Appendix

Prenatal diagnosis through Chromosomal Microarray Analysis (CMA)
UTV2014/510

Appendix 3 Excluded studies and studies with high risk of bias

Excluded studies	
The Observatory. Genetic Testing. MLO: Medical Laboratory Observer, 2013; 45 (1): 6-6.	Not relevant study design: comment
Agatisa PK,Mercer MB,Leek AC,Smith MB,Philipson E, Farrell RM. A first look at women's perspectives on noninvasive prenatal testing to detect sex chromosome aneuploidies and microdeletion syndromes. Prenat Diagn, 2015; 35 (7): 692-8.	Not relevant PICO: intervention
Alesi V,Bertoli M,Sinibaldi L, Novelli A. The clinical utility and indications of chromosomal microarray analysis in prenatal diagnosis. Bjog, 2013; 120 (1): 119-20.	Not relevant study design: comment
Armengol L,Nevado J,Serra-Juhe C,Plaja A,Mediano C,Garcia-Santiago FA, et al. Clinical utility of chromosomal microarray analysis in invasive prenatal diagnosis. Hum Genet, 2012; 131 (3): 513-23.	Micrarray platform not specified
Avent ND. Refining noninvasive prenatal diagnosis with single-molecule next-generation sequencing. Clin Chem, 2012; 58 (4): 657-8.	Not relevant study design : editorial
Bao B,Wang Y,Hu H,Yao H,Li Y,Tang S, et al. Karyotypic and molecular genetic changes associated with fetal cardiovascular abnormalities: results of a retrospective 4-year ultrasonic diagnosis study. Int J Biol Sci, 2013; 9 (5): 463-71.	Not relevant PICO: Population less than 20 patients
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. Eur J Hum Genet, 2015.	Targeted prenatal genetic testing for a specific aberration
Benachi A,Letourneau A,Kleinfinger P,Senat MV,Gautier E,Favre R, et al. Cell-free DNA analysis in maternal plasma in cases of fetal abnormalities detected on ultrasound examination. Obstet Gynecol, 2015; 125 (6): 1330-7.	Not relevant PICO: outcome
Bi W,Breman A,Shaw CA,Stankiewicz P,Gambin T,Lu X, et al. Detection of >/=1Mb microdeletions and microduplications in a single cell using custom oligonucleotide arrays. Prenatal Diagnosis 2012; 32 (1): 10-20.	Not relevant PICO: population

Bianchi DW,Platt LD,Goldberg JD,Abuhamad AZ,Sehnert AJ, Rava RP. Genome-wide fetal aneuploidy detection by maternal plasma DNA sequencing. Obstet Gynecol, 2012; 119 (5): 890-901.	Not relevant PICO: intervention
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. Obstet Gynecol, 2013; 121 (5): 1057-62.	Not relevant PICO: intervention
Bianchi DW, Wilkins-Haug L. Integration of noninvasive DNA testing for aneuploidy into prenatal care: what has happened since the rubber met the road? Clin Chem, 2014; 60 (1): 78-87.	Not relevant study design
Bianchi DW,Parsa S,Bhatt S,Halks-Miller M,Kurtzman K,Sehnert AJ, et al. Fetal sex chromosome testing by maternal plasma DNA sequencing: clinical laboratory experience and biology. Obstet Gynecol, 2015; 125 (2): 375-82.	Not relevant PICO: intervention
Biesecker LG, Green RC. Diagnostic clinical genome and exome sequencing. New England Journal of Medicine, 2014; 370 (25): 2418-25.	Not relevant study design: comment
Blackburn HL,Schroeder B,Turner C,Shriver CD,Ellsworth DL, Ellsworth RE. Management of Incidental Findings in the Era of Next-generation Sequencing. Curr Genomics, 2015; 16 (3): 159-74.	Not relevant study design
Bouffard C,Viville S, Knoppers BM. Genetic diagnosis of embryos: clear explanation, not rhetoric, is needed. CMAJ: Canadian Medical Association Journal, 2009; 181 (6-7): 387-91.	Not relevant study design
Bovolenta M,Rimessi P,Dolcini B,Ravani A,Ferlini A, Gualandi F. Prenatal diagnosis of Duchenne muscular dystrophy by comparative genomic hybridization. Clin Genet, 2010; 77 (5): 503-6.	Not relevant study design case report
Brady PD,Devriendt K,Deprest J, Vermeesch JR. Array-based approaches in prenatal diagnosis. Methods Mol Biol 2012; 838151-71.	Not relevant study design
Brady PD, Vermeesch JR. Genomic microarrays: a technology overview. Prenat Diagn, 2012; 32 (4): 336-43.	Not relevant study design
Burnell L,Verchere C,Pugash D,Loock C,Robertson S,Lehman A, et al. Additional post-natal diagnoses following antenatal diagnosis of isolated cleft lip +/- Palate. Archives of Disease in Childhood: Fetal and Neonatal Edition, 2014; 99 (4): F286-F90.	Not relevant PICO: intervention
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. Gene 2012; 504 (1): 144-9.	Not relevant study design

