

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

Excluded studies, page 1–35

Systematic review 35

Studies with high risk of bias, page 36–38

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

<p>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.</p>	<p>Not relevant PICO: outcome</p>
<p>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. <i>Obstet Gynecol</i>, 2015; 125 (6): 1330-7.</p>	<p>Not relevant PICO: outcome</p>
<p>Bernhardt BA, Soucier D, Hanson K, Savage MS, Jackson L, Wapner RJ. Women's experiences receiving abnormal prenatal chromosomal microarray testing results. <i>Genet Med</i>, 2013; 15 (2): 139-45.</p>	<p>Not relevant PICO: intervention</p>
<p>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. <i>Prenat Diagn</i> 2012; 32 (1): 10-20.</p>	<p>Not relevant PICO: population</p>
<p>Bianchi DW, Wilkins-Haug L. Integration of noninvasive DNA testing for aneuploidy into prenatal care: what has happened since the rubber met the road? <i>Clin Chem</i>, 2014; 60 (1): 78-87.</p>	<p>Not relevant study design</p>
<p>Biesecker LG, Green RC. Diagnostic clinical genome and exome sequencing. <i>New England Journal of Medicine</i>, 2014; 370 (25): 2418-25.</p>	<p>Not relevant study design: comment</p>
<p>Blackburn HL, Schroeder B, Turner C, Shriver CD, Ellsworth DL, Ellsworth RE. Management of Incidental Findings in the Era of Next-generation Sequencing. <i>Curr Genomics</i>, 2015; 16 (3): 159-74.</p>	<p>Not relevant PICO: intervention</p>
<p>Bouffard C, Viville S, Knoppers BM. Genetic diagnosis of embryos: clear explanation, not rhetoric, is needed. <i>CMAJ: Canadian Medical Association Journal</i>, 2009; 181 (6-7): 387-91.</p>	<p>Not relevant study design</p>
<p>Bovolenta M, Rimessi P, Dolcini B, Ravani A, Ferlini A, Gualandi F. Prenatal diagnosis of Duchenne muscular dystrophy by comparative genomic hybridization. <i>Clin Genet</i>, 2010; 77 (5): 503-6.</p>	<p>Not relevant study design: case report</p>

<p>Brady PD,DeKoninck P,Fryns JP,Devriendt K,Depest JA, Vermeesch JR. Identification of dosage-sensitive genes in fetuses referred with severe isolated congenital diaphragmatic hernia. <i>Prenat Diagn</i>, 2013; 33 (13): 1283-92.</p>	<p>Not relevant PICO: intervention</p>
<p>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. <i>Genet Med</i>, 2014; 16 (6): 469-76.</p>	<p>Not relevant PICO: intervention</p>
<p>Brady PD,Devriendt K,Depest J, Vermeesch JR. Array-based approaches in prenatal diagnosis. <i>Methods Mol Biol</i> 2012; 838:151-71.</p>	<p>Not relevant study design</p>
<p>Brady PD, Vermeesch JR. Genomic microarrays: a technology overview. <i>Prenat Diagn</i>, 2012; 32 (4): 336-43.</p>	<p>Not relevant study design</p>
<p>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. <i>Prenat Diagn</i>, 2012; 32 (4): 351-61.</p>	<p>Not relevant PICO: intervention</p>
<p>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. <i>Archives of Disease in Childhood: Fetal and Neonatal Edition</i>, 2014; 99 (4): F286-F90.</p>	<p>Not relevant PICO: intervention</p>
<p>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; 504 (1): 144-9.</p>	<p>Not relevant study design</p>
<p>Callaway JL,Huang S,Karampetsou E, Crolla JA. Perspective on the technical challenges involved in the implementation of array-CGH in prenatal diagnostic testing. <i>Mol Biotechnol</i> 2014; 56 (4): 312-8.</p>	<p>Not relevant PICO: outcome</p>

<p>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. <i>Prenat Diagn</i>, 2013; 33 (12): 1119-23.</p>	<p>Not relevant PICO: intervention</p>
<p>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. <i>Eur J Endocrinol</i> 2014; 171 (2): 253-62.</p>	<p>Not relevant PICO: population</p>
<p>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. <i>Prenat Diagn</i>, 2014; 34 (5): 478-86.</p>	<p>Not relevant PICO: intervention</p>
<p>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. <i>Hum Mol Genet</i>, 2014; 23 (12): 3269-77.</p>	<p>Not relevant PICO: population</p>
<p>Cavalli P,Cavallari U, Novelli A. Array CGH in routine prenatal diagnosis practice. <i>Prenat Diagn</i> 2012; 32 (7): 708-9; author reply 11-2.</p>	<p>Not relevant study design: letter</p>
<p>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. <i>Eur J Obstet Gynecol Reprod Biol</i>, 2010; 150 (1): 28-33.</p>	<p>Targeted prenatal genetic testing for a specific aberration</p>
<p>Char D. Preventive Genomic Sequencing and Care of the Individual Patient. <i>American Journal of Bioethics</i>, 2015; 15 (7): 32-3.</p>	<p>Not relevant study design</p>
<p>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. <i>Aust N Z J Obstet Gynaecol</i>, 2014; 54 (1): 46-52.</p>	<p>Not relevant PICO: intervention</p>

<p>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.</p>	<p>Not relevant study design: case study</p>
<p>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.</p>	<p>Not relevant study design: case report</p>
<p>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.</p>	<p>Not relevant study design: case study</p>
<p>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.</p>	<p>Not relevant study design: case report</p>
<p>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.</p>	<p>Not relevant study design: case study</p>
<p>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.</p>	<p>Not relevant study design: case study</p>
<p>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.</p>	<p>Not relevant PICO: intervention</p>
<p>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</p>	<p>Not relevant PICO intervention</p>

hybridization. <i>Ultrasound Obstet Gynecol</i> 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. <i>Genome Med</i> , 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]. <i>Zhonghua Yan Ke Za Zhi</i> 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. <i>Prenat Diagn</i> , 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. <i>Proc Natl Acad Sci U S A</i> , 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. <i>Bjog</i> 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. <i>Fetal and Maternal Medicine Review</i> , 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

<p>microarrays for invasive fetal testing. <i>Ultrasound Obstet Gynecol</i>, 2015; 45 (4): 394-401.</p>	
<p>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. <i>Prenat Diagn</i>, 2009; 29 (12): 1156-66.</p>	<p>Not relevant PICO: intervention</p>
<p>Coromilas A, Wynn J, Haverfield E, Chung WK. Nonspecific phenotype of Noonan syndrome diagnosed by whole exome sequencing. <i>Clin Case Rep</i>, 2015; 3 (4): 237-9.</p>	<p>Not relevant study design: case report</p>
<p>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. <i>Am J Psychiatry</i>, 2013; 170 (12): 1498.</p>	<p>Not relevant study design</p>
<p>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. <i>Clin Genet</i>, 2012; 81 (2): 128-41.</p>	<p>Not relevant PICO: intervention</p>
<p>D'Angelo G, Di Rienzo T, Ojetti V. Microarray analysis in gastric cancer: a review. <i>World J Gastroenterol</i> 2014; 20 (34): 11972-6.</p>	<p>Not relevant PICO: population</p>
<p>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. <i>Genet Med</i>, 2008; 10 (1): 13-8.</p>	<p>Not relevant PICO: intervention</p>
<p>Davies N. Prenatal screening with microarray technology. <i>MLO: Medical Laboratory Observer</i>, 2015; 47 (5): 8-12.</p>	<p>Not relevant PICO: intervention</p>

