Washington State Health Care Authority

Cell-free DNA Prenatal Screening for Chromosomal Aneuploidies

Appendix H: Excluded Studies

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Health Technology Assessment Program (HTA)

Washington State Health Care Authority
PO Box 42712
Olympia, WA 98504-2712
(360) 725-5126
www.hca.wa.gov/hta
shtap@hca.wa.gov

Cell-free DNA Prenatal Screening for Chromosomal Aneuploidies

Draft Evidence Report

Prepared by:

Center for Evidence-based Policy
Oregon Health & Science University
3030 SW Moody, Suite 250
Portland, OR 97201
Phone: 503.494.2182
Fax: 503.494.3807
http://centerforevidencebasedpolicy.org/

Authors:

Beth Shaw, MSc,
Valerie King, MD, MPH,
Shannon Robalino MSc,
Michelle Eide,
Curtis Harrod, PhD, MPH

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Appendix H. Excluded Studies

Reference	Reason for Exclusion
Corrigenda: Non-invasive prenatal testing for aneuploidy and beyond: Challenges of responsible innovation in prenatal screening (European Journal of Human Genetics (2015) 23 (1438-1450) DOI: 10.1038/ejhg.2015.57). *European Journal of Human Genetics.* 2015. 23:1592	Not relevant publication type
Ainsworth, A. J., Holman, M. A., Codsi, E., Wick, M.. Use of Genetic Testing after Abnormal Screening Ultrasound: A Descriptive Cohort Study. *Gynecologic & Obstetric Investigation.* 2018. 83:466-470	Not appropriate population
Al-Alfy, M. E., El-Noury, M. A., Azmy, O. M., Bebars, M. A., Alsharnoubi, G., Dawood, R. M., Nagy, O. A., Ibrahim, A. K.. Molecular detection of CFFDNA for early laboratory diagnosis of X linked disorders carriers. *Middle East Fertility Society Journal.* 2017. 22:260-263	Not appropriate setting or country
Alberti, A., Salomon, L. J., Le Lorc'h, M., Couloux, A., Bussieres, L., Goupil, S., Malan, V., Pelletier, E., Hyon, C., Vialard, F., Rozenberg, P., Bouhanna, P., Oury, J. F., Schmitz, T., Romana, S., Weissenbach, J., Vekemans, M., Ville, Y.. Non-invasive prenatal testing for trisomy 21 based on analysis of cell-free fetal DNA circulating in the maternal plasma. *Prenatal Diagnosis.* 2015. 35:471-6	Not appropriate population