Callaway JL,Huang S,Karampetsou E, Crolla JA. Perspective on the technical challenges involved in the implementation of array-CGH in prenatal diagnostic testing. Mol Biotechnol 2014; 56 (4): 312-8.	Not relevant PICO: outcome
Canton AP,Costa SS,Rodrigues TC,Bertola DR,Malaquias AC,Correa FA, et al. Genome-wide screening of copy number variants in children born small for gestational age reveals several candidate genes involved in growth pathways. Eur J Endocrinol 2014; 171 (2): 253-62.	Not relevant PICO: population
Carss KJ,Hillman SC,Parthiban V,McMullan DJ,Maher ER,Kilby MD, et al. Exome sequencing improves genetic diagnosis of structural fetal abnormalities revealed by ultrasound. Hum Mol Genet, 2014; 23 (12): 3269-77.	Not relevant PICO: intervention
Cavalli P,Cavallari U, Novelli A. Array CGH in routine prenatal diagnosis practice. Prenat Diagn 2012; 32 (7): 708-9; author reply 11-2.	Not relevant study design letter
Chan K,Yam I,Leung KY,Tang M,Chan TK, Chan V. Detection of paternal alleles in maternal plasma for non-invasive prenatal diagnosis of beta-thalassemia: a feasibility study in southern Chinese. Eur J Obstet Gynecol Reprod Biol, 2010; 150 (1): 28-33.	Targeted prenatal genetic testing for a specific aberration
Char D. Preventive Genomic Sequencing and Care of the Individual Patient. American Journal of Bioethics, 2015; 15 (7): 32-3.	Not relevant study design
Chen CP,Chang SD,Chueh HY,Su YN,Su JW,Chern SR, et al. Rapid positive confirmation of trisomy 21 mosaicism at amniocentesis by interphase FISH, QF-PCR and aCGH on uncultured amniocytes. Taiwan J Obstet Gynecol, 2012; 51 (3): 475-80.	Not relevant study design case study
Chen CP,Su YN,Chern SR,Chen YT,Su JW,Pan CW, et al. Prenatal diagnosis of trisomy 8 mosaicism. Taiwan J Obstet Gynecol, 2012; 51 (4): 666-8.	Not relevant study design: case report
Chen CP,Su YN,Chern SR,Tsai FJ,Hsu CY,Lee CC, et al. Prenatal diagnosis of a 4.9-Mb deletion of 10q11.21 --> q11.23 by array comparative genomic hybridization. Taiwan J Obstet Gynecol, 2010; 49 (1): 117-9.	Not relevant study design: case study
Chen CP,Su YN,Chern SR,Wu PS,Su JW,Town DD, et al. Prenatal diagnosis of an interstitial deletion of 10q (10q11.21 --> q21.1): array comparative genomic hybridization characterization and literature review. Taiwan J Obstet Gynecol, 2012; 51 (4): 672-6.	Not relevant study design: case report

Chen CP,Su YN,Hsu CY,Tsai FJ,Chien SC,Chern SR, et al. Abnormally flat facial profile on two- and three-dimensional ultrasound and array comparative genomic hybridization for the diagnosis of Pallister-Killian syndrome. Taiwan J Obstet Gynecol, 2010; 49 (1): 124-8.	Not relevant study design: case study
Chen CP,Su YN,Tsai FJ,Chern SR,Hsu CY,Huang MC, et al. Rapid genome-wide aneuploidy diagnosis using uncultured amniocytes and array comparative genomic hybridization in pregnancy with abnormal ultrasound findings detected in late second and third trimesters. Taiwan J Obstet Gynecol, 2010; 49 (1): 120-3.	Not relevant study design: case study
Chen CP,Wu PC,Lin CJ,Su YN,Chern SR,Tsai FJ, et al. Balanced reciprocal translocations detected at amniocentesis. Taiwan J Obstet Gynecol, 2010; 49 (4): 455-67.	Not relevant PICO: intervention
Chen M,Yang YS,Shih JC,Lin WH,Lee DJ,Lin YS, et al. Microdeletions/duplications involving TBX1 gene in fetuses with conotruncal heart defects which are negative for 22q11.2 deletion on fluorescence in-situ hybridization. Ultrasound Obstet Gynecol 2014; 43 (4): 396-403.	Not relevant PICO: population less than 20 patients
Chen S,Ge H,Wang X,Pan X,Yao X,Li X, et al. Haplotype-assisted accurate non-invasive fetal whole genome recovery through maternal plasma sequencing. Genome Med, 2013; 5 (2): 18.	Not relevant study design: case study
Chen S,Lau TK,Zhang C,Xu C,Xu Z,Hu P, et al. A method for noninvasive detection of fetal large deletions/duplications by low coverage massively parallel sequencing. Prenat Diagn, 2013; 33 (6): 584-90.	Not relevant PICO: intervention
Chen X,Zhao KX, Zhao C. [The development and importance of molecular diagnosis in hereditary retinal diseases]. Zhonghua Yan Ke Za Zhi 2013; 49 (6): 484-7.	Language
Chitty LS,Mason S,Barrett AN,McKay F,Lench N,Daley R, et al. Non-invasive prenatal diagnosis of achondroplasia and thanatophoric dysplasia: next generation sequencing allows for a safer, more accurate and comprehensive approach. Prenat Diagn, 2015.	Targeted prenatal genetic testing for a specific aberration
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. Proc Natl Acad Sci U S A, 2008; 105 (51): 20458-63.	Not relevant PICO: outcome