<p>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. <i>J Community Genet</i>, 2013; 4 (1): 125-35.</p>	<p>Not relevant PICO: population</p>
<p>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. <i>Genet Test Mol Biomarkers</i>, 2010; 14 (3): 421-4.</p>	<p>Not relevant PICO: population</p>
<p>Dickinson JE, Harcourt E, Murch A. The selective use of rapid aneuploidy screening in prenatal diagnosis. <i>Australian & New Zealand Journal of Obstetrics & Gynaecology</i>, 2009; 49 (1): 28-33.</p>	<p>Not relevant PICO: intervention</p>
<p>Dondorp WJ, Page-Christiaens GC, de Wert GM. Genomic futures of prenatal screening: ethical reflection. <i>Clin Genet</i>, 2015.</p>	<p>Not relevant study design</p>
<p>Donley G, Hull SC, Berkman BE. Prenatal whole genome sequencing: just because we can, should we? <i>Hastings Cent Rep</i> 2012; 42 (4): 28-40.</p>	<p>Not relevant study design</p>
<p>Donnelly JC, Platt LD, Rebarber A, Zachary J, Grobman WA, Wapner RJ. Association of copy number variants with specific ultrasonographically detected fetal anomalies. <i>Obstet Gynecol</i>, 2014; 124 (1): 83-90.</p>	<p>Not relevant PICO: intervention</p>
<p>Donnelly JC, Wapner RJ, Simpson JL, Author A, Department o, Obstetrics, et al. Can microarray analysis reveal additional abnormalities in prenatal diagnosis? <i>Expert Review of Obstetrics and Gynecology</i>, 2013; 8 (3): 193-95.</p>	<p>Not relevant study design</p>
<p>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. <i>J Prenat Med</i>, 2014; 8 (1-2): 31-5.</p>	<p>Not relevant PICO: population</p>
<p>Dugoff L. Application of genomic technology in prenatal diagnosis. <i>N Engl J Med</i> 2012; 367 (23): 2249-51.</p>	<p>Not relevant study design : editorial</p>

<p>Duncan A, Langlois S. Use of array genomic hybridization technology in prenatal diagnosis in Canada. <i>J Obstet Gynaecol Can</i>, 2011; 33 (12): 1256-9.</p>	<p>Not relevant PICO: intervention</p>
<p>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. <i>Prenat Diagn</i> 2014.</p>	<p>Not relevant PICO: Intervention</p>
<p>Eng CM, Yang Y, Plon SE. Genetic diagnosis through whole-exome sequencing... <i>N Engl J Med</i>. 2014 Mar 13;370(11):1067-8; <i>N Engl J Med</i>. 2013 Oct 17;369(16):1502-11; <i>N Engl J Med</i>. 2014 Mar 13;370(11):1067. <i>New England Journal of Medicine</i>, 2014; 370 (11): 1068-68.</p>	<p>Not relevant study design: letter</p>
<p>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. <i>Biomed Res Int</i>, 2013; 2013346762.</p>	<p>Not relevant PICO: intervention</p>
<p>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. <i>Mol Cytogenet</i>, 2010; 324.</p>	<p>Not relevant PICO: intervention</p>
<p>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. <i>Prenat Diagn</i>, 2012; 32 (4): 362-70.</p>	<p>Not relevant PICO: intervention</p>
<p>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. <i>J Med Genet</i>, 2010; 47 (9): 586-94.</p>	<p>Not relevant PICO: population</p>

<p>Fan HC, Quake SR. Sensitivity of noninvasive prenatal detection of fetal aneuploidy from maternal plasma using shotgun sequencing is limited only by counting statistics. <i>PLoS One</i>, 2010; 5 (5): e10439.</p>	<p>Not relevant PICO: Outcome</p>
<p>Faucett A, Savage M. Chromosomal microarray testing. <i>JAAPA: Journal of the American Academy of Physician Assistants</i> (Haymarket Media, Inc.) 2012; 25 (1): 65-66.</p>	<p>Not relevant PICO: intervention</p>
<p>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. <i>J Ultrasound Med</i>, 2011; 30 (7): 1021-9.</p>	<p>Not relevant study design : case series</p>
<p>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. <i>Mol Cytogenet</i>, 2012; 5 (1): 38.</p>	<p>Not relevant PICO: intervention</p>
<p>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. <i>Ultrasound obstet gynecol</i> 2013; 41: 610-620. <i>Ultrasound Obstet Gynecol</i>, 2013; 41 (6): 608.</p>	<p>Not relevant study design: comment</p>
<p>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. <i>Fertil Steril</i>, 2014; 101 (5): 1375-82.</p>	<p>Not relevant PICO: population</p>
<p>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. <i>Prenat Diagn</i>, 2011; 31 (13): 1270-82.</p>	<p>Not relevant PICO: intervention</p>

<p>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. <i>Eur J Hum Genet</i>, 2013; 21 (7): 725-30.</p>	<p>Not relevant PICO: intervention</p>
<p>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. <i>Preventive Medicine</i>, 2012; 55 (5): 514-20.</p>	<p>Not relevant PICO: population</p>
<p>Foglieni B, Galbiati S, Ferrari M, Cremonesi L. A fast microelectronic array for screening and prenatal diagnosis of beta-thalassemia. <i>Methods Mol Biol</i>, 2008; 444:169-82.</p>	<p>Targeted prenatal genetic testing for a specific aberration</p>
<p>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. <i>Expert Opin Biol Ther</i> 2012; 12 Suppl 1S181-7.</p>	<p>Targeted prenatal genetic testing for a specific aberration</p>
<p>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? <i>Bjog</i>, 2013; 120 (5): 594-606.</p>	<p>Not relevant PICO: intervention</p>
<p>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. <i>Biomed Res Int</i> 2014; 2014590298.</p>	<p>Not relevant PICO: intervention</p>
<p>Gekas J, Sergi C, Kamnasaran D. Molecular prenatal diagnosis of a sporadic alobar holoprosencephalic fetus: genotype-phenotype correlations. <i>J Prenat Med</i>, 2012; 6 (3): 36-9.</p>	<p>Not relevant study design: case study</p>
<p>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 villous</p>	<p>Not relevant study design</p>

<p>sampling. The so called "Next Generation Prenatal Diagnosis" (NGPD). <i>J Prenat Med</i>, 2014; 8 (1-2): 1-10.</p>	
<p>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. <i>Ultrasound Obstet Gynecol</i>, 2015.</p>	<p>Not relevant PICO: intervention</p>
<p>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. <i>Am J Obstet Gynecol</i>, 2014; 211 (6): 711-12.</p>	<p>Not relevant study design</p>
<p>Grati FR,Vialard F, Gross S. BACs-on-Beads (BoBs) assay for the genetic evaluation of prenatal samples and products of conception. <i>Methods Mol Biol</i> 2015; 1227259-78.</p>	<p>Not relevant PICO: intervention</p>
<p>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. <i>Prenat Diagn</i>, 2012; 32 (4): 383-8.</p>	<p>Not relevant PICO: intervention</p>
<p>Gruchy N,Lebrun M,Herlicoviez M,Alliet J,Gourdier D,Kottler ML, et al. Supernumerary marker chromosomes management in prenatal diagnosis. <i>Am J Med Genet A</i>, 2008; 146a (21): 2770-6.</p>	<p>Not relevant PICO: intervention</p>
<p>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. <i>J Neurochem</i>, 2012; 122 (3): 641-9.</p>	<p>Not relevant PICO: intervention</p>
<p>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. <i>Ultrasound Obstet Gynecol</i>, 2013; 41 (1): 95-6.</p>	<p>Not relevant PICO: intervention</p>

<p>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.</p>	<p>Not relevant PICO: intervention</p>
<p>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.</p>	<p>Not relevant PICO: intervention</p>
<p>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.</p>	<p>Not relevant PICO: population</p>
<p>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.</p>	<p>Not relevant study design : editorial</p>
<p>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.</p>	<p>Not relevant PICO: outcome</p>
<p>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.</p>	<p>Not relevant PICO: intervention</p>
<p>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.</p>	<p>Not relevant study design: letter</p>