Allach El Khattabi, L., Brun, S., Gueguen, P., Chatron, N., Guichoux, E., Schutz, S., Nectoux, J., Sorlin, A., Quere, M., Boudjarane, J., Tsatsaris, V., Mandelbrot, L., Schluth-Bolard, C., Dupont, J. M., Rooryck, C., H. Consortium. Performance of Semiconductor sequencing platform for non-invasive prenatal genetic screening for fetal aneuploidies: results from a multicenter prospective cohort study in a clinical setting. *Ultrasound in Obstetrics & Gynecology.* 2018. 06:06	Not appropriate population
Alldred, S. K., Deeks, J. J., Guo, B., Neilson, J. P., Alfirevic, Z.. Second trimester serum tests for Down's Syndrome screening. *Cochrane Database of Systematic Reviews.* 2012. #volume#:CD009925	Not intervention or test of interest
Alldred, S. K., Takwoingi, Y., Guo, B., Pennant, M., Deeks, J. J., Neilson, J. P., Alfirevic, Z.. First trimester serum tests for Down's syndrome screening. *Cochrane Database of Systematic Reviews.* 2015. #volume#:#pages#	Not intervention or test of interest
Amorim Costa, C.. Non-invasive prenatal screening for chromosomal abnormalities using circulating cell-free fetal DNA in maternal plasma: Current applications, limitations and prospects. *Egyptian Journal of Medical Human Genetics.* 2017. 18:1-7	Not relevant publication type
An, N., Li, L. L., Wang, R. X., Li, L. L., Yue, J. M., Liu, R. Z.. Clinical and cytogenetic results of a series of amniocentesis cases from Northeast China: a report of 2500 cases. *Genetics & Molecular Research.* 2015. 14:15660-7	Not appropriate setting or country
Andrew, C., Koshy, T., Gopal, S., Paul, S. F. D.. A retrospective exploratory study of fetal genetic invasive procedures at a University Hospital. *Journal of Obstetrics & Gynaecology.* 2018. 38:906-910	Not appropriate setting or country
Anonymous, . Committee Opinion No. 640: Cell-Free DNA Screening For Fetal Aneuploidy. *Obstetrics & Gynecology.* 2015. 126:e31-7	Other - withdrawn guidance
Anonymous, . Committee Opinion Summary No. 640: Cell-Free DNA Screening For Fetal Aneuploidy. *Obstetrics & Gynecology.* 2015. 126:691-2	Other - withdrawn guidance
Arbabi, A., Rampasek, L., Brudno, M.. Cell-free DNA fragment-size distribution analysis for non-invasive prenatal CNV prediction. *Bioinformatics.* 2016. 32:1662-9	Not intervention or test of interest

