Appendix 3 Excludes studies and studies with high risk of bias/Bilaga 3 exkluderade studier och studier av låg kvalitet

Excluded studies	
The Observatory. Genetic Testing. MLO: Medical Laboratory Observer, 2013; 45 (1): 6-6.	Not relevant study design: comment
Ahn JW,Bint S,Irving MD,Kyle PM,Akolekar R,Mohammed SN, et al. A new direction for prenatal chromosome microarray testing: software-targeting for detection of clinically significant chromosome imbalance without equivocal findings. PeerJ, 2014; 2e354.	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.	Not relevant PICO: intervention
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: 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. Eur J Hum Genet, 2015.	Not relevant PICO: outcome
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
Bernhardt BA,Soucier D,Hanson K,Savage MS,Jackson L, Wapner RJ. Women's experiences receiving abnormal prenatal chromosomal microarray testing results. Genet Med, 2013; 15 (2): 139-45.	Not relevant PICO: intervention
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. Prenat Diagn2012; 32 (1): 10-20.	Not relevant PICO: population
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
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 PICO: intervention
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,DeKoninck P,Fryns JP,Devriendt K,Deprest JA, Vermeesch JR. Identification of dosage-sensitive genes in fetuses referred with severe isolated congenital diaphragmatic hernia. Prenat Diagn, 2013; 33 (13): 1283-92.	Not relevant PICO: intervention
Brady PD,Delle Chiaie B,Christenhusz G,Dierickx K,Van Den Bogaert K,Menten B, et al. A prospective study of the clinical utility of prenatal chromosomal microarray analysis in fetuses with ultrasound abnormalities and an exploration of a framework for reporting unclassified variants and risk factors. Genet Med, 2014; 16 (6): 469-76.	Not relevant PICO: intervention
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
Breman A,Pursley AN,Hixson P,Bi W,Ward P,Bacino CA, et al. Prenatal chromosomal microarray analysis in a diagnostic laboratory; experience with >1000 cases and review of the literature. Prenat Diagn, 2012; 32 (4): 351-61.	Not relevant PICO: intervention
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

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.	Not relevant PICO: intervention
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
Carey L,Scott F,Murphy K,Mansfield N,Barahona P,Leigh D, et al. Prenatal diagnosis of chromosomal mosaicism in over 1600 cases using array comparative genomic hybridization as a first line test. Prenat Diagn, 2014; 34 (5): 478-86.	Not relevant PICO: intervention
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: population
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
Charan P,Woodrow N,Walker SP,Ganesamoorthy D,McGillivray G, Palma-Dias R. High-resolution microarray in the assessment of fetal anomalies detected by ultrasound. Aust N Z J Obstet Gynaecol, 2014; 54 (1): 46-52.	Not relevant PICO: intervention

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	Not relevant PICO intervention

hybridization. Ultrasound Obstet Gynecol 2014; 43 (4): 396-403.	
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 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	Not relevant PICO: outcome

microarrays for invasive fetal testing. Ultrasound Obstet Gynecol, 2015; 45 (4): 394-401.	
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.	Not relevant PICO: intervention
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
D'Amours G,Kibar Z,Mathonnet G,Fetni R,Tihy F,Desilets V, et al. Whole-genome array CGH identifies pathogenic copy number variations in fetuses with major malformations and a normal karyotype. Clin Genet, 2012; 81 (2): 128-41.	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
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 PICO: intervention
Davies N. Prenatal screening with microarray technology. MLO: Medical Laboratory Observer, 2015; 47 (5): 8-12.	Not relevant PICO: intervention

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
Donley G,Hull SC, Berkman BE. Prenatal whole genome sequencing: just because we can, should we? Hastings Cent Rep 2012; 42 (4): 28-40.	Not relevant study design
Donnelly JC,Platt LD,Rebarber A,Zachary J,Grobman WA, Wapner RJ. Association of copy number variants with specific ultrasonographically detected fetal anomalies. Obstet Gynecol, 2014; 124 (1): 83-90.	Not relevant PICO: intervention
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
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: population
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 PICO: intervention
Dupont C,Grati FR,Choy KW,Jaillard S,Toutain J,Maurin ML, et al. Prenatal diagnosis of 24 cases of microduplication 22q11.2: an investigation of phenotype-genotype correlations. Prenat Diagn 2014.	Not relevant PICO: Intervention
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
Evangelidou P,Alexandrou A,Moutafi M,Ioannides M,Antoniou P,Koumbaris G, et al. Implementation of high resolution whole genome array CGH in the prenatal clinical setting: advantages, challenges, and review of the literature. Biomed Res Int, 2013; 2013346762.	Not relevant PICO: intervention
Evangelidou P,Sismani C,Ioannides M,Christodoulou C,Koumbaris G,Kallikas I, et al. Clinical application of whole-genome array CGH during prenatal diagnosis: Study of 25 selected pregnancies with abnormal ultrasound findings or apparently balanced structural aberrations. Mol Cytogenet, 2010; 324.	Not relevant PICO: intervention
Faas BH,Feenstra I,Eggink AJ,Kooper AJ,Pfundt R,van Vugt JM, et al. Non-targeted whole genome 250K SNP array analysis as replacement for karyotyping in fetuses with structural ultrasound anomalies: evaluation of a one-year experience. Prenat Diagn, 2012; 32 (4): 362-70.	Not relevant PICO: intervention
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, 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: Outcome
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 PICO: intervention
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
Filges I,Kang A,Klug V,Wenzel F,Heinimann K,Tercanli S, et al. Array comparative genomic hybridization in prenatal diagnosis of first trimester pregnancies at high risk for chromosomal anomalies. Mol Cytogenet, 2012; 5 (1): 38.	Not relevant PICO: intervention
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: population
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.	Not relevant PICO: intervention