<p>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. <i>Bjog</i> 2013; 120 (10): 1296.</p>	<p>Not relevant study design</p>
<p>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? <i>J Matern Fetal Neonatal Med</i>, 2014; 27 (7): 649-57.</p>	<p>Not relevant PICO: intervention</p>
<p>Hillman SC,McMullan DJ,Williams D, Maher ER, Kilby MD. Microarray comparative genomic hybridization in prenatal diagnosis: a review. <i>Ultrasound Obstet Gynecol</i> 2012; 40 (4): 385-91.</p>	<p>Not relevant study design</p>
<p>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). <i>Ultrasound in Obstetrics and Gynecology</i>, 2011; 37 (1): 6-14.</p>	<p>Not relevant PICO: intervention</p>
<p>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. <i>Am J Med Genet A</i>, 2013; 161a (7): 1619-27.</p>	<p>Not relevant PICO: intervention</p>
<p>Holm S, Ploug T. Patient Choice and Preventive Genomic Sequencing—More Trouble Upstream. <i>American Journal of Bioethics</i>, 2015; 15 (7): 24-6.</p>	<p>Not relevant study design</p>
<p>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. <i>Prenat Diagn</i>, 2014; 34 (5): 496-9.</p>	<p>Not relevant PICO: intervention</p>
<p>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. <i>Prenat Diagn</i>, 2014.</p>	<p>Not relevant PICO: population</p>

<p>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? <i>Ultrasound in Obstetrics & Gynecology</i>, 2014; 43 (6): 620-24.</p>	<p>Not relevant PICO: intervention</p>
<p>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. <i>Ultrasound Obstet Gynecol</i> 2014; 43: 254-264. <i>Ultrasound Obstet Gynecol</i>, 2014; 43 (3): 245.</p>	<p>Not relevant study design</p>
<p>Jacob HJ. Next-generation sequencing for clinical diagnostics. <i>New England Journal of Medicine</i>, 2013; 369 (16): 1557-58.</p>	<p>Not relevant study design: comment</p>
<p>Jacob HJ. Genetic diagnosis through whole-exome sequencing... <i>N Engl J Med</i>. 2013 Oct 17;369(16):1557-8; <i>N Engl J Med</i>. 2014 Mar 13;370(11):1068. <i>New England Journal of Medicine</i>, 2014; 370 (11): 1069-69.</p>	<p>Not relevant study design: comment</p>
<p>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. <i>Ultrasound Obstet Gynecol</i>, 2014.</p>	<p>Not relevant PICO: intervention</p>
<p>Jensen TJ,Kim SK,van den Boom D,Deciu C, Ehrich M. Noninvasive detection of a balanced fetal translocation from maternal plasma. <i>Clin Chem</i>, 2014; 60 (10): 1298-305.</p>	<p>Not relevant study design: case study</p>
<p>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. <i>PLoS One</i>, 2013; 8 (3): e57381.</p>	<p>Not relevant PICO: intervention</p>
<p>Jeon KC,Chen LS, Goodson P. Decision to abort after a prenatal diagnosis of sex chromosome abnormality:</p>	<p>Not relevant study design</p>

a systematic review of the literature. <i>Genet Med</i> , 2012; 14 (1): 27-38.	
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. <i>PLoS One</i> , 2014; 9 (10): e110240.	Not relevant PICO: outcome
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). <i>Int J Clin Exp Pathol</i> , 2014; 7 (10): 7003-11.	Not relevant study design: case study
Jin Y,Miao Z,Ge J,Zhang W,Li S, Liu X. [Prenatal diagnosis of fetal chromosome aneuploidy by massively parallel genomic sequencing]. <i>Zhonghua Yi Xue Za Zhi</i> , 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. <i>Fetal Diagn Ther</i> , 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. <i>Ultraschall Med</i> , 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. <i>Prenat Diagn</i> , 2015.	Not relevant PICO: intervention
Kan AS,Lau ET,Tang WF,Chan SS,Ding SC,Chan KY, et al. Whole-genome array CGH evaluation for replacing prenatal karyotyping in Hong Kong. <i>PLoS One</i> , 2014; 9 (2): e87988.	Not relevant PICO: intervention
Kang JU, Koo SH. Clinical implementation of chromosomal microarray technology in prenatal diagnosis. (Review). <i>Mol Med Rep</i> , 2012; 6 (6): 1219-22.	Not relevant study design

<p>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. <i>Prenat Diagn</i>, 2014; 34 (1): 98-101.</p>	<p>Not relevant study design: case study</p>
<p>Kashork CD, Theisen A, Shaffer LG. Prenatal diagnosis using array CGH. <i>Methods Mol Biol</i>, 2008; 444:59-69.</p>	<p>Not relevant PICO: intervention</p>
<p>Kitzman JO, Snyder MW, Ventura M, Lewis AP, Qiu R, Simmons LE, et al. Noninvasive whole-genome sequencing of a human fetus. <i>Sci Transl Med</i> 2012; 4 (137): 137ra76.</p>	<p>Not relevant study design: case study</p>
<p>Kleeman L, Bianchi DW, Shaffer LG, Rorem E, Cowan J, Craigo SD, et al. Use of array comparative genomic hybridization for prenatal diagnosis of fetuses with sonographic anomalies and normal metaphase karyotype. <i>Prenat Diagn</i>, 2009; 29 (13): 1213-7.</p>	<p>Not relevant PICO: intervention</p>
<p>Klugman S, Suskin B, Spencer BL, Dar P, Bajaj K, Powers J, et al. Clinical utility of chromosomal microarray analysis in prenatal diagnosis: report of first 6 months in clinical practice. <i>J Matern Fetal Neonatal Med</i>, 2014; 27 (13): 1333-8.</p>	<p>Not relevant PICO: intervention</p>
<p>Konialis C, Pangalos C. Dilemmas in Prenatal Chromosomal Diagnosis Revealed Through a Single Center's 30 Years' Experience and 90,000 Cases. <i>Fetal Diagn Ther</i>, 2015.</p>	<p>Not relevant PICO: intervention</p>
<p>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. <i>Expert Opin Biol Ther</i> 2012; 12 Suppl 1: S27-33.</p>	<p>Not relevant PICO: population</p>
<p>Lalatta F, Tint GS. Counseling parents before prenatal diagnosis: do we need to say more about the sex chromosome aneuploidies? <i>Am J Med Genet A</i>, 2013; 161a (11): 2873-9.</p>	<p>Not relevant study design</p>
<p>Lam KW, Jiang P, Liao GJ, Chan KC, Leung TY, Chiu RW, et al. Noninvasive prenatal diagnosis of monogenic diseases by targeted massively parallel sequencing of</p>	<p>Targeted prenatal genetic testing for a specific aberration</p>

maternal plasma: application to beta-thalassemia. Clin Chem 2012; 58 (10): 1467-75.	
Lamb AN,Rosenfeld JA,Coppinger J,Dodge ET,Dabell MP,Torchia BS, et al. Defining the impact of maternal cell contamination on the interpretation of prenatal microarray analysis. Genetics in Medicine, 2012; 14 (11): 914-21.	Not relevant PICO: outcome
Lan JH, Zhang Q. Clinical applications of next-generation sequencing in histocompatibility and transplantation. Curr Opin Organ Transplant, 2015; 20 (4): 461-7.	Not relevant study design
Lapaire O,Johnson KL, Bianchi DW. Method for extraction of high-quantity and -quality cell-free DNA from amniotic fluid. Methods Mol Biol, 2008; 444303-9.	Not relevant PICO: intervention
Lau TK,Chan MK,Salome Lo PS,Chan HYC,Chan WK,Koo TY, et al. Non-invasive prenatal screening of fetal sex chromosomal abnormalities: perspective of pregnant women. The Journal of Maternal-Fetal & Neonatal Medicine, 2012; 25 (12): 2616-19.	Not relevant PICO: Intervention
Lau TK,Chen F,Pan X,Pooh RK,Jiang F,Li Y, et al. Noninvasive prenatal diagnosis of common fetal chromosomal aneuploidies by maternal plasma DNA sequencing. J Matern Fetal Neonatal Med, 2012; 25 (8): 1370-4.	Not relevant PICO: outcome
Lebo RV,Novak RW,Wolfe K,Michelson M,Robinson H, Mancuso MS. Discordant circulating fetal DNA and subsequent cytogenetics reveal false negative, placental mosaic, and fetal mosaic cfDNA genotypes. J Transl Med, 2015; 13 (1): 260.	Not relevant PICO: outcome
Lebo RV, Tonk VS. Analyzing the most frequent disease loci in targeted patient categories optimizes disease gene identification and test accuracy worldwide. J Transl Med, 2015; 13 (1): 16.	Not relevant PICO: outcome