Choy KW,Kwok YK,Cheng YK,Wong KM,Wong HK,Leung KO, et al. Diagnostic accuracy of the BACs-on-Beads assay versus karyotyping for prenatal detection of chromosomal abnormalities: a retrospective consecutive case series. Bjog 2014; 121 (10): 1245-52.	Not relevant PICO: intervention
Choy KW,Tsang PT,Leung TY,Wang CC,Lau TK,Author A, et al. The application of microarray based comparative genomic hybridization in prenatal diagnosis. Fetal and Maternal Medicine Review, 2008; 19 (2): 119-33.	Not relevant study design
Cohen K,Tzika A,Wood H,Berri S,Roberts P,Mason G, et al. Diagnosis of fetal submicroscopic chromosomal abnormalities in failed array CGH samples: copy number by sequencing as an alternative to microarrays for invasive fetal testing. Ultrasound Obstet Gynecol, 2015; 45 (4): 394-401.	Not relevant PICO: Control
Comas C,Echevarria M,Rodriguez MA,Prats P,Rodriguez I, Serra B. Initial experience with non-invasive prenatal testing of cell-free DNA for major chromosomal anomalies in a clinical setting. J Matern Fetal Neonatal Med, 2015; 28 (10): 1196-201.	Not relevant PICO: intervention
Coppinger J,Alliman S,Lamb AN,Torchia BS,Bejjani BA, Shaffer LG. Whole-genome microarray analysis in prenatal specimens identifies clinically significant chromosome alterations without increase in results of unclear significance compared to targeted microarray. Prenat Diagn, 2009; 29 (12): 1156-66.	Data also part of a later publication
Coromilas A,Wynn J,Haverfield E, Chung WK. Nonspecific phenotype of Noonan syndrome diagnosed by whole exome sequencing. Clin Case Rep, 2015; 3 (4): 237-9.	Not relevant study design: case report
Costain G,McDonald-McGinn DM, Bassett AS. Prenatal genetic testing with chromosomal microarray analysis identifies major risk variants for schizophrenia and other later-onset disorders. Am J Psychiatry, 2013; 170 (12): 1498.	Not relevant study design
Dan S,Chen F,Choy KW,Jiang F,Lin J,Xuan Z, et al. Prenatal detection of aneuploidy and imbalanced chromosomal arrangements by massively parallel sequencing. PLoS One, 2012; 7 (2): e27835.	Not relevant PICO: intervention
D'Angelo G,Di Rienzo T, Ojetti V. Microarray analysis in gastric cancer: a review. World J Gastroenterol 2014; 20 (34): 11972-6.	Not relevant PICO: population
Dar Pe,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. American Journal of Obstetrics and Gynecology, 2014; 211 (5): 527.e1-27.e17.	Not relevant PICO: intervention

Darilek S,Ward P,Pursley A,Plunkett K,Furman P,Magoulas P, et al. Pre- and postnatal genetic testing by array-comparative genomic hybridization: genetic counseling perspectives. Genet Med, 2008; 10 (1): 13-8.	Not relevant study design
Davies N. Prenatal screening with microarray technology. MLO: Medical Laboratory Observer, 2015; 47 (5): 8-12.	Not relevant study design
de Jong A,Dondorp WJ,Krumeich A,Boonekamp J,van Lith JM, de Wert GM. The scope of prenatal diagnosis for women at increased risk for aneuploidies: views and preferences of professionals and potential users. J Community Genet, 2013; 4 (1): 125-35.	Not relevant PICO: population
Deshpande M,Harper J,Holloway M,Palmer R, Wang R. Evaluation of array comparative genomic hybridization for genetic analysis of chorionic villus sampling from pregnancy loss in comparison to karyotyping and multiplex ligation-dependent probe amplification. Genet Test Mol Biomarkers, 2010; 14 (3): 421-4.	Not relevant PICO: population
Dickinson JE,Harcourt E, Murch A. The selective use of rapid aneuploidy screening in prenatal diagnosis. Australian & New Zealand Journal of Obstetrics & Gynaecology, 2009; 49 (1): 28-33.	Not relevant PICO: intervention
Dondorp WJ,Page-Christiaens GC, de Wert GM. Genomic futures of prenatal screening: ethical reflection. Clin Genet, 2015.	Not relevant study design
Donnelly JC,Wapner RJ,Simpson JL,Author A,Department o,Obstetrics, et al. Can microarray analysis reveal additional abnormalities in prenatal diagnosis? Expert Review of Obstetrics and Gynecology, 2013; 8 (3): 193-95.	Not relevant study design
Drury S,Williams H,Trump N,Boustred C,Lench N,Scott RH, et al. Exome sequencing for prenatal diagnosis of fetuses with sonographic abnormalities. Prenat Diagn, 2015.	Not relevant PICO: intervention
Dugo N,Padula F,Mobili L,Brizzi C,D'Emidio L,Cignini P, et al. Six consecutive false positive cases from cell-free fetal DNA testing in a single referring centre. J Prenat Med, 2014; 8 (1-2): 31-5.	Not relevant PICO: intervention
Dugoff L. Application of genomic technology in prenatal diagnosis. N Engl J Med 2012; 367 (23): 2249-51.	Not relevant study design : editorial
Duncan A, Langlois S. Use of array genomic hybridization technology in prenatal diagnosis in Canada. J Obstet Gynaecol Can, 2011; 33 (12): 1256-9.	Not relevant study design
Dupont C,Grati FR,Choy KW,Jaillard S,Toutain J,Maurin ML, et al. Prenatal diagnosis of 24 cases of microduplication 22q11.2:	Not relevant PICO: Population less than 20 patients