Fiorentino F,Napoletano S,Caiazzo F,Sessa M,Bono S,Spizzichino L, et al. Chromosomal microarray analysis as a first-line test in pregnancies with a priori low risk for the detection of submicroscopic chromosomal abnormalities. Eur J Hum Genet, 2013; 21 (7): 725-30.	Not relevant PICO: intervention
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 PICO: population
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
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.	Targeted prenatal genetic testing for a specific aberration
Ganesamoorthy D,Bruno DL,McGillivray G,Norris F,White SM,Adroub S, et al. Meeting the challenge of interpreting high-resolution single nucleotide polymorphism array data in prenatal diagnosis: does increased diagnostic power outweigh the dilemma of rare variants? Bjog, 2013; 120 (5): 594-606.	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
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	Not relevant study design

sampling. The so called "Next Generation Prenatal Diagnosis" (NGPD). J Prenat Med, 2014; 8 (1-2): 1-10.	
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.	Not relevant PICO: intervention
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,Decamp M,Richard N,Jeanne-Pasquier C,Benoist G,Mittre H, et al. Array CGH analysis in high-risk pregnancies: comparing DNA from cultured cells and cell-free fetal DNA. Prenat Diagn, 2012; 32 (4): 383-8.	Not relevant PICO: intervention
Gruchy N,Lebrun M,Herlicoviez M,Alliet J,Gourdier D,Kottler ML, et al. Supernumerary marker chromosomes management in prenatal diagnosis. Am J Med Genet A, 2008; 146a (21): 2770-6.	Not relevant PICO: intervention
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: 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: intervention
Held KR,Zahn S,Author A,Mvz genteQ GmbH Lf,Humangenetik FHG,Correspondence A, et al. Microarray-based comparative genomic hybridization for prenatal diagnosis: Indications and clinical evaluation ORIGINAL (NON-ENGLISH) TITLE Pranataler Array: Indikationen, Bewertung. Medizinische Genetik, 2014; 26 (4): 398-404.	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: population
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,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 (6): 610-20.	Not relevant PICO: intervention
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 outweigh the dilemma of rare variants. Bjog 2013; 120 (10): 1296.	Not relevant study design
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.	Not relevant PICO: intervention
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
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.	Not relevant PICO: intervention
Hillman SC,Skelton J,Quinlan-Jones E,Wilson A, Kilby MD. "If it helps..." the use of microarray technology in prenatal testing: patient and partners reflections. Am J Med Genet A, 2013; 161a (7): 1619-27.	Not relevant PICO: intervention
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: population