Lee CN,Lin SY,Lin CH,Shih JC,Lin TH, Su YN. Clinical utility of array comparative genomic hybridisation for prenatal diagnosis: a cohort study of 3171 pregnancies. <i>Bjog</i> , 2012; 119 (5): 614-25.	Not relevant PICO: intervention
Leung TY,Vogel I,Lau TK,Chong W,Hyett JA,Petersen OB, et al. Identification of submicroscopic chromosomal aberrations in fetuses with increased nuchal translucency and apparently normal karyotype. <i>Ultrasound Obstet Gynecol</i> , 2011; 38 (3): 314-9.	Not relevant PICO: intervention
Levenson D. Array CGH increasingly used in prenatal and postnatal testing. <i>Am J Med Genet A</i> 2012; 158a (3): viii-ix.	Not relevant study design : case series
Li P,Pomianowski P,DiMaio MS,Florio JR,Rossi MR,Xiang B, et al. Genomic characterization of prenatally detected chromosomal structural abnormalities using oligonucleotide array comparative genomic hybridization. <i>Am J Med Genet A</i> , 2011; 155a (7): 1605-15.	Not relevant study design : case series
Liao C,Fu F,Li R,Xie GE,Zhang YL,Li J, et al. Implementation of high-resolution SNP arrays in the investigation of fetuses with ultrasound malformations: 5 years of clinical experience. <i>Clin Genet</i> , 2014; 86 (3): 264-9.	Not relevant PICO: intervention
Liao C,Li R,Fu F,Xie G,Zhang Y,Pan M, et al. Prenatal diagnosis of congenital heart defect by genome-wide high-resolution SNP array. <i>Prenat Diagn</i> , 2014; 34 (9): 858-63.	Not relevant PICO: intervention
Liao GJ,Chan KC,Jiang P,Sun H,Leung TY,Chiu RW, et al. Noninvasive prenatal diagnosis of fetal trisomy 21 by allelic ratio analysis using targeted massively parallel sequencing of maternal plasma DNA. <i>PLoS One</i> , 2012; 7 (5): e38154.	Not relevant PICO: outcome
Liao GJ,Lun FM,Zheng YW,Chan KC,Leung TY,Lau TK, et al. Targeted massively parallel sequencing of maternal plasma DNA permits efficient and unbiased detection of fetal alleles. <i>Clin Chem</i> , 2011; 57 (1): 92-101.	Not relevant PICO: outcome

Lichtenbelt KD,Knoers NV, Schuring-Blom GH. From karyotyping to array-CGH in prenatal diagnosis. <i>Cytogenet Genome Res</i> 2011; 135 (3-4): 241-50.	Not relevant study design
Liehr T,Karamysheva T,Merkas M,Brecevic L,Hamid AB,Ewers E, et al. Somatic mosaicism in cases with small supernumerary marker chromosomes. <i>Curr Genomics</i> , 2010; 11 (6): 432-9.	Not relevant study design
Lin TH,Lin SY, Lee CN,Shih JC,Chung CH, Su YN. Should array CGH be applied to all routine prenatal care? <i>Prenat Diagn</i> 2012; 32 (7): 710; author reply 11-2.	Not relevant study design: letter
Lin XL,Xie FN,Tang SH,Xu XQ,Wu H,Zheng ZK, et al. [Prenatal diagnosis and clinical counseling for fetal chromosomal reciprocal translocations]. <i>Zhonghua Yi Xue Yi Chuan Xue Za Zhi</i> 2013; 30 (5): 612-5.	Language
Lo JO,Feist CD,Hashima J, Shaffer BL. Jacobsen syndrome detected by noninvasive prenatal testing. <i>Obstet Gynecol</i> , 2015; 125 (2): 387-9.	Not relevant study design: case report
Lo YM. Noninvasive fetal whole-genome sequencing from maternal plasma: feasibility studies and future directions. <i>Clin Chem</i> 2013; 59 (4): 601-3.	Not relevant study design
Lund IC,Christensen R,Petersen OB,Vogel I, Vestergaard EM. Chromosomal microarray in fetuses with increased nuchal translucency. <i>Ultrasound Obstet Gynecol</i> , 2014.	Not relevant PICO: intervention
Luo CY,Ma DY,Zhang JJ,Hu P,Cao L, Ji XQ, et al. [Prenatal diagnosis of 22q11.2 microdeletion by multiplex ligation-dependent probe amplification]. <i>Zhonghua Fu Chan Ke Za Zhi</i> 2013; 48 (11): 824-7.	Language
Lv W,Wei X,Guo R,Liu Q,Zheng Y,Chang J, et al. Non-invasive Prenatal Testing for Wilson Disease by Use of Circulating Single-Molecule Amplification and Resequencing Technology (cSMART). <i>Clin Chem</i> , 2014.	Targeted prenatal genetic testing for a specific aberration
Lv W,Wei X,Guo R,Liu Q,Zheng Y,Chang J, et al. Noninvasive prenatal testing for Wilson disease by use of circulating single-molecule amplification and resequencing technology (cSMART). <i>Clin Chem</i> , 2015; 61 (1): 172-81.	Targeted prenatal genetic testing for a specific aberration

<p>Maarse W,Rozendaal AM,Pajkr E,Vermeij-Keers C,Mink van der Molen AB, van den Boogaard MJ. A systematic review of associated structural and chromosomal defects in oral clefts: when is prenatal genetic analysis indicated? J Med Genet, 2012; 49 (8): 490-8.</p>	<p>Not relevant PICO: intervention</p>
<p>Macera MJ,Sobrinho 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, 2014.</p>	<p>Not relevant study design: case study</p>
<p>Macera MJ,Sobrinho 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.</p>	<p>Not relevant study design: case report</p>
<p>Machado IN,Heinrich JK, Barini R. Genomic imbalances detected through array CGH in fetuses with holoprosencephaly. Arq Neuropsiquiatr, 2011; 69 (1): 3-8.</p>	<p>Not relevant PICO: population</p>
<p>Mackie FL,Carss KJ,Hillman SC,Hurles ME,Kilby MD,Author A, et al. Exome sequencing in fetuses with structural malformations. Journal of Clinical Medicine, 2014; 3 (3): 747-62.</p>	<p>Not relevant study design</p>
<p>Mademont-Soler I,Morales C,Soler A,Martinez-Crespo JM,Shen Y,Margarit E, et al. Prenatal diagnosis of chromosomal abnormalities in fetuses with abnormal cardiac ultrasound findings: evaluation of chromosomal microarray-based analysis. Ultrasound Obstet Gynecol, 2013; 41 (4): 375-82.</p>	<p>Not relevant PICO: intervention</p>
<p>Madjunkova S,Tong Li C,Vlasschaert M,Adams M,Chitayat D,Maire G, et al. QF-PCR rapid aneuploidy screen and aCGH analysis of cell free fetal (cff) DNA in supernatant of compromised amniotic fluids (AF). Prenat Diagn 2014; 34 (10): 970-6.</p>	<p>Not relevant PICO: less than 20 patients</p>
<p>Makela NL,Birch PH,Friedman JM, Marra CA. Parental perceived value of a diagnosis for intellectual disability (ID): a qualitative comparison of families</p>	<p>Not relevant study design</p>