Reference	Reason for Exclusion
Artieri, C. G., Haverty, C., Evans, E. A., Goldberg, J. D., Haque, I. S., Yaron, Y., Muzzey, D.. Noninvasive prenatal screening at low fetal fraction: comparing whole-genome sequencing and single-nucleotide polymorphism methods. *Prenatal Diagnosis.* 2017. 37:482-490	Not intervention or test of interest
Ashoor, G., Poon, L., Syngelaki, A., Mosimann, B., Nicolaides, K. H.. Fetal fraction in maternal plasma cell-free DNA at 11-13 weeks' gestation: effect of maternal and fetal factors. *Fetal Diagnosis & Therapy.* 2012. 31:237-43	Not appropriate population
Ashoor, G., Syngelaki, A., Poon, L. C., Rezende, J. C., Nicolaides, K. H.. Fetal fraction in maternal plasma cell-free DNA at 11-13 weeks' gestation: relation to maternal and fetal characteristics. *Ultrasound in Obstetrics & Gynecology.* 2013. 41:26-32	Not outcomes of interest
Ashoor, G., Syngelaki, A., Wagner, M., Birdir, C., Nicolaides, K. H.. Chromosome-selective sequencing of maternal plasma cell-free DNA for first-trimester detection of trisomy 21 and trisomy 18. *American Journal of Obstetrics & Gynecology.* 2012. 206:322.e1-5	Not appropriate population
Ayres, A. C., Whitty, J. A., Ellwood, D. A.. A cost-effectiveness analysis comparing different strategies to implement noninvasive prenatal testing into a Down syndrome screening program. *Australian & New Zealand Journal of Obstetrics & Gynaecology.* 2014. 54:412-7	Not appropriate setting or country
Badeau, M., Lindsay, C., Blais, J., Takwoingi, Y., Langlois, S., Légaré, F., Giguère, Y., Turgeon, A. F., Witteman, W., Rousseau, F.. Genomics-based non-invasive prenatal testing for detection of fetal chromosomal aneuploidy in pregnant women. *Cochrane Database of Systematic Reviews.* 2015. 2015:#pages#	Not relevant publication type
Baer, R. J., Currier, R. J., Norton, M. E., Flessel, M. C., Goldman, S., Towner, D., Jelliffe-Pawlowski, L. L.. Obstetric, perinatal, and fetal outcomes in pregnancies with false-positive integrated screening results. *Obstetrics & Gynecology.* 2014. 123:603-9	Not intervention or test of interest
Baer, R. J., Currier, R. J., Norton, M. E., Flessel, M. C., Goldman, S., Towner, D., Jelliffe-Pawlowski, L. L.. Outcomes of pregnancies with more than one positive prenatal screening result in the first or second trimester. *Prenatal Diagnosis.* 2015. 35:1223-31	Not appropriate population
Baer, R. J., Flessel, M. C., Jelliffe-Pawlowski, L. L., Goldman, S., Hudgins, L., Hull, A. D., Norton, M. E., Currier, R. J.. Detection Rates for Aneuploidy by First-Trimester and Sequential Screening. *Obstetrics & Gynecology.* 2015. 126:753-9	Not intervention or test of interest
Balslev-Harder, M., Richter, S. R., Kjaergaard, S., Johansen, P.. Correlation between Z score, fetal fraction, and sequencing reads in non-invasive prenatal testing. *Prenatal Diagnosis.* 2017. 37:943-945	Not intervention or test of interest
Barrett, A. N., Xiong, L., Tan, T. Z., Advani, H. V., Hua, R., Laureano-Asibal, C., Soong, R., Biswas, A., Nagarajan, N., Choolani, M.. Measurement of fetal fraction in cell-free DNA from maternal plasma using a panel of insertion/deletion polymorphisms. *PLoS ONE [Electronic Resource].* 2017. 12:e0186771	Not intervention or test of interest
Barringer, S. N., Sandlin, A. T., Magann, E. F.. Non-Invasive Prenatal Testing (NIPT) in Arkansas: Prenatal Genetics Clinic Experience and Lessons Learned. *Journal of the Arkansas Medical Society.* 2016. 113:38-40, 42	Not appropriate population
Basaran, S., Has, R., Kalelioglu, I. H., Karaman, B., Kirgiz, M., Dehgan, T., Satkin, B. N., Sivrikoz, T. S., Yuksel, A.. Follow-Up Studies of cf-DNA Testing from 101 Consecutive Fetuses and Related Ultrasound Findings. *Ultraschall in der Medizin.* 2018. 25:25	Not appropriate setting or country
Bayindir, B., Dehaspe, L., Brison, N., Brady, P., Ardui, S., Kammoun, M., Van der Veken, L., Lichtenbelt, K., Van den Bogaert, K., Van Houdt, J., Peeters, H., Van Esch, H., de Ravel, T., Legius, E., Devriendt, K., Vermeesch, J. R.. Noninvasive prenatal testing using a novel analysis pipeline to screen for all autosomal fetal aneuploidies improves pregnancy management. *European Journal of Human Genetics.* 2015. 23:1286-93	Not appropriate population

