, Pineda-Alvarez D, et al. Non-Invasive Prenatal Screening for Aneuploidy: Positive Predictive Values Based on Cytogenetic Findings. <i>Am J Obstet Gynecol</i> , 2015.	Low quality
Poon CF, Tse WC, Kou KO, Leung KY. Uptake of Noninvasive Prenatal Testing in Chinese Women Following Positive Down Syndrome Screening. <i>Fetal Diagn Ther</i> , 2014.	Low quality
Sago H, Sekizawa A. Nationwide demonstration project of next-generation sequencing of cell-free DNA in maternal plasma in Japan: one-year experience. <i>Prenat Diagn</i> , 2014.	Low quality
Sehnert AJ, Rhees B, Comstock D, de Feo E, Heilek G, Burke J, et al. Optimal detection of fetal chromosomal abnormalities by massively parallel DNA sequencing of cell-free fetal DNA from maternal blood. <i>Clinical chemistry</i> . 2011 Jul;57(7):1042-9.	Low quality
Shah FT, French KS, Osann KE, Bocian M, Jones MC, Kerty L, et al. Impact of cell-free fetal DNA screening on patients' choice of invasive procedures after a positive california prenatal screen result. <i>Journal of Clinical Medicine</i> , 2014; 3 (3): 849-64.	Low quality
Zhang M, Li T, Chen J, Li L, Zhou C, Wang Y, et al. Non-invasive prenatal diagnosis of trisomy 21 by dosage ratio of fetal chromosome-specific epigenetic markers in maternal plasma. <i>Journal of Huazhong University of Science and Technology Medical sciences = Hua zhong ke ji da xue xue bao Yi xue Ying De wen ban = Huazhong keji daxue xuebao Yixue Yingdewen ban</i> . 2011 Oct;31(5):687-92.	Low quality
Zimmermann B, Hill M, Gemelos G, Demko Z, Banjevic M, Baner J, et al. Noninvasive prenatal aneuploidy testing of chromosomes 13, 18, 21, X, and Y, using targeted sequencing of polymorphic loci. <i>Prenatal diagnosis</i> . 2012 Dec;32(13):1233-41.	Low quality
Wallerstein R, Jelks A, Garabedian MJ. A new model for providing cell-free DNA and risk assessment for chromosome abnormalities in a public hospital setting. <i>J Pregnancy</i> , 2014; 2014962720.	Low quality

Wang JC,Sahoo T,Schonberg S,Kopita KA,Ross L,Patek K, et al. Discordant noninvasive prenatal testing and cytogenetic results: a study of 109 consecutive cases. Genet Med, 2014.	Low quality
Wax JR,Cartin A,Chard R,Lucas FL, Pinette MG. Noninvasive prenatal testing: Impact on genetic counseling, invasive prenatal diagnosis, and trisomy 21 detection. J Clin Ultrasound, 2015; 43 (1): 1-6.	Low quality
Fang Y,Wang G,Wang C,Suo F,Gu M, Xia Y. The Diagnosis Pattern of Mid-Trimester Fetal Chromosomal Aneuploidy in Xuzhou and the Clinical Applications. Cell Biochem Biophys, 2015.	Low quality