with and without a diagnosis for their child's ID. <i>Am J Med Genet A</i> , 2009; 149a (11): 2393-402.	
Malvestiti F, De Toffol S, Grimi B, Chinetti S, Marcato L, Agrati C, et al. De novo small supernumerary marker chromosomes detected on 143,000 consecutive prenatal diagnoses: chromosomal distribution, frequencies, and characterization combining molecular cytogenetics approaches. <i>Prenat Diagn</i> , 2014; 34 (5): 460-8.	Not relevant PICO: intervention
Mansour S. Re: 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. F. Scott, K. Murphy, L. Carey, W. Greville, N. Mansfield, P. Barahona, R. Robertson and A. McLennan. <i>Ultrasound Obstet Gynecol</i> 2013; 41: 500-507. <i>Ultrasound Obstet Gynecol</i> 2013; 41 (5): 489.	Not relevant study design
Marle N, Martinet D, Aboura A, Joly-Helas G, Andrieux J, Flori E, et al. Molecular characterization of 39 de novo sSMC: contribution to prognosis and genetic counselling, a prospective study. <i>Clin Genet</i> , 2014; 85 (3): 233-44.	Not relevant PICO: intervention
Maya I, Davidov B, Gershovitz L, Zalstein Y, Taub E, Coppinger J, et al. Diagnostic utility of array-based comparative genomic hybridization (aCGH) in a prenatal setting. <i>Prenat Diagn</i> , 2010; 30 (12-13): 1131-7.	Not relevant PICO: intervention
Meng J, Matarese C, Crivello J, Wilcox K, Wang D, DiAdamo A, et al. Changes in and Efficacies of Indications for Invasive Prenatal Diagnosis of Cytogenomic Abnormalities: 13 Years of Experience in a Single Center. <i>Med Sci Monit</i> , 2015; 21:1942-8.	Not relevant PICO: intervention
Meng M, Huo R, Han MY, Chi FL, Dai P, He L, et al. Detection of common deafness mutation by maternal plasma cell-free DNA. <i>Eur Rev Med Pharmacol Sci</i> 2014; 18 (10): 1544-8.	Targeted prenatal genetic testing for a specific aberration

Mennuti MT,Chandrasekaran S,Khalek N, Dugoff L. Cell-free DNA Screening and Sex Chromosome Abnormalities. Prenat Diagn, 2015.	Not relevant study design
Mercier S,Dubourg C,Belleguic M,Pasquier L,Loget P,Lucas J, et al. Genetic counseling and "molecular" prenatal diagnosis of holoprosencephaly (HPE). Am J Med Genet C Semin Med Genet, 2010; 154c (1): 191-6.	Not relevant PICO: Intervention
Meschia JF,Nalls M,Matarin M,Brott TG,Brown RD, Jr.,Hardy J, et al. Siblings with ischemic stroke study: results of a genome-wide scan for stroke loci. Stroke (00392499), 2011; 42 (10): 2726-32.	Not relevant PICO: population
Mosca-Boidron AL,Faivre L,Aho S,Marle N,Truntzer C,Rousseau T, et al. An improved method to extract DNA from 1 ml of uncultured amniotic fluid from patients at less than 16 weeks' gestation. PLoS One 2013; 8 (4): e59956.	Not relevant PICO: outcome
Nepomnyashchaya YN,Artemov AV,Roumiantsev SA,Roumyantsev AG, Zhavoronkov A. Non-invasive prenatal diagnostics of aneuploidy using next-generation DNA sequencing technologies, and clinical considerations. Clin Chem Lab Med 2013; 51 (6): 1141-54.	Not relevant study design
New MI,Tong YK,Yuen T,Jiang P,Pina C,Chan KC, et al. Noninvasive prenatal diagnosis of congenital adrenal hyperplasia using cell-free fetal DNA in maternal plasma. J Clin Endocrinol Metab, 2014; 99 (6): E1022-30.	Targeted prenatal genetic testing for a specific aberration
Novelli A,Cavalli P, Bernardini L. The future of prenatal diagnosis: karyotype, microarray or both? Technical and ethical considerations. Expert Rev Proteomics 2013; 10 (2): 131-4.	Not relevant study design
Oitmaa E,Peters M,Vaidla K,Andreson R,Magi R,Slavin G, et al. Molecular diagnosis of Down syndrome using quantitative APEX-2 microarrays. Prenat Diagn, 2010; 30 (12-13): 1170-7.	Not relevant PICO: outcome

<p>Oneda B,Baldinger R,Reissmann R,Reshetnikova I,Krejci P,Masood R, et al. High-resolution chromosomal microarrays in prenatal diagnosis significantly increase diagnostic power. Prenat Diagn, 2014; 34 (6): 525-33.</p>	<p>Not relevant PICO: intervention</p>
<p>Ormond KE,Wheeler MT,Hudgins L,Klein TE,Butte AJ,Altman RB, et al. Challenges in the clinical application of whole-genome sequencing. Lancet, 2010; 375 (9727): 1749-51.</p>	<p>Not relevant study design</p>
<p>Palomaki G,Kloza E,Lambert-Messerlian G,van den Boom D,Ehrich M,Deciu C, et al. Circulating cell free (ccf) DNA testing: are some test failures informative? Prenat Diagn, 2014.</p>	<p>Not relevant PICO: outcome</p>
<p>Palomaki GE,Kloza EM,Lambert-Messerlian GM,Haddow JE,Neveux LM,Ehrich M, et al. DNA sequencing of maternal plasma to detect Down syndrome: an international clinical validation study. Genet Med, 2011; 13 (11): 913-20.</p>	<p>Not relevant PICO: outcome</p>
<p>Papasavva T,van Ijcken WF,Kockx CE,van den Hout MC,Kountouris P,Kythreotis L, et al. Next generation sequencing of SNPs for non-invasive prenatal diagnosis: challenges and feasibility as illustrated by an application to beta-thalassaemia. Eur J Hum Genet, 2013; 21 (12): 1403-10.</p>	<p>Targeted prenatal genetic testing for a specific aberration</p>
<p>Park SJ,Jung EH,Ryu RS,Kang HW,Ko JM,Kim HJ, et al. Clinical implementation of whole-genome array CGH as a first-tier test in 5080 pre and postnatal cases. Mol Cytogenet, 2011; 412.</p>	<p>Not relevant PICO: Intervention</p>
<p>Phadke S. Cytogenetic microarray in prenatal and postnatal diagnosis. Mol Cytogenet 2014; 7 (Suppl 1 Proceedings of the International Conference on Human): I32.</p>	<p>Not relevant study design: conference abstract</p>
<p>Pieters JJ,Kooper AJ,Eggink AJ,Verhaak CM,Otten BJ,Braat DD, et al. Parents' perspectives on the unforeseen finding of a fetal sex chromosomal aneuploidy. Prenat Diagn, 2011; 31 (3): 286-92.</p>	<p>Not relevant study design</p>