Reference	Reason for Exclusion
Bayon, J. C., Orruno, E., Portillo, M. I., Asua, J.. The consequences of implementing non-invasive prenatal testing with cell-free foetal DNA for the detection of Down syndrome in the Spanish National Health Service: a cost-effectiveness analysis. *Cost Effectiveness & Resource Allocation.* 2019. 17:6	Not appropriate setting or country
Beamon, C. J., Hardisty, E. E., Harris, S. C., Vora, N. L.. A single center's experience with noninvasive prenatal testing. *Genetics in Medicine.* 2014. 16:681-7	Not appropriate population
Beck, V., Opdekamp, S., Enzlin, P., Done, E., Gucciardo, L., El Handouni, N., van Mieghem, T., Lewi, L., Deprest, J.. Psychosocial aspects of invasive fetal therapy as compared to prenatal diagnosis and risk assessment. *Prenatal Diagnosis.* 2013. 33:334-40	Not intervention or test of interest
Becker, D. A., Tang, Y., Jacobs, A. P., Biggio, J. R., Edwards, R. K., Subramaniam, A.. Sensitivity of prenatal ultrasound for detection of trisomy 18. *Journal of Maternal-Fetal & Neonatal Medicine.* 2018. #volume#:1-7	Not appropriate population
Belloin, C., Jacquemard, F., Bernabé-Dupont, C., Viot, G., Lohmann, L., Grangé, G.. The noninvasive prenatal testing for Down's Syndrome. Retrospective study of 8821 patients. *Revue Sage - Femme.* 2017. 16:243-249	Not in English
Benachi, A., Letourneau, A., Kleinfinger, P., Senat, M. V., Gautier, E., Favre, R., Bidat, L., Houfflin-Debarge, V., Bouyer, J., Costa, J. M., Collaborative, SEquencage a Haut Debit et Aneuploidies Study Group. Cell-free DNA analysis in maternal plasma in cases of fetal abnormalities detected on ultrasound examination. *Obstetrics & Gynecology.* 2015. 125:1330-7	Not appropriate population
Bestwick, J. P., Wald, N. J.. Antenatal reflex DNA screening for trisomy 18 and trisomy 13 in addition to Down's syndrome. *Journal of Medical Screening.* 2016. 23:171-174	Not appropriate population
Beulen, L., Faas, B. H. W., Feenstra, I., van Vugt, J. M. G., Bekker, M. N.. Clinical utility of non-invasive prenatal testing in pregnancies with ultrasound anomalies. *Ultrasound in Obstetrics & Gynecology.* 2017. 49:721-728	Not appropriate population
Beulen, L., Faas, B. H. W., Feenstra, I., Van Vugt, J. M. G., Bekker, M. N.. Clinical Utility of Noninvasive Prenatal Testing in Pregnancies with Ultrasound Anomalies. *Obstetrical and Gynecological Survey.* 2017. 72:640-642	Not relevant publication type
Beulen, L., Grutters, J. P. C., Faas, B. H., Feenstra, I., Van Vugt, J. M. G., Bekker, M. N.. The consequences of implementing non-invasive prenatal testing in Dutch National Health Care: A cost-effectiveness analysis: Editorial comment. *Obstetrical and Gynecological Survey.* 2015. 70:162-164	Not relevant publication type
Beulen, L., Grutters, J. P., Faas, B. H., Feenstra, I., van Vugt, J. M., Bekker, M. N.. The consequences of implementing non-invasive prenatal testing in Dutch national health care: a cost-effectiveness analysis. *European Journal of Obstetrics, Gynecology, & Reproductive Biology.* 2014. 182:53-61	Not appropriate setting or country
Bevilacqua, E., Gil, M. M., Nicolaides, K. H., Ordonez, E., Cirigliano, V., Dierickx, H., Willems, P. J., Jani, J. C.. Performance of screening for aneuploidies by cell-free DNA analysis of maternal blood in twin pregnancies. *Ultrasound in Obstetrics & Gynecology.* 2015. 45:61-6	Not appropriate population
Bevilacqua, E., Jani, J. C., Letourneau, A., Duiella, S. F., Kleinfinger, P., Lohmann, L., Resta, S., Cos Sanchez, T., Fils, J. F., Mirra, M., Benachi, A., Costa, J. M.. Cell-Free DNA Analysis in Maternal Blood: Differences in Estimates between Laboratories with Different Methodologies Using a Propensity Score Approach. *Fetal Diagnosis & Therapy.* 2019. 45:302-311	Not appropriate population
Bevilacqua, E., Ordonez, E., Hurtado, I., Rueda, L., Mazzone, E., Cirigliano, V., Jani, J. C.. Screening for Sex Chromosome Aneuploidy by Cell-Free DNA Testing: Patient Choice and Performance. *Fetal Diagnosis & Therapy.* 2018. 44:98-104	Not appropriate population