an investigation of phenotype-genotype correlations. Prenat Diagn 2014.	
Eng CM,Yang Y, Plon SE. Genetic diagnosis through whole-exome sequencing... N Engl J Med. 2014 Mar 13;370(11):1067–8; N Engl J Med. 2013 Oct 17;369(16):1502–11; N Engl J Med. 2014 Mar 13;370(11):1067. New England Journal of Medicine, 2014; 370 (11): 1068–68.	Not relevant study design: letter
Faas BH,van der Burgt I,Kooper AJ,Pfundt R,Hehir-Kwa JY,Smits AP, et al. Identification of clinically significant, submicroscopic chromosome alterations and UPD in fetuses with ultrasound anomalies using genome-wide 250k SNP array analysis. J Med Genet, 2010; 47 (9): 586–94.	Not relevant PICO: population
Fan HC,Gu W,Wang J,Blumenfeld YJ,El-Sayed YY, Quake SR. Non-invasive prenatal measurement of the fetal genome. Nature 2012; 487 (7407): 320–4.	Not relevant PICO: intervention
Fan HC, Quake SR. Sensitivity of noninvasive prenatal detection of fetal aneuploidy from maternal plasma using shotgun sequencing is limited only by counting statistics. PLoS One, 2010; 5 (5): e10439.	Not relevant PICO: intervention
Faucett A, Savage M. Chromosomal microarray testing. JAAPA: Journal of the American Academy of Physician Assistants (Haymarket Media, Inc.), 2012; 25 (1): 65–66.	Not relevant study design
Filges I,Kang A,Hench J,Wenzel F,Bruder E,Miny P, et al. Fetal polydactyly: a study of 24 cases ascertained by prenatal sonography. J Ultrasound Med, 2011; 30 (7): 1021–9.	Not relevant study design : case series
Fiorentino F. Re: Use of prenatal chromosomal microarray: prospective cohort study and systematic review and meta-analysis. S. C. Hillman, D. J. McMullan, G. Hall, F. S. Togneri, N. James, E. J. Maher, C. H. Meller, D. Williams, R. J. Wapner, E. R. Maher and M. D. Kilby. Ultrasound obstet gynecol 2013; 41: 610–620. Ultrasound Obstet Gynecol, 2013; 41 (6): 608.	Not relevant study design: comment
Fiorentino F,Biricik A,Bono S,Spizzichino L,Cotroneo E,Cottone G, et al. Development and validation of a next-generation sequencing-based protocol for 24-chromosome aneuploidy screening of embryos. Fertil Steril, 2014; 101 (5): 1375–82.	Not relevant PICO: intervention
Fiorentino F,Caiazzo F,Napolitano S,Spizzichino L,Bono S,Sessa M, et al. Introducing array comparative genomic hybridization into routine prenatal diagnosis practice: a prospective study on over 1000 consecutive clinical cases. Prenat Diagn, 2011; 31 (13): 1270–82.	Data also part of a later publication

Fisher A,Bonner C,Biankin AV, Juraskova I. Factors influencing intention to undergo whole genome screening in future healthcare: A single-blind parallel-group randomised trial. Preventive Medicine, 2012; 55 (5): 514–20.	Not relevant study design
Foglieni B,Galbiati S,Ferrari M, Cremonesi L. A fast microelectronic array for screening and prenatal diagnosis of beta-thalassemia. Methods Mol Biol, 2008; 444169–82.	Targeted prenatal genetic testing for a specific aberration
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. Prenat Diagn, 2013; 33 (6): 569–74.	Not relevant PICO: intervention
Galbiati S,Brisci A,Damin F,Gentilin B,Curcio C,Restagno G, et al. Fetal DNA in maternal plasma: a noninvasive tool for prenatal diagnosis of beta-thalassemia. Expert Opin Biol Ther 2012; 12 Suppl 1S181–7.	Not relevant PICO: intervention
Garcia-Herrero S,Campos-Galindo I,Martinez-Conejero JA,Serra V,Olmo I,Lara C, et al. BACs-on-Beads technology: a reliable test for rapid detection of aneuploidies and microdeletions in prenatal diagnosis. Biomed Res Int 2014; 2014590298.	Not relevant PICO: intervention
Gekas J,Sergi C, Kamnasaran D. Molecular prenatal diagnosis of a sporadic alobar holoprosencephalic fetus: genotype-phenotype correlations. J Prenat Med, 2012; 6 (3): 36–9.	Not relevant study design: case study
Gil MM,Quezada MS,Revello R,Akolekar R, Nicolaides KH. Analysis of cell-free DNA in maternal blood in screening for fetal aneuploidies: updated meta-analysis. Ultrasound Obstet Gynecol, 2015; 45 (3): 249–66.	Not relevant study design
Giorlandino C,Mesoraca A,Bizzoco D,Dello Russo C,Cima A,Di Giacomo G, et al. Introducing the next generation sequencing in genomic amnio and villuos sampling. The so called "Next Generation Prenatal Diagnosis" (NGPD). J Prenat Med, 2014; 8 (1–2): 1–10.	Not relevant study design
Grati FR,Ferreira JC, Bajaj K. Noninvasive prenatal screening for fetal trisomies 21, 18, 13 and the common sex chromosome aneuploidies from maternal blood using massively parallel genomic sequencing of DNA. Am J Obstet Gynecol, 2014; 211 (6): 711–12.	Not relevant study design
Grati FR,Vialard F, Gross S. BACs-on-Beads (BoBs) assay for the genetic evaluation of prenatal samples and products of conception. Methods Mol Biol 2015; 1227259–78.	Not relevant PICO: intervention
Gruchy N,Lebrun M,Herlicoviez M,Alliet J,Gourdier D,Kottler ML, et al. Supernumerary marker chromosomes management	Not relevant PICO: intervention