<p>Pieters JJ, Verhaak CM, Braat DD, van Leeuwen E, Smits AP. Experts' opinions on the benefit of an incidental prenatal diagnosis of sex chromosomal aneuploidy: a qualitative interview survey. <i>Prenat Diagn</i>, 2012; 32 (12): 1151-7.</p>	<p>Not relevant PICO: population</p>
<p>Pietropolli A, Vicario R, Peconi C, Zampatti S, Quitadamo MC, Capogna MV, et al. Transabdominal coelocentesis as early source of fetal DNA for chromosomal and molecular diagnosis. <i>J Matern Fetal Neonatal Med</i> 2014; 27 (16): 1656-60.</p>	<p>Not relevant PICO: outcome</p>
<p>Pooh RK. Sonogenetics in fetal neurology. <i>Semin Fetal Neonatal Med</i> 2012; 17 (6): 353-9.</p>	<p>Not relevant PICO: intervention</p>
<p>Rabinowitz M, Valenti E, Pettersen B, Sigurjonsson S, Hill M, Zimmermann B. Noninvasive aneuploidy detection by multiplexed amplification and sequencing of polymorphic Loci. <i>Obstet Gynecol</i>, 2014; 123 Suppl 1167s.</p>	<p>Not relevant study design</p>
<p>Rad S, Turner AL, Ratousi D, Aghajanian P, Williams J, 3rd, Esakoff TF. Predictors for choosing array-comparative genomic hybridization for prenatal diagnosis. <i>Obstet Gynecol</i> 2014; 123 Suppl 1100s-1s.</p>	<p>Not relevant study design: conference abstract</p>
<p>Rajcan-Separovic E. Chromosome microarrays in human reproduction. <i>Hum Reprod Update</i>, 2012; 18 (5): 555-67.</p>	<p>Not relevant study design</p>
<p>Rava RP, Srinivasan A, Sehnert AJ, Bianchi DW. Circulating fetal cell-free DNA fractions differ in autosomal aneuploidies and monosomy X. <i>Clin Chem</i>, 2014; 60 (1): 243-50.</p>	<p>Not relevant PICO: outcome</p>
<p>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. <i>CMAJ: Canadian Medical Association Journal</i>, 2015; 187 (6): E190-7.</p>	<p>Not relevant study design</p>
<p>Rembouskos G, Passamonti U, De Robertis V, Tempesta A, Campobasso G, Volpe G, et al. Aberrant right subclavian artery (ARSA) in unselected population at first and second trimester</p>	<p>Not relevant PICO: intervention</p>

ultrasonography. Prenat Diagn, 2012; 32 (10): 968-75.	
Riedijk S,Diderich KEM,van der Steen SL,Govaerts LCP,Joosten M,Knapen MFCM, et al. The psychological challenges of replacing conventional karyotyping with genomic SNP array analysis in prenatal testing. Journal of Clinical Medicine, 2014; 3 (3): 713-23.	Not relevant study design
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
Robinson WP,Penaherrera MS,Jiang R,Avila L,Sloan J,McFadden DE, et al. Assessing the role of placental trisomy in preeclampsia and intrauterine growth restriction. Prenat Diagn, 2010; 30 (1): 1-8.	Not relevant PICO: outcome
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
Rosenfeld JA,Morton SA,Hummel C,Sulpizio SG,McDaniel LD,Schultz RA, et al. Experience Using a Rapid Assay for Aneuploidy and Microdeletion/Microduplication Detection in over 2,900 Prenatal Specimens. Fetal Diagn Ther, 2014; 36 (3): 231-41.	Not relevant PICO: intervention
Ryan A,Baner J,Demko Z,Hill M,Sigurjonsson S,Baird ML, et al. Informatics-based, highly accurate, noninvasive prenatal paternity testing. Genet Med 2013; 15 (6): 473-7.	Not relevant PICO: outcome

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.	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. <i>Prenat Diagn</i> , 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. <i>Am J Obstet Gynecol</i> , 2014.	Not relevant PICO: Intervention
Sayres LC, Allyse M, Cho MK. Integrating stakeholder perspectives into the translation of cell-free fetal DNA testing for aneuploidy. <i>Genome Med</i> , 2012; 4 (6): 49.	Not relevant study design
Schmid M, Stary S, Springer S, Bettelheim D, Husslein P, Streubel B. Prenatal microarray analysis as second-tier diagnostic test: single-center prospective study. <i>Ultrasound Obstet Gynecol</i> , 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. <i>Ultrasound Obstet Gynecol</i> , 2009; 34 (6): 618-22.	Not relevant PICO: intervention
Schulz-Hanke I. [Prenatal diagnosis - how powerful is DNA microarray compared to karyotyping?]. <i>Z Geburtshilfe Neonatol</i> , 2013; 217 (1): 3-4.	Not relevant study design
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. <i>Ultrasound Obstet Gynecol</i> , 2013; 41 (5): 500-7.	Not relevant PICO: intervention

<p>Shaffer LG,Coppinger J,Alliman S,Torchia BA,Theisen A,Ballif BC, et al. Comparison of microarray-based detection rates for cytogenetic abnormalities in prenatal and neonatal specimens. <i>Prenat Diagn</i>, 2008; 28 (9): 789-95.</p>	<p>Not relevant PICO: intervention</p>
<p>Shaffer LG,Dabell MP,Fisher AJ,Coppinger J,Bandholz AM,Ellison JW, et al. Experience with microarray-based comparative genomic hybridization for prenatal diagnosis in over 5000 pregnancies. <i>Prenat Diagn</i>, 2012; 32 (10): 976-85.</p>	<p>Not relevant PICO: intervention</p>
<p>Shaffer LG,Dabell MP,Rosenfeld JA,Neill NJ,Ballif BC,Coppinger J, et al. Referral patterns for microarray testing in prenatal diagnosis. <i>Prenatal Diagnosis</i>, 2012; 32 (4): 344-50.</p>	<p>Not relevant PICO: intervention</p>
<p>Shaffer LG,Rosenfeld JA,Dabell MP,Coppinger J,Bandholz AM,Ellison JW, et al. Detection rates of clinically significant genomic alterations by microarray analysis for specific anomalies detected by ultrasound. <i>Prenat Diagn</i>, 2012; 32 (10): 986-95.</p>	<p>Not relevant PICO: intervention</p>
<p>Shen GS,Zhang S, He PY. [Application of gene array chip in diagnosis of cytogenetics of recurrent spontaneous abortion]. <i>Marias beställning</i>, 2013; 48 (7): 515-8.</p>	<p>Language</p>
<p>Sheth F,Rahman M,Liehr T,Desai M,Patel B,Modi C, et al. Prenatal screening of cytogenetic anomalies - a Western Indian experience. <i>BMC Pregnancy Childbirth</i>, 2015; 1590.</p>	<p>Not relevant PICO: Intervention</p>
<p>Sillence KA,Madgett TE,Roberts LA,Overton TG,Avent ND,Author A, et al. Non-invasive screening tools for down's syndrome: A review. <i>Diagnostics</i>, 2013; 3 (2): 291-314.</p>	<p>Not relevant study design</p>
<p>Srebniak M,Boter M,Oudesluijs G,Joosten M,Govaerts L, Van Opstal D, et al. Application of SNP array for rapid prenatal diagnosis: implementation, genetic counselling and diagnostic flow. <i>Eur J Hum Genet</i>, 2011; 19 (12): 1230-7.</p>	<p>Not relevant PICO: intervention</p>