Reference	Reason for Exclusion
Bevilacqua, E., Resta, S., Carlin, A., Kang, X., Cos Sanchez, T., de Marchin, J., Jani, J. C.. Profile of women choosing the Harmony Prenatal Test. *Expert Review of Molecular Diagnostics.* 2018. 18:591-599	Not appropriate population
Bianchi, D. W., Parsa, S., Bhatt, S., Halks-Miller, M., Kurtzman, K., Sehnert, A. J., Swanson, A.. Fetal sex chromosome testing by maternal plasma DNA sequencing: clinical laboratory experience and biology. *Obstetrics & Gynecology.* 2015. 125:375-82	Not appropriate population
Bianchi, D. W., Platt, L. D., Goldberg, J. D., Abuhamad, A. Z., Sehnert, A. J., Rava, R. P., MatErnal, BLood I. S. Source to Accurately diagnose fetal aneuploidy Study Group. Genome-wide fetal aneuploidy detection by maternal plasma DNA sequencing. *Obstetrics & Gynecology.* 2012. 119:890-901	Not appropriate population
Bianchi, D. W., Prosen, T., Platt, L. D., Goldberg, J. D., Abuhamad, A. Z., Rava, R. P., Sehnert, A. J., MatErnal, BLood I. S. Source to Accurately diagnose fetal aneuploidy Study Group. Massively parallel sequencing of maternal plasma DNA in 113 cases of fetal nuchal cystic hygroma. *Obstetrics & Gynecology.* 2013. 121:1057-62	Not appropriate population
Bianchi, D. W.. Cherchez la femme: Maternal Incidental Findings Can Explain Discordant Prenatal Cell-free DNA Sequencing Results. *Obstetrical and Gynecological Survey.* 2019. 74:72-75	Not relevant publication type
Birko, S., Ravitsky, V., Dupras, C., Le Clerc-Blain, J., Lemoine, M. E., Affdal, A. O., Haidar, H., Laberge, A. M.. The value of non-invasive prenatal testing: preferences of Canadian pregnant women, their partners, and health professionals regarding NIPT use and access. *BMC Pregnancy & Childbirth.* 2019. 19:22	Not outcomes of interest
Bjerregaard, L., Stenbakken, A. B., Andersen, C. S., Kristensen, L., Jensen, C. V., Skovbo, P., Sorensen, A. N.. The rate of invasive testing for trisomy 21 is reduced after implementation of NIPT. *Danish Medical Journal.* 2017. 64:#pages#	Not appropriate population
Blais, J., Giroux, S., Caron, A., Clement, V., Dionne-Laporte, A., Jouan, L., Gauthier, J., MacLeod, T., Moore, R., Parker, J., Swanson, L., Zhao, Y., Rouleau, G., Karsan, A., Langlois, S., Rousseau, F.. Non-invasive prenatal aneuploidy testing: Critical diagnostic performance parameters predict sample z-score values. *Clinical Biochemistry.* 2018. 59:69-77	Not intervention or test of interest
Borruto, F., Treisser, A., Abdelkrim, S. B., Comparetto, C.. The end of amniocentesis? From TriTest to PrenaTest™. *Donald School Journal of Ultrasound in Obstetrics and Gynecology.* 2013. 7:213-218	Not relevant publication type
Brar, H., Wang, E., Struble, C., Musci, T. J., Norton, M. E.. The fetal fraction of cell-free DNA in maternal plasma is not affected by a priori risk of fetal trisomy. *Journal of Maternal-Fetal & Neonatal Medicine.* 2013. 26:143-5	Not appropriate population
Breman, A. M., Chow, J. C., U'Ren, L., Normand, E. A., Qdaisat, S., Zhao, L., Henke, D. M., Chen, R., Shaw, C. A., Jackson, L., Yang, Y., Vossaert, L., Needham, R. H., Chang, E. J., Campton, D., Werbin, J. L., Seubert, R. C., Van den Veyver, I. B., Stilwell, J. L., Kaldjian, E. P., Beaudet, A. L.. Evidence for feasibility of fetal trophoblastic cell-based noninvasive prenatal testing. *Prenatal Diagnosis.* 2016. 36:1009-1019	Not intervention or test of interest
Breman, A. M., Chow, J. C., U'Ren, L., Normand, E. A., Qdaisat, S., Zhao, L., Henke, D. M., Chen, R., Shaw, C. A., Jackson, L., Yang, Y., Vossaert, L., Needham, R. H. V., Chang, E. J., Campton, D., Werbin, J. L., Seubert, R. C., Van Den Veyver, I. B., Stilwell, J. L., Kaldjian, E. P., Beaudet, A. L.. Evidence for Feasibility of Fetal Trophoblastic Cell-Based Noninvasive Prenatal Testing. *Obstetrical and Gynecological Survey.* 2017. 72:4-5	Not relevant publication type