in prenatal diagnosis. Am J Med Genet A, 2008; 146a (21): 2770–6.	
Gu H,Li H,Zhang L,Luan H,Huang T,Wang L, et al. Diagnostic role of microRNA expression profile in the serum of pregnant women with fetuses with neural tube defects. J Neurochem, 2012; 122 (3): 641–9.	Not relevant PICO: intervention
Guerra C,Rendeiro P,Pereira E,Rosmaninho A,Nogueira R,Pereira S, et al. Microarray for skeletal dysplasias: thanatophoric dysplasia diagnosed in utero using microarray technology. Ultrasound Obstet Gynecol, 2013; 41 (1): 95–6.	Not relevant PICO: Population less than 20 patients
Guex N,Iseli C,Syngelaki A,Deluen C,Pescia G,Nicolaides KH, et al. A robust second-generation genome-wide test for fetal aneuploidy based on shotgun sequencing cell-free DNA in maternal blood. Prenat Diagn, 2013; 33 (7): 707–10.	Not relevant PICO: intervention
Hall GK,Mackie FL,Hamilton S,Evans A,McMullan DJ,Williams D, et al. Chromosomal microarray analysis allows prenatal detection of low level mosaic autosomal aneuploidy. Prenat Diagn, 2014; 34 (5): 505–7.	Not relevant PICO: Population less than 20 patients
Helgeson J,Wardrop J,Boomer T,Almasri E,Paxton WB,Saldivar JS, et al. Clinical outcome of subchromosomal events detected by whole-genome noninvasive prenatal testing. Prenat Diagn, 2015.	Not relevant PICO: intervention
Hill M,Twiss P,Verhoef TI,Drury S,McKay F,Mason S, et al. Non-invasive prenatal diagnosis for cystic fibrosis: detection of paternal mutations, exploration of patient preferences and cost analysis. Prenat Diagn, 2015.	Not relevant PICO: intervention
Hillman S,McMullan D,Maher E, Kilby M. Clinical utility of array comparative genomic hybridisation for prenatal diagnosis: a cohort study of 3171 pregnancies... BJOG. 2012 Apr;119(5):614–25. BJOG: An International Journal of Obstetrics & Gynaecology, 2012; 119 (10): 1281–82.	Not relevant study design : editorial
Hillman SC,Barton PM,Roberts TE,Maher ER,McMullan DM, Kilby MD. BAC chromosomal microarray for prenatal detection of chromosome anomalies in fetal ultrasound anomalies: an economic evaluation. Fetal Diagn Ther 2014; 36 (1): 49–58.	Not relevant PICO: outcome
Hillman SC,McMullan DJ,Maher ER, Kilby MD. Clinical utility of array comparative genomic hybridisation for prenatal diagnosis: a cohort study of 3171 pregnancies. Bjog, 2012; 119 (10): 1281–2; author reply 82.	Not relevant study design: letter
Hillman SC,McMullan DJ,Maher ER, Kilby MD. Meeting the challenge of interpreting high-resolution single nucleotide polymorphism array data: does increased diagnostic power	Not relevant study design

