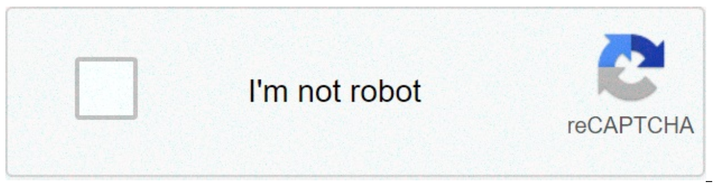

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How to read congenital anomaly scan

An anomaly scan takes a close look at your baby and your womb (uterus) at about 20 weeks of pregnancy. The person carrying out the scan (sonographer) will check that your baby is developing normally. Reasons to have this scan Reassure you that your baby is developing normally. Confirm the gestational age of your pregnancy. Confirm the number of fetuses and, if twins, whether they are identical or not. Detect birth defects, such as a spina bifida or heart problems. If you are concerned about the chances of chromosome issues, this scan can search for subtle markers that may suggest a higher risk that your baby may have a syndrome. If you want to know your baby's gender this can usually be seen at this scan. When you attend for this scan we will tell you about everything that we see, unless you advise us that there are certain things that you don't want to know about, such as your baby's gender or markers for chromosome issues. This ultrasound scan is very accurate but unfortunately it cannot diagnose 100% of congenital abnormalities. If the scan is complete, we would expect to pick up at least 95% of cases of spina bifida, 80% of cases of cleft lip or palate, and 60% to 70% of cases of congenital heart disease. This scan can also identify 50% to 70% of cases of Down syndrome, but only an amniocentesis can give you this information for certain. It is also important to realise that ultrasound scans in pregnancy do not detect problems like cerebral palsy or autism. Sometimes babies with chromosomal abnormalities have signs called ultrasound markers. These include thick skin in the neck, excess fluid in the kidneys, short arms or legs, white spots in the baby's heart or abdomen, or choroid plexus cysts in the brain. While some babies with chromosomal abnormalities have these markers, it is important to remember that many normal babies also have these signs. If the scan suggests a problem, you will be told this immediately. You will be able to discuss the findings immediately with a consultant who specialises in fetal medicine. The only way to diagnose or exclude a chromosomal problem for certain is to have an amniocentesis. If you would prefer not to know about these markers please inform us prior to the scan. A full support service will be available for you should any issue be detected, including a referral to an appropriate pediatrician. A copy of your report will be sent to your referring hospital, doctor or midwife to ensure good communication. In case of a twin pregnancy the price is €300, since the baby is examined head to toe and scan will take longer as it is essentially two anatomy reviews. Price Anomaly scan price is € 200/300 Working off-campus? Learn about our remote access options Volume 51, Issue 4 p. 463-469 This article has been selected for Journal Club. Click here to view slides and discussion points. en The main aim of this study was to assess the proportion and type of congenital anomalies, both structural and chromosomal, that can be detected at an early scan performed at 12–13 weeks' gestation, compared with at the 20-week structural anomaly scan offered under the present screening policy. Secondary aims were to evaluate the incidence of false-positive findings and ultrasound markers at both scans, and parental choice regarding termination of pregnancy (TOP). Sonographers accredited for nuchal translucency (NT) measurement were asked to participate in the study after undergoing additional training to improve their skills in late first-trimester fetal anatomy examination. The early scans were performed according to a structured protocol, in six ultrasound practices and two referral centers in the north-east of The Netherlands. All women opting for the combined test (CT) or with an increased a-priori risk of fetal anomalies were offered a scan at 12–13 weeks' gestation (study group). All women with a continuing pregnancy were offered, as part of the 'usual care', a 20-week anomaly scan. The study group consisted of 5237 women opting for the CT and 297 women with an increased a-priori risk of anomalies (total, 5534). In total, 51 structural and 34 chromosomal anomalies were detected prenatally in the study population, and 18 additional structural anomalies were detected after birth. Overall, 54/85 (63.5%) anomalies were detected at the early scan (23/51 (45.1%) structural and all chromosomal anomalies presenting with either an increased risk at first-trimester screening or structural anomalies (31/34)). All particularly severe anomalies were detected at the early scan (all cases of neural tube defect, omphalocele, megacystis, and multiple severe congenital and severe skeletal anomalies). NT was increased in 12/23 (52.2%) cases of structural anomaly detected at the early scan. Of the 12 cases of heart defects, four (33.3%) were detected at the early scan, five (41.7%) at the 20-week scan and three (25.0%) after birth. False-positive diagnoses at the early scan and at the 20-week scan occurred in 0.1% and 0.6% of