Reference	Reason for Exclusion
Brison, N., Neofytou, M., Dehaspe, L., Bayindir, B., Van Den Bogaert, K., Dardour, L., Peeters, H., Van Esch, H., Van Buggenhout, G., Vogels, A., de Ravel, T., Legius, E., Devriendt, K., Vermeesch, J. R.. Predicting fetoplacental chromosomal mosaicism during non-invasive prenatal testing. *Prenatal Diagnosis.* 2018. 38:258-266	Not appropriate population
Brison, N., Van Den Bogaert, K., Dehaspe, L., van den Oever, J. M., Janssens, K., Blaumeiser, B., Peeters, H., Van Esch, H., Van Buggenhout, G., Vogels, A., de Ravel, T., Legius, E., Devriendt, K., Vermeesch, J. R.. Accuracy and clinical value of maternal incidental findings during noninvasive prenatal testing for fetal aneuploidies. *Genetics in Medicine.* 2017. 19:306-313	Not outcomes of interest
Byers, H. M., Neufeld-Kaiser, W., Chang, E. Y., Tsuchiya, K., Oehler, E. S., Adam, M. P.. Discordant sex between fetal screening and postnatal phenotype requires evaluation. *Journal of Perinatology.* 2019. 39:28-33	Not appropriate study design
Calabrese, G., Baldi, M., Fantasia, D., Sessa, M. T., Kalantar, M., Holzhauer, C., Alunni-Fabbroni, M., Palka, G., Sitar, G.. Detection of chromosomal aneuploidies in fetal cells isolated from maternal blood using single-chromosome dual-probe FISH analysis. *Clinical Genetics.* 2012. 82:131-9	Not intervention or test of interest
Calabrese, G., Fantasia, D., Alfonsi, M., Morizio, E., Celentano, C., Guanciali Franchi, P., Sabbatinelli, G., Palka, C., Benn, P., Sitar, G.. Aneuploidy screening using circulating fetal cells in maternal blood by dual-probe FISH protocol: a prospective feasibility study on a series of 172 pregnant women. *Molecular Genetics & Genomic Medicine.* 2016. 4:634-640	Not intervention or test of interest
Canick, J. A., Kloza, E. M., Lambert-Messerlian, G. M., Haddow, J. E., Ehrich, M., van den Boom, D., Bombard, A. T., Deciu, C., Palomaki, G. E.. DNA sequencing of maternal plasma to identify Down syndrome and other trisomies in multiple gestations. *Prenatal Diagnosis.* 2012. 32:730-4	Not appropriate population
Chai, H., DiAdamo, A., Grommisch, B., Boyle, J., Amato, K., Wang, D., Wen, J., Li, P.. Integrated FISH, Karyotyping and aCGH Analyses for Effective Prenatal Diagnosis of Common Aneuploidies and Other Cytogenomic Abnormalities. *Medical Sciences.* 2019. 7:23	Not appropriate population
Chan, N., Smet, M. E., Sandow, R., da Silva Costa, F., McLennan, A.. Implications of failure to achieve a result from prenatal maternal serum cell-free DNA testing: a historical cohort study. *BJOG: An International Journal of Obstetrics & Gynaecology.* 2018. 125:848-855	Not appropriate population
Chan, N., Smet, M. E., Sandow, R., Da Silva Costa, F., McLennan, A.. Implications of Failure to Achieve a Result from Prenatal Maternal Serum Cell-Free DNA Testing: A Historical Cohort Study. *Obstetrical and Gynecological Survey.* 2018. 73:611-613	Not relevant publication type
Chan, Y. M., Leung, W. C., Chan, W. P., Leung, T. Y., Cheng, Y. K., Sahota, D. S.. Women's uptake of non-invasive DNA testing following a high-risk screening test for trisomy 21 within a publicly funded healthcare system: findings from a retrospective review. *Prenatal Diagnosis.* 2015. 35:342-7	Not appropriate population
Chen, A., Tenhunen, H., Torkki, P., Heinonen, S., Lillrank, P., Stefanovic, V.. Considering medical risk information and communicating values: A mixed-method study of women's choice in prenatal testing. *PLoS ONE [Electronic Resource].* 2017. 12:e0173669	Not appropriate population
Chen, E. Z., Chiu, R. W., Sun, H., Akolekar, R., Chan, K. C., Leung, T. Y., Jiang, P., Zheng, Y. W., Lun, F. M., Chan, L. Y., Jin, Y., Go, A. T., Lau, E. T., To, W. W., Leung, W. C., Tang, R. Y., Au-Yeung, S. K., Lam, H., Kung, Y. Y., Zhang, X., van Vugt, J. M., Minekawa, R., Tang, M. H., Wang, J., Oudejans, C. B., Lau, T. K., Nicolaides, K. H., Lo, Y. M.. Noninvasive prenatal diagnosis of fetal trisomy 18 and trisomy 13 by maternal plasma DNA sequencing. *PLoS ONE [Electronic Resource].* 2011. 6:e21791	Not intervention or test of interest