outweigh the dilemma of rare variants. Bjog 2013; 120 (10): 1296.	
Hillman SC,McMullan DJ,Silcock L,Maher ER, Kilby MD. How does altering the resolution of chromosomal microarray analysis in the prenatal setting affect the rates of pathological and uncertain findings? J Matern Fetal Neonatal Med, 2014; 27 (7): 649–57.	Data also part of a later publication included in the systematic report
Hillman SC,McMullan DJ,Williams D,Maher ER, Kilby MD. Microarray comparative genomic hybridization in prenatal diagnosis: a review. Ultrasound Obstet Gynecol 2012; 40 (4): 385–91.	Not relevant study design
Holm S, Ploug T. Patient Choice and Preventive Genomic Sequencing—More Trouble Upstream. American Journal of Bioethics, 2015; 15 (7): 24–6.	Not relevant study design
Hooks J,Wolfberg AJ,Wang ET,Struble CA,Zahn J,Juneau K, et al. Non-invasive risk assessment of fetal sex chromosome aneuploidy through directed analysis and incorporation of fetal fraction. Prenat Diagn, 2014; 34 (5): 496–9.	Not relevant PICO: intervention
Hua R,Barrett AN,Tan TZ,Huang Z,Mahyuddin AP,Ponnusamy S, et al. Detection of aneuploidy from single fetal nucleated red blood cells using whole genome sequencing. Prenat Diagn, 2014.	Not relevant PICO: intervention
Hui L. Re: Non-invasive prenatal testing for fetal chromosomal abnormalities by low-coverage whole-genome sequencing of maternal plasma DNA: review of 1982 consecutive cases in a single center. T. K. Lau, S. W. Cheung, P. S. S. Lo, A. N. Pursley, M. K. Chan, F. Jiang, H. Zhang, W. Wang, L. F. J. Jong, O. K. C. Yuen, H. Y. C. Chan, W. S. K. Chan and K. W. Choy. Ultrasound Obstet Gynecol 2014; 43: 254–264. Ultrasound Obstet Gynecol, 2014; 43 (3): 245.	Not relevant study design
Jacob HJ. Next-generation sequencing for clinical diagnostics. New England Journal of Medicine, 2013; 369 (16): 1557–58.	Not relevant study design: comment
Jacob HJ. Genetic diagnosis through whole-exome sequencing... N Engl J Med. 2013 Oct 17;369(16):1557–8; N Engl J Med. 2014 Mar 13;370(11):1068. New England Journal of Medicine, 2014; 370 (11): 1069–69.	Not relevant study design: comment
Jensen TJ,Kim SK,van den Boom D,Deciu C, Ehrich M. Noninvasive detection of a balanced fetal translocation from maternal plasma. Clin Chem, 2014; 60 (10): 1298–305.	Not relevant study design: case study

Jensen TJ,Zwiefelhofer T,Tim RC,Dzakula Z,Kim SK,Mazloom AR, et al. High-throughput massively parallel sequencing for fetal aneuploidy detection from maternal plasma. PLoS One, 2013; 8 (3): e57381.	Not relevant PICO: intervention
Jeon KC,Chen LS, Goodson P. Decision to abort after a prenatal diagnosis of sex chromosome abnormality: a systematic review of the literature. Genet Med, 2012; 14 (1): 27–38.	Not relevant PICO: outcome
Jeon YJ,Zhou Y,Li Y,Guo Q,Chen J,Quan S, et al. The feasibility study of non-invasive fetal trisomy 18 and 21 detection with semiconductor sequencing platform. PLoS One, 2014; 9 (10): e110240.	Not relevant PICO: intervention
Jia Y,Zhao H,Shi D,Peng W,Xie L,Wang W, et al. Genetic effects of a 13q31.1 microdeletion detected by noninvasive prenatal testing (NIPT). Int J Clin Exp Pathol, 2014; 7 (10): 7003–11.	Not relevant study design: case study
Jiang F,Ren J,Chen F,Zhou Y,Xie J,Dan S, et al. Noninvasive Fetal Trisomy (NIFTY) test: an advanced noninvasive prenatal diagnosis methodology for fetal autosomal and sex chromosomal aneuploidies. BMC Med Genomics, 2012; 557.	Not relevant PICO: intervention
Jin Y,Miao Z,Ge J,Zhang W,Li S, Liu X. [Prenatal diagnosis of fetal chromosome aneuploidy by massively parallel genomic sequencing]. Zhonghua Yi Xue Za Zhi, 2014; 94 (23): 1788–90.	Language
Juneau K,Bogard PE,Huang S,Mohseni M,Wang ET,Ryvkin P, et al. Microarray-Based Cell-Free DNA Analysis Improves Noninvasive Prenatal Testing. Fetal Diagn Ther, 2014.	Not relevant PICO: outcome
Kagan KO,Hoopmann M,Hammer R,Stressig R, Kozlowski P. Screening for chromosomal abnormalities by first trimester combined screening and noninvasive prenatal testing. Ultraschall Med, 2015; 36 (1): 40–6.	Not relevant PICO: outcome
Kalynchuk EJ,Althouse A,Parker LS,Saller DN, Jr., Rajkovic A. Prenatal whole-exome sequencing: parental attitudes. Prenat Diagn, 2015.	Not relevant PICO: intervention
Kang JU, Koo SH. Clinical implementation of chromosomal microarray technology in prenatal diagnosis. (Review). Mol Med Rep, 2012; 6 (6): 1219–22.	Not relevant study design
Karampetsou E,Morrogh D,Ballard T,Waters JJ,Lench N, Chitty LS. Confined placental mosaicism: implications for fetal chromosomal analysis using microarray comparative genomic hybridization. Prenat Diagn, 2014; 34 (1): 98–101.	Not relevant study design: case study
Kashork CD,Theisen A, Shaffer LG. Prenatal diagnosis using array CGH. Methods Mol Biol, 2008; 44459–69.	Not relevant PICO: intervention