<p>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. <i>Mol Cytogenet</i>, 2012; 5 (1): 14.</p>	<p>Not relevant PICO: intervention</p>
<p>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. <i>Hum Mutat</i>, 2013; 34 (9): 1298-303.</p>	<p>Not relevant PICO: Intervention</p>
<p>Srebniak MI, Van Opstal D, Joosten M, Diderich KE, de Vries FA, Riedijk S, et al. Whole-genome array as a first-line cytogenetic test in prenatal diagnosis. <i>Ultrasound Obstet Gynecol</i>, 2015; 45 (4): 363-72.</p>	<p>Not relevant study design</p>
<p>Straver R, Sistermans EA, Holstege H, Visser A, Oudejans CB, Reinders MJ. WISECONDOR: detection of fetal aberrations from shallow sequencing maternal plasma based on a within-sample comparison scheme. <i>Nucleic Acids Res</i>, 2014; 42 (5): e31.</p>	<p>Not relevant PICO: outcome</p>
<p>Srinivasan A, Bianchi DW, Huang H, Sehnert AJ, Rava RP. Noninvasive detection of fetal subchromosome abnormalities via deep sequencing of maternal plasma. <i>Am J Hum Genet</i>, 2013; 92 (2): 167-76.</p>	<p>Not relevant PICO: population</p>
<p>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. <i>Biomed Res Int</i>, 2015; 2015426379.</p>	<p>Not relevant PICO: intervention</p>
<p>Sun LH, Li L, Wang XW, Zhu YZ, Chai YC, Li XH, et al. [Prenatal screening and diagnosis of genetic deafness by microarray]. <i>Marias beställning</i>, 2012; 47 (12): 991-5.</p>	<p>Language</p>
<p>Tabor HK, Murray JC, Gammill HS, Kitzman JO, Snyder MW, Ventura M, et al. Non-invasive fetal genome sequencing: opportunities and challenges. <i>Marias beställning</i>, 2012; 158a (10): 2382-4.</p>	<p>Not relevant study design</p>

Talkowski ME,Ordulu Z,Pillalamarri V,Benson CB,Blumenthal I,Connolly S, et al. Clinical diagnosis by whole-genome sequencing of a prenatal sample. <i>N Engl J Med</i> , 2012; 367 (23): 2226-32.	Not relevant study design: case study
Tang S,Lv J,Chen X,Bai L,Li H,Chen C, et al. Prenatal Diagnosis of DNA Copy Number Variations by Genomic Single-Nucleotide Polymorphism Array in Fetuses with Congenital Heart Defects. <i>Fetal Diagn Ther</i> , 2015.	Not relevant PICO: intervention
Thung DT,Beulen L,Hehir-Kwa J,Faas BH,Author A,Radboud University Medical Center N, et al. Implementation of whole genome massively parallel sequencing for noninvasive prenatal testing in laboratories. <i>Expert Review of Molecular Diagnostics</i> , 2015; 15 (1): 111-24.	Not relevant study design
Timbs AT,Rugless MJ,Gallienne AE,Haywood AM,Henderson SJ, Old JM. Prenatal diagnosis of hemoglobinopathies by pyrosequencing: a more sensitive and rapid approach to fetal genotyping. <i>Hemoglobin</i> , 2012; 36 (2): 144-50.	targeted prenatal genetic testing for a specific aberration
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. <i>J Ultrasound Med</i> , 2009; 28 (12): 1735-42.	Not relevant study design: case report
Tyreman M,Abbott KM,Willatt LR,Nash R,Lees C,Whittaker J, et al. High resolution array analysis: diagnosing pregnancies with abnormal ultrasound findings. <i>J Med Genet</i> , 2009; 46 (8): 531-41.	Not relevant PICO: intervention
Valduga M,Philippe C,Bach Segura P,Thiebaugeorges O,Miton A,Beri M, et al. A retrospective study by oligonucleotide array-CGH analysis in 50 fetuses with multiple malformations. <i>Prenat Diagn</i> , 2010; 30 (4): 333-41.	Not relevant PICO: intervention
Walser SA,Kellom KS,Palmer SC, Bernhardt BA. Comparing genetic counselor's and patient's perceptions of needs in prenatal chromosomal microarray testing. <i>Prenat Diagn</i> , 2015.	Not relevant PICO: intervention

<p>Van den Veyver IB,Patel A,Shaw CA,Pursley AN,Kang SH,Simovich MJ, et al. Clinical use of array comparative genomic hybridization (aCGH) for prenatal diagnosis in 300 cases. Prenat Diagn, 2009; 29 (1): 29-39.</p>	<p>Not relevant PICO: intervention</p>
<p>van der Steen SL,Diderich KE,Riedijk SR,Verhagen-Visser J,Govaerts LC,Joosten M, et al. Pregnant couples at increased risk for common aneuploidies choose maximal information from invasive genetic testing. Clin Genet, 2014.</p>	<p>Not relevant PICO: intervention</p>
<p>van der Zwaag PA,Jongbloed JDH, van Tintelen JP. Genetic diagnosis through whole-exome sequencing... N Engl J Med. 2013 Oct 17;369(16):1502-11. New England Journal of Medicine, 2014; 370 (11): 1067-67.</p>	<p>Not relevant study design: letter</p>
<p>Van Opstal D,de Vries F,Govaerts L,Boter M,Lont D,van Veen S, et al. Benefits and burdens of using a SNP array in pregnancies at increased risk for the common aneuploidies. Hum Mutat, 2015; 36 (3): 319-26.</p>	<p>Not relevant PICO: intervention</p>
<p>Vanakker O,Vilain C,Janssens K, Van der Aa N,Smits G,Bandelier C, et al. Implementation of genomic arrays in prenatal diagnosis: the Belgian approach to meet the challenges. Marias beställning, 2014; 57 (4): 151-6.</p>	<p>Not relevant study design</p>
<p>Wapner RJ,Martin CL,Levy B,Ballif BC,Eng CM,Zachary JM, et al. Chromosomal microarray versus karyotyping for prenatal diagnosis. N Engl J Med, 2012; 367 (23): 2175-84.</p>	<p>Not relevant PICO: intervention</p>
<p>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.</p>	<p>Not relevant PICO: Population</p>
<p>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.</p>	<p>Not relevant study design</p>

<p>Wen S,Wei F,He Y,Xu W,Xie R,Zhang X, et al. [Application of noninvasive fetal trisomy testing based on massively parallel sequencing for the detection of chromosomal deletions and duplications]. <i>Marias beställning</i>, 2014; 31 (5): 557-60.</p>	<p>Language</p>
<p>Wenstrom KD. Microarray analysis: elegant, accurate, and expensive. <i>Marias beställning</i>, 2014; 124 (2 Pt 1): 199-201.</p>	<p>Not relevant study design : editorial</p>
<p>Westerfield LE,Stover SR,Mathur VS,Nassef SA,Carter TG,Yang Y, et al. Reproductive genetic counseling challenges associated with diagnostic exome sequencing in a large academic private reproductive genetic counseling practice. <i>Prenat Diagn</i>, 2015.</p>	<p>Not relevant PICO: intervention</p>
<p>Vestergaard EM,Christensen R,Petersen OB, Vogel I. Prenatal diagnosis: array comparative genomic hybridization in fetuses with abnormal sonographic findings. <i>Acta Obstet Gynecol Scand</i>, 2013; 92 (7): 762-8.</p>	<p>Not relevant PICO: intervention</p>
<p>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. <i>PLoS ONE</i>, 2012; 7:9 Article Number: e45073.</p>	<p>Not relevant PICO: intervention</p>
<p>Vialard F,Molina Gomes D,Leroy B,Quarello E,Escalona A,Le Sciellour C, et al. Array comparative genomic hybridization in prenatal diagnosis: another experience. <i>Fetal Diagn Ther</i>, 2009; 25 (2): 277-84.</p>	<p>Not relevant PICO: intervention</p>
<p>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. <i>J Obstet Gynaecol Can</i>, 2014; 36 (10): 927-39.</p>	<p>Not relevant PICO: intervention</p>
<p>Wit MC,Srebniak MI,Govaerts LC,Opstal D,Galjaard RJ, Go AT. Additional value of prenatal genomic array testing in fetuses with isolated structural ultrasound abnormalities and a normal karyotype: a systematic review of the literature (Provisional abstract).</p>	<p>Not relevant PICO: intervention</p>