Reference	Reason for Exclusion
Chen, Y. P., He, Z. Q., Shi, Y., Zhou, Q., Cai, Z. M., Yu, B., Wang, T.. Not all chromosome aberrations can be detected by NIPT in women at advanced maternal age: A multicenter retrospective study. *Clinica Chimica Acta.* 2018. 486:232-236	Not appropriate setting or country
Chen, Y., Lai, Y., Yi, S., Tang, Y., Lei, Y., Yi, S., Shen, Y., Wei, H.. Non-invasive prenatal screening for fetal aneuploidy in twin pregnancies by cell-free DNA analysis. *Clinical and Experimental Obstetrics and Gynecology.* 2018. 45:656-660	Not appropriate setting or country
Cheng, S. S. W., Chan, K. Y. K., Leung, K. K. P., Au, P. K. C., Tam, W. K., Li, S. K. M., Luk, H. M., Kan, A. S. Y., Chung, B. H. Y., Lo, I. F. M., Tang, M. H. Y.. Experience of chromosomal microarray applied in prenatal and postnatal settings in Hong Kong. *American Journal of Medical Genetics. Part C, Seminars in Medical Genetics.* 2019. 181:196-207	Not appropriate population
Cheng, Y., Leung, W. C., Leung, T. Y., Choy, K. W., Chiu, R., Lo, T. K., Kwok, K. Y., Sahota, D. S.. Women's preference for non-invasive prenatal DNA testing versus chromosomal microarray after screening for Down syndrome: a prospective study. *BJOG: An International Journal of Obstetrics & Gynaecology.* 2018. 125:451-459	Not appropriate setting or country
Chetty, S., Garabedian, M. J., Norton, M. E.. Uptake of noninvasive prenatal testing (NIPT) in women following positive aneuploidy screening. *Prenatal Diagnosis.* 2013. 33:542-6	Not appropriate population
Chitty, L. S., Wright, D., Hill, M., Verhoef, T. I., Daley, R., Lewis, C., Mason, S., McKay, F., Jenkins, L., Howarth, A., Cameron, L., McEwan, A., Fisher, J., Kroese, M., Morris, S.. Uptake, outcomes, and costs of implementing non-invasive prenatal testing for Down's syndrome into NHS maternity care: prospective cohort study in eight diverse maternity units. *BMJ.* 2016. 354:i3426	Not appropriate population
Chitty, L. S., Wright, D., Hill, M., Verhoef, T. I., Daley, R., Lewis, C., Mason, S., McKay, F., Jenkins, L., Howarth, A., et al., . Uptake, outcomes, and costs of implementing non-invasive prenatal testing for down syndrome into NHS maternity care: prospective cohort study in eight diverse maternity units. *Obstetrical & gynecological survey.* 2016. 71:637-639	Not relevant publication type