Kitzman JO,Snyder MW,Ventura M,Lewis AP,Qiu R,Simmons LE, et al. Noninvasive whole-genome sequencing of a human fetus. Sci Transl Med 2012; 4 (137): 137ra76.	Not relevant study design: case study
Korabecna M,Pazourkova E,Horinek A,Mokrejsova M, Tesar V. Alterations in methylation status of immune response genes promoters in cell-free DNA during a hemodialysis procedure. Expert Opin Biol Ther 12 Suppl 1S27–33.	Not relevant PICO: intervention
Korostelev S,Totchiev G,Kanivets I, Gnetetskaya V. Association of non-invasive prenatal testing and chromosomal microarray analysis for prenatal diagnostics. Gynecol Endocrinol, 2014; 30 Suppl 113–6.	Not relevant PICO: intervention
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Porreco RP,Garite TJ,Maurel K,Marusiak B,Ehrich M,van den Boom D, et al. Noninvasive prenatal screening for fetal trisomies 21, 18, 13 and the common sex chromosome aneuploidies from maternal blood using massively parallel genomic sequencing of DNA. Am J Obstet Gynecol, 2014; 211 (4): 365.e1–12.	Not relevant PICO: intervention

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Schulz-Hanke I. [Prenatal diagnosis - how powerful is DNA microarray compared to karyotyping?]. Z Geburtshilfe Neonatol, 2013; 217 (1): 3–4.	Not relevant study design
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. Clin Chem, 2011; 57 (7): 1042–9.	Not relevant PICO: intervention
Shaffer LG,Dabell MP,Rosenfeld JA,Neill NJ,Ballif BC,Coppinger J, et al. Referral patterns for microarray testing in prenatal diagnosis. Prenatal Diagnosis, 2012; 32 (4): 344–50.	Not relevant PICO: control and outcome unclear
Shaw SW,Hsiao CH,Chen CY,Ren Y,Tian F,Tsai C, et al. Noninvasive prenatal testing for whole fetal chromosomal aneuploidies: a multicenter prospective cohort trial in Taiwan. Fetal Diagn Ther, 2014; 35 (1): 13–7.	Not relevant PICO: intervention
Shen GS,Zhang S, He PY. [Application of gene array chip in diagnosis of cytogenetics of recurrent spontaneous abortion]. Zhonghua Fu Chan Ke Za Zhi 2013; 48 (7): 515–8.	Language
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Sillence KA,Madgett TE,Roberts LA,Overton TG,Avent ND,Author A, et al. Non-invasive screening tools for down's syndrome: A review. Diagnostics, 2013; 3 (2): 291–314.	Not relevant study design
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Talkowski ME,Ordulu Z,Pillalamarri V,Benson CB,Blumenthal I,Connolly S, et al. Clinical diagnosis by whole-genome sequencing of a prenatal sample. N Engl J Med, 2012; 367 (23): 2226–32.	Not relevant study design: case study
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Timor-Tritsch IE,Kapp S,Berg R,Bejjani BA,Adams SA,Monteagudo A, et al. Greig cephalopolysyndactyly syndrome: diagnosis based on prenatal sonographic features coupled with comparative genomic hybridization. J Ultrasound Med, 2009; 28 (12): 1735–42.	Not relevant study design: case report
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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.	Not relevant PICO: intervention
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Warren JE,Turok DK,Maxwell TM,Brothman AR, Silver RM. Array comparative genomic hybridization for genetic evaluation of fetal loss between 10 and 20 weeks of gestation. Obstet Gynecol, 2009; 114 (5): 1093–102.	Not relevant PICO: Population

Wegner RD,Stumm M, Hofmann W. Non-invasive prenatal diagnosis using massively parallel sequencing - first experience in Germany. Mol Cytogenet, 2014; 7 (Suppl 1 Proceedings of the International Conference on Human): I14.	Not relevant study design
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White HE,Dent CL,Hall VJ,Crolla JA,Chitty LS,Author A, et al. Evaluation of a Novel Assay for Detection of the Fetal Marker RASSF1A: Facilitating Improved Diagnostic Reliability of Noninvasive Prenatal Diagnosis. PLoS ONE, 2012; 7:9 Article Number: e45073.	Not relevant PICO: intervention
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Wilson RD,Wilson RD,Audibert F,Brock JA,Campagnolo C,Carroll J, et al. Prenatal screening, diagnosis, and pregnancy management of fetal neural tube defects. J Obstet Gynaecol Can, 2014; 36 (10): 927–39.	Not relevant PICO: intervention
Vlatkovic IB,Hafner T,Miskovic B,Vicic A,Poljak B, Stipoljev F. Prenatal diagnosis of sex chromosome aneuploidies and disorders of sex development - a retrospective analysis of 11-year data. Journal of Perinatal Medicine, 2014; 42 (4): 529–34.	Not relevant PICO: intervention
Voelkerding KV, Lyon E. Digital fetal aneuploidy diagnosis by next-generation sequencing. Clin Chem, 2010; 56 (3): 336–8.	Not relevant study design: editorial
Xie W,Tan Y,Li X,Lin G,Jiang H,Chen F, et al. Rapid detection of aneuploidies on a benchtop sequencing platform. Prenat Diagn 2013; 33 (3): 232–7.	Not relevant PICO: Population
Xu HB,Yang H,Liu G, Chen H. Systematic review of accuracy of prenatal diagnosis for abnormal chromosome diseases by microarray technology. Genet Mol Res, 2014; 13 (4): 9115–21.	Not relevant PICO: intervention.