Huang J,Poon LC,Akolekar R,Choy KW,Leung TY, Nicolaides KH. Is high fetal nuchal translucency associated with submicroscopic chromosomal abnormalities on array CGH? Ultrasound in Obstetrics & Gynecology, 2014; 43 (6): 620-24.	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
Jansen FA,Blumenfeld YJ,Fisher A,Cobben JM,Odibo AO,Borrell A, et al. Array Comparative Genomic Hybridization and Fetal Congenital Heart Defects - A systematic review and meta-analysis. Ultrasound Obstet Gynecol, 2014.	Not relevant PICO: intervention
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
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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
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Macera MJ,Sobrino A,Levy B,Jobanputra V,Aggarwal V,Mills A, et al. Prenatal diagnosis of chromothripsis, with nine breaks characterized by karyotyping, FISH, microarray and whole-genome sequencing. Prenat Diagn, 2015; 35 (3): 299-301.	Not relevant study design: case report
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Pooh RK. Sonogenetics in fetal neurology. Semin Fetal Neonatal Med 2012; 17 (6): 353-9.	Not relevant PICO: intervention
Rabinowitz M,Valenti E,Pettersen B,Sigurjonsson S,Hill M, Zimmermann B. Noninvasive aneuploidy detection by multiplexed amplification and sequencing of polymorphic Loci. Obstet Gynecol, 2014; 123 Suppl 1167s.	Not relevant study design
Rad S,Turner AL,Ratousi D,Aghajanian P,Williams J, 3rd, Esakoff TF. Predictors for choosing array-comparative genomic hybridization for prenatal diagnosis. Obstet Gynecol 2014; 123 Suppl 1100s-1s.	Not relevant study design: conference abstract
Rajcan-Separovic E. Chromosome microarrays in human reproduction. Hum Reprod Update, 2012; 18 (5): 555-67.	Not relevant study design
Rava RP,Srinivasan A,Sehnert AJ, Bianchi DW. Circulating fetal cell-free DNA fractions differ in autosomal aneuploidies and monosomy X. Clin Chem, 2014; 60 (1): 243-50.	Not relevant PICO: outcome
Regier DA,Peacock SJ,Pataky R,van der Hoek K,Jarvik GP,Hoch J, et al. Societal preferences for the return of incidental findings from clinical genomic sequencing: a discrete-choice experiment. CMAJ: Canadian Medical Association Journal, 2015; 187 (6): E190-7.	Not relevant study design
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Rieneck K,Bak M,Jonson L,Clausen FB,Krog GR,Tommerup N, et al. Next-generation sequencing: proof of concept for antenatal prediction of the fetal Kell blood group phenotype from cell-free fetal DNA in maternal plasma. Transfusion, 2013; 53 (11 Suppl 2): 2892-8.	Not relevant PICO: outcome
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Rooryck C,Toutain J,Cailley D,Bouron J,Horovitz J,Lacombe D, et al. Prenatal diagnosis using array-CGH: a French experience. Eur J Med Genet, 2013; 56 (7): 341-5.	Not relevant PICO: intervention
Rosen JM. Genetic diagnosis through whole-exome sequencing... N Engl J Med. 2013 Oct 17;369(16):1557-8. New England Journal of Medicine, 2014; 370 (11): 1068-68.	Not relevant study design : editorial
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Sago H, Sekizawa A. Nationwide demonstration project of next-generation sequencing of cell-free DNA in maternal plasma in Japan: one-year experience. Prenat Diagn, 2014.	Not relevant PICO: outcome
Sago H, Sekizawa A. Nationwide demonstration project of next-generation sequencing of cell-free DNA in maternal plasma in Japan: 1-year experience. Prenat Diagn, 2015; 35 (4): 331-6.	Not relevant PICO: outcome
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.	Not relevant PICO: Intervention
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Schmid M,Stary S,Springer S,Bettelheim D,Husslein P, Streubel B. Prenatal microarray analysis as second-tier diagnostic test: single-center prospective study. Ultrasound Obstet Gynecol, 2013; 41 (3): 267-73.	Not relevant PICO: intervention
Schou KV,Kirchhoff M,Nygaard U,Jorgensen C, Sundberg K. Increased nuchal translucency with normal karyotype: a follow-up study of 100 cases supplemented with CGH and MLPA analyses. Ultrasound Obstet Gynecol, 2009; 34 (6): 618-22.	Not relevant PICO: intervention
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Scott F,Murphy K,Carey L,Greville W,Mansfield N,Barahona P, et al. Prenatal diagnosis using combined quantitative fluorescent polymerase chain reaction and array comparative genomic hybridization analysis as a first-line test: results from over 1000 consecutive cases. Ultrasound Obstet Gynecol, 2013; 41 (5): 500-7.	Not relevant PICO: intervention

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Shen GS,Zhang S, He PY. [Application of gene array chip in diagnosis of cytogenetics of recurrent spontaneous abortion]. Marias beställning, 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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Srebniak MI,Boter M,Oudesluijs GO,Cohen-Overbeek T,Govaerts LC,Diderich KE, et al. Genomic SNP array as a gold standard for prenatal diagnosis of foetal ultrasound abnormalities. Mol Cytogenet, 2012; 5 (1): 14.	Not relevant PICO: intervention
Srebniak MI,Mout L,Van Opstal D, Galjaard RJ. 0.5 Mb array as a first-line prenatal cytogenetic test in cases without ultrasound abnormalities and its implementation in clinical practice. Hum Mutat, 2013; 34 (9): 1298-303.	Not relevant PICO: Intervention
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Srinivasan A,Bianchi DW,Huang H,Sehnert AJ, Rava RP. Noninvasive detection of fetal subchromosome abnormalities via deep sequencing of maternal plasma. Am J Hum Genet, 2013; 92 (2): 167-76.	Not relevant PICO: population
Sun L,Wu Q,Jiang SW,Yan Y,Wang X,Zhang J, et al. Prenatal Diagnosis of Central Nervous System Anomalies by High-Resolution Chromosomal Microarray Analysis. Biomed Res Int, 2015; 2015426379.	Not relevant PICO: intervention
Sun LH,Li L,Wang XW,Zhu YZ,Chai YC,Li XH, et al. [Prenatal screening and diagnosis of genetic deafness by microarray]. Marias beställning, 2012; 47 (12): 991-5.	Language
Tabor HK,Murray JC,Gammill HS,Kitzman JO,Snyder MW,Ventura M, et al. Non-invasive fetal genome sequencing: opportunities and challenges. Marias beställning, 2012; 158a (10): 2382-4.	Not relevant study design

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