Chiu, R. W., Akolekar, R., Zheng, Y. W., Leung, T. Y., Sun, H., Chan, K. C., Lun, F. M., Go, A. T., Lau, E. T., To, W. W., Leung, W. C., Tang, R. Y., Au-Yeung, S. K., Lam, H., Kung, Y. Y., Zhang, X., van Vugt, J. M., Minekawa, R., Tang, M. H., Wang, J., Oudejans, C. B., Lau, T. K., Nicolaides, K. H., Lo, Y. M.. Non-invasive prenatal assessment of trisomy 21 by multiplexed maternal plasma DNA sequencing: large scale validity study. *BMJ.* 2011. 342:c7401	Not appropriate population
Chiu, R. W., Chan, K. C., Gao, Y., Lau, V. Y., Zheng, W., Leung, T. Y., Foo, C. H., Xie, B., Tsui, N. B., Lun, F. M., Zee, B. C., Lau, T. K., Cantor, C. R., Lo, Y. M.. Noninvasive prenatal diagnosis of fetal chromosomal aneuploidy by massively parallel genomic sequencing of DNA in maternal plasma. *Proceedings of the National Academy of Sciences of the United States of America.* 2008. 105:20458-63	Not intervention or test of interest
Chiu, R. W., Sun, H., Akolekar, R., Clouser, C., Lee, C., McKernan, K., Zhou, D., Nicolaides, K. H., Lo, Y. M.. Maternal plasma DNA analysis with massively parallel sequencing by ligation for noninvasive prenatal diagnosis of trisomy 21. *Clinical Chemistry.* 2010. 56:459-63	Not intervention or test of interest
Chu, T., Bunce, K., Hogge, W. A., Peters, D. G.. Statistical considerations for digital approaches to non-invasive fetal genotyping. *Bioinformatics.* 2010. 26:2863-6	Not appropriate study design
Chu, T., Shaw, P. A., Yeniterzi, S., Dunkel, M., Rajkovic, A., Hogge, W. A., Bunce, K. D., Peters, D. G.. Comparative evaluation of the Minimally-Invasive Karyotyping (MINK) algorithm for non-invasive prenatal testing. *PLoS ONE [Electronic Resource].* 2017. 12:e0171882	Not appropriate population
Cicatiello, R., Pignataro, P., Izzo, A., Mollo, N., Pezone, L., Maruotti, G. M., Sarno, L., Sglavo, G., Conti, A., Genesio, R., Nitsch, L.. Chromosomal Microarray Analysis versus Karyotyping in Fetuses with Increased Nuchal Translucency. *Medical Sciences.* 2019. 7:27	Not appropriate population

Reference	Reason for Exclusion
Cirigliano, V., Ordonez, E., Rueda, L., Syngelaki, A., Nicolaides, K. H.. Performance of the neoBona test: a new paired-end massively parallel shotgun sequencing approach for cell-free DNA-based aneuploidy screening. *Ultrasound in Obstetrics & Gynecology.* 2017. 49:460-464	Not intervention or test of interest
Cirigliano, V., Voglino, G., Ordonez, E., Marongiu, A., Paz Canadas, M., Ejarque, M., Rueda, L., Lloveras, E., Fuster, C., Adinolfi, M.. Rapid prenatal diagnosis of common chromosome aneuploidies by QF-PCR, results of 9 years of clinical experience. *Prenatal Diagnosis.* 2009. 29:40-9	Not appropriate population
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