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Yatsenko SA,Peters DG,Saller DN,Chu T,Clemens M, Rajkovic A. Maternal cell-free DNA-based screening for fetal microdeletion and the importance of careful diagnostic follow-up. Genet Med, 2015.	Not relevant study design: case report
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Yu SC,Jiang P,Choy KW,Chan KC,Won HS,Leung WC, et al. Noninvasive prenatal molecular karyotyping from maternal plasma. PLoS One, 2013; 8 (4): e60968.	Not relevant PICO: intervention
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Yuan Y,Jiang F,Hua S,Du B,Hao Y,Ye L, et al. Feasibility study of semiconductor sequencing for noninvasive prenatal detection of fetal aneuploidy. Clin Chem, 2013; 59 (5): 846–9.	Not relevant PICO: outcome

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Zilina O,Teek R,Tammur P,Kuuse K,Yakoreva M,Vaidla E, et al. Chromosomal microarray analysis as a first-tier clinical diagnostic test: Estonian experience. Mol Genet Genomic Med, 2014; 2 (2): 166–75.	Not relevant PICO
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Zwijnenburg PJG,Lakeman P,Pfundt R,Klein Wassink-Ruiter JS,Kerstjens-Frederikse WS,Van Ravenswaaij-Arts CMA, et al. Detection of submicroscopic chromosomal abnormalities using microarray analysis: The value and pitfalls in prenatal and postnatal diagnosis ORIGINAL (NON-ENGLISH) TITLE Detectie van submicroscopische chromosomale afwijkingen door middel van array-diagnostiek: De meerwaarde en de valkuilen in de prenatale en postnatale diagnostiek. Tijdschrift voor Kindergeneeskunde, 2014; 82 (1): 3–18.	Language

Systematic reviews	
Callaway JL,Shaffer LG,Chitty LS,Rosenfeld JA, Crolla JA. The clinical utility of microarray technologies applied to prenatal cytogenetics in the presence of a normal conventional karyotype: a review of the literature. Prenat Diagn, 2013; 33 (12): 1119–23.	Systematic review Differences in inclusion exclusion criteria: includes studies published before 2008 and studies were they uses custom made microarrays. Includes studies with a high risk of bias in the meta-analysis. High risk of bias: Not presenting a clear PICO. The full search is not presented. Only one database is used for the literature

	search. No quality assessment of the included studies. Pooling of data was done in spite of heterogeneity issues.
Grande M,Jansen FA,Blumenfeld YJ,Fisher A,Odibo AO,Haak MC, et al. Genomic microarray in fetuses with increased nuchal translucency and normal karyotype - a systematic review and meta-analysis. Ultrasound Obstet Gynecol, 2015.	Systematic review Differences in inclusion/exclusion criteria: includes studies were they used custom made microarrays. Uses a cutoff of >5Mb for aberrations to be considered detectable by the karyotype. Studies with fewerthan 15 cases were excluded from the meta-analysis. Includes studies with a high risk of bias in the meta-analysis. Moderate risk of bias: The quality assessment is not presented for each of the included studies separately.
Hillman SC,Pretlove S,Coomarasamy A,McMullan DJ,Davison EV,Maher ER, et al. Additional information from array comparative genomic hybridization technology over conventional karyotyping in prenatal diagnosis: a systematic review and meta-analysis (Provisional abstract). Ultrasound in Obstetrics and Gynecology, 2011; 37 (1): 6–14.	Systematic review Differences in inclusion exclusion criteria: includes studies published before 2008 and studies were they uses custom made microarrays. Combines pathogenic CNV:s and VOUS in the meta-analysis. Includes studies with a high risk of bias in the meta-analysis. High risk of bias: The full search is not presented. The quality assessment is not presented for each of the included studies separately. Pooling of data was done in spite of heterogeneity issues.

Hillman SC, McMullan DJ, Hall G, Togneri FS, James N, Maher EJ, et al. Use of prenatal chromosomal microarray: prospective cohort study and systematic review and meta-analysis. Ultrasound Obstet Gynecol 2013;41:610–20.	Differences in inclusion exclusion criteria: includes studies published before 2008 and studies were they uses custom made microarrays. Includes studies with a high risk of bias in the meta-analysis. High risk of bias: The full search is not presented. The quality assessment is not presented for each of the included studies separately. Pooling of data was done in spite of heterogeneity issues.
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Saldarriaga W,Garcia-Perdomo HA,Arango-Pineda J, Fonseca J. Karyotype versus genomic hybridization for the prenatal diagnosis of chromosomal abnormalities: a metaanalysis. Am J Obstet Gynecol, 2014.	Systematic review Differences in inclusion exclusion criteria: Uses CNV:s identified with microarray plus abnormal karyotype as the true positives. Includes studies were they uses custom made microarrays or were the microarray platform was not specified. Includes studies with a high risk of bias in the meta-analysis. High risk of bias: Pooling of data was done in spite of heterogeneity issues.
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	algorithm used for the pooling of the data is not presented, nor the weight of the different studies.

Studies with high risk of bias/ Studier med låg kvalitet
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