Ultrasound in Obstetrics and Gynecology, 2014; 43 (2): 139-46.	
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. Marias beställning, 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.
Xu Z,Geng Q,Luo F,Xu F,Li P, Xie J. Multiplex ligation-dependent probe amplification and array comparative genomic hybridization analyses for prenatal diagnosis of cytogenomic abnormalities. Mol Cytogenet, 2014; 7 (1): 84.	Not relevant PICO: intervention
Yakut S,Cetin Z,Simsek M,Mendicioglu, II,Toru HS,Karauzum SB, et al. Rare Structural Chromosomal Abnormalities in Prenatal Diagnosis; Clinical and Cytogenetic Findings on 10125 Prenatal Cases. Turk Patoloji Derg, 2014.	Not relevant PICO:intervention
Yan Y,Wu Q,Zhang L,Wang X,Dan S,Deng D, et al. Detection of submicroscopic chromosomal aberrations by array-based comparative genomic hybridization in fetuses with congenital heart disease. Ultrasound Obstet Gynecol, 2014; 43 (4): 404-12.	Not relevant PICO: intervention

<p>Yatsenko SA,Davis S,Hendrix NW,Surti U,Emery S,Canavan T, et al. Application of chromosomal microarray in the evaluation of abnormal prenatal findings. Clin Genet, 2013; 84 (1): 47-54.</p>	<p>Not relevant PICO: intervention</p>
<p>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.</p>	<p>Not relevant study design: case report</p>
<p>Yilmaz Z,Sahin FI,Bulakbasi T,Yuregir OO,Tarim E, Yanik F. Ethical considerations regarding parental decisions for termination following prenatal diagnosis of sex chromosome abnormalities. Genet Couns, 2008; 19 (3): 345-52.</p>	<p>Not relevant study design</p>
<p>Yohe S,Hauge A,Bunjer K,Kemmer T,Bower M,Schomaker M, et al. Clinical Validation of Targeted Next-Generation Sequencing for Inherited Disorders. Archives of Pathology & Laboratory Medicine, 2015; 139 (2): 204-10.</p>	<p>Not relevant PICO: population</p>
<p>Yoo SK,Chan Lim B,Byeun J,Hwang H,Joong Kim K,Seung Hwang Y, et al. Noninvasive Prenatal Diagnosis of Duchenne Muscular Dystrophy: Comprehensive Genetic Diagnosis in Carrier, Proband, and Fetus. Clin Chem, 2015.</p>	<p>Targeted prenatal genetic testing for a specific aberration</p>
<p>Yu SCY,Chan KCA,Zheng YWL,Jiang P,Liao GJW,Sun H, et al. Size-based molecular diagnostics using plasma DNA for noninvasive prenatal testing. Proceedings of the National Academy of Sciences of the United States of America, 2014; 111 (23): 8583-88.</p>	<p>Not relevant study design</p>
<p>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.</p>	<p>Not relevant PICO: outcome</p>
<p>Zhang J,Hu P,Luo C, Ji Q,Zhou J,Liu A, et al. [Application of multiplex ligation-dependent probe amplification for rapid detection of aneuploidies and structural chromosomal abnormalities in prenatal diagnosis]. Marias beställning, 2014; 31 (1): 11-5.</p>	<p>Language</p>

<p>Zhu R,Zhu X,Yang Y,Duan H,Zhang Y,Wu X, et al. [Application of different technologies for distinguishing true and pseudo mosaicisms during prenatal diagnosis]. <i>Marias beställning</i>, 2014; 31 (5): 636-40.</p>	<p>Language</p>
<p>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. <i>Mol Genet Genomic Med</i>, 2014; 2 (2): 166-75.</p>	<p>Not relevant PICO: intervention</p>
<p>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. <i>Tijdschrift voor Kindergeneeskunde</i>, 2014; 82 (1): 3-18.</p>	<p>Language</p>
<p>Systematic review</p>	
<p>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. <i>Ultrasound Obstet Gynecol</i>, 2015; 45 (3): 249–66.</p>	<p>Systematic review Differences in inclusion exclusion criteria: includes studies published from 2011. 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. Pooling of data was not made using the bivariate model or the hierarchical summary receiver operating characteristic model.</p>

Studies with high risk of bias/Studier med låg kvalitet
Agatista 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. <i>Prenat Diagn</i> , 2015; 35 (7): 692-8.
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. <i>Obstet Gynecol</i> , 2015; 125 (2): 375-82.
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>Obstet Gynecol</i> , 2013; 121 (5): 1057-62.
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. <i>PLoS One</i> , 2012; 7 (2): e27835.
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. <i>American Journal of Obstetrics and Gynecology</i> , 2014; 211 (5): 527.e1-27.e17.
Drury S, Williams H, Trump N, Boustred C, Lench N, Scott RH, et al. Exome sequencing for prenatal diagnosis of fetuses with sonographic abnormalities. <i>Prenat Diagn</i> , 2015.
Fan HC, Gu W, Wang J, Blumenfeld YJ, El-Sayed YY, Quake SR. Non-invasive prenatal measurement of the fetal genome. <i>Marias beställning</i> , 2012; 487 (7407): 320-4.
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>Prenat Diagn</i> , 2013; 33 (6): 569-74.
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. <i>Prenat Diagn</i> , 2013; 33 (7): 707-10.

Jensen TJ,Dzakula Z,Deciu C,van den Boom D, Ehrich M. Detection of microdeletion 22q11.2 in a fetus by next-generation sequencing of maternal plasma. <i>Clin Chem</i> , 2012; 58 (7): 1148-51.
Korostelev S,Totchiev G,Kanivets I, Gnetetskaya V. Association of non-invasive prenatal testing and chromosomal microarray analysis for prenatal diagnostics. <i>Gynecol Endocrinol</i> , 2014; 30 Suppl 113-6.
Lau TK,Jiang FM,Stevenson RJ,Lo TK,Chan LW,Chan MK, et al. Secondary findings from non-invasive prenatal testing for common fetal aneuploidies by whole genome sequencing as a clinical service. <i>Prenat Diagn</i> , 2013; 33 (6): 602-8.
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; 111 (20): 7415-20.
Meck JM,Kramer Dugan E,Matyakhina L,Aviram A,Trunca C,Pineda-Alvarez D, et al. Noninvasive prenatal screening for aneuploidy: positive predictive values based on cytogenetic findings. <i>Am J Obstet Gynecol</i> , 2015; 213 (2): 214.e1-5.
Nicolaidis KH,Musci TJ,Struble CA,Syngelaki A, Gil MM. Assessment of fetal sex chromosome aneuploidy using directed cell-free DNA analysis. <i>Fetal Diagn Ther</i> , 2014; 35 (1): 1-6.
Nicolaidis KH,Syngelaki A,del Mar Gil M,Quezada MS, Zinevich Y. Prenatal detection of fetal triploidy from cell-free DNA testing in maternal blood. <i>Fetal Diagn Ther</i> , 2014; 35 (3): 212-7.
Samango-Sprouse C,Banjevic M,Ryan A,Sigurjonsson S,Zimmermann B,Hill M, et al. SNP-based non-invasive prenatal testing detects sex chromosome aneuploidies with high accuracy. <i>Prenatal Diagnosis</i> , 2013; 33 (7): 643-49.
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>Clin Chem</i> , 2011; 57 (7): 1042-9.
Shi X,Zhang Z,Cram DS, Liu C. Feasibility of noninvasive prenatal testing for common fetal aneuploidies in an early gestational window. <i>Clin Chim Acta</i> , 2015; 43924-8.

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.

Wapner RJ,Babiarz JE,Levy B,Stosic M,Zimmermann B,Sigurjonsson S, et al. Expanding the scope of non-invasive prenatal testing: Detection of fetal microdeletion syndromes. *Am J Obstet Gynecol*, 2014.

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.

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. *Prenatal Diagnosis*, 2012; 32 (13): 1233-41.