cases, respectively, whereas ultrasound markers were detected in 1.4% and 3.0% of cases, respectively. After first- or second-trimester diagnosis of an anomaly, parents elected TOP in 83.3% and 25.8% of cases, respectively. An early scan performed at 12–13 weeks' gestation by a competent sonographer can detect about half of the prenatally detectable structural anomalies and 100% of those expected to be detected at this stage. Particularly severe anomalies, often causing parents to choose TOP, are amenable to early diagnosis. The early scan is an essential part of modern pregnancy care. Copyright © 2017 ISUOG. Published by John Wiley & Sons Ltd. es Eficacia de la ecografía de 12–13 semanas para el diagnóstico temprano de anomalías congénitas fetales en la era del ADN fetal El objetivo principal de este estudio fue evaluar la proporción y el tipo de anomalías congénitas, tanto estructurales como cromosómicas, que se pueden detectar en una ecografía temprana realizada a las 12–13 semanas de gestación, en comparación con la ecografía a las 20 semanas para detectar anomalías estructurales que se ofrece bajo la política de cribado actual. Los objetivos secundarios fueron evaluar la incidencia de falsos positivos y de marcadores ecográficos en ambas ecografías, y la elección de los padres con respecto a la interrupción legal del embarazo (ILE). Se invitó a especialistas en técnicas de ecografía acreditados(as) para la medición de la translucencia nucal (TN) a que participasen en el estudio, después de recibir formación adicional para mejorar su experiencia en el examen de la anatomía fetal de finales del primer trimestre. Las ecografías tempranas se realizaron bajo un protocolo estructurado en seis clínicas de ultrasonido y dos centros de especialistas en el noreste de los Países Bajos. A todas las mujeres que optaron por la prueba combinada (PC) o con un mayor riesgo a priori de anomalías fetales, se les ofreció una ecografía a las 12–13 semanas de gestación (grupo de estudio). A todas las mujeres que continuaron con la gestación, se les ofreció, como parte del 'cuidado habitual', una ecografía a las 20 semanas para detectar posibles anomalías. El grupo de estudio consistió en 5237 mujeres que optaron por la PC y 297 mujeres con un mayor riesgo a priori de anomalías (5534 en total). En total, se detectaron 51 anomalías prenatales estructurales y 34 cromosómicas en la población de estudio y 18 anomalías estructurales adicionales después del nacimiento. En total, se detectaron 54/85 (63.5%) anomalías en la ecografía temprana (23/51 (45.1%) anomalías estructurales y todas las cromosómicas que presentaban o un mayor riesgo en el cribado del primer trimestre o anomalías estructurales (31/34)). En la ecografía temprana se detectaron todas las anomalías particularmente graves (todos los casos con defectos del tubo neural, onfalocele, megavejiga y anomalías múltiples graves congénitas o del esqueleto). La TN se incrementó en 12/23 (52.2%) casos de anomalía estructural detectada en la ecografía temprana. De los 12 casos de defectos cardíacos, cuatro (33.3%) se detectaron en la ecografía temprana, cinco (41.7%) en la ecografías de 20 semanas y tres (25.0%) después del nacimiento. Los falsos positivos en la ecografía temprana y en la ecografía de 20 semanas ocurrieron en el 0,1% y el 0,6% de los casos, respectivamente, mientras que los marcadores ecográficos se detectaron en el 1,4% y 3,0% de los casos, respectivamente. Después del diagnóstico de una anomalía en el primer o segundo trimestre, los padres eligieron la ILE en el 83,3% y 25,8% de los casos, respectivamente. Una ecografía temprana realizada entre las 12–13 semanas de gestación por especialistas competentes en ecografía puede detectar aproximadamente la mitad de las anomalías estructurales prenatales detectables y el 100% de las que se espera que se detecten en esta etapa. Las anomalías particularmente graves, que a menudo provocan que los padres elijan la ILE, son susceptibles de un diagnóstico temprano. La ecografía temprana es una parte esencial de la atención moderna del embarazo. zh 在游离DNA时代通过孕12~13周扫描早期诊断胎儿先天畸形的有效性 本研究的主要目的是评估孕12~13周早期扫描能够发现的先天异常(结构和染色体)的比例和类型,并与目前筛查政策下20周结构异常扫描进行比较。次要目的是评估假阳性结果发生率和2次扫描的超声标志物,以及父母对终止妊娠(termination of pregnancy,TOP)的选择。 参与本研究的超声医师均具备颈项透明层(nuchal translucency,NT)测量技能认证,在接受额外培训提高孕早期末胎儿解剖检查技能后,在荷兰东北部的6个超声检查部门和2个转诊中心,根据结构化方案进行早期扫描。所有选择进行联合检测(combined test,CT)或胎儿畸形先验风险增加的孕妇在孕12~13周进行扫描(研究组)。所有继续妊娠的孕妇接受孕20周畸形扫描(“常规检查”的一部分)。 研究组包括选择CT检查的5237例孕妇和畸形先验风险升高的297例孕妇(共5534例)。研究人群中产前检查共发现51例结构异常和34例染色体异常,出生后又另外发现18例结构异常。共63.5%(54/85)的异常在早期扫描时发现,45.1%(23/51)的结构异常和所有的染色体异常表现为孕早期筛查风险增加或结构异常(31/34)。早期扫描发现所有特别严重的畸形(所有神经管缺陷、脐膨出、巨膀胱、多发严重先天性和严重骨骼畸形病例)。52.2%(12/23)的早期扫描发现的结构异常病例中NT增厚。12例心脏缺陷病例中,4例(33.3%)在早期扫描时发现,5例(41.7%)在孕20周扫描时发现,3例(25.0%)在出生后发现。早期扫描和孕20周扫描时诊断假阳性率分别为0.1%和0.6%,然而分别 在1.4%和3.0%的病例中发现超声标志物。在孕早期和孕中期诊断畸形后,分别有83.3%和25.8%的病例父母选择终止妊娠。 由经验丰富的超声医生在孕12~13周进行早期扫描,能够发现近一半的产前可检测的结构异常,人们希望在这一阶段发现所有畸形。应该早期诊断特别严重的畸形,父母常因此选择终止妊娠。早期扫描是现代产科保健必不可少的一部分。 Ten years after the introduction of the Dutch prenatal screening program aimed at increasing the reproductive choices of parents with a diagnosis of fetal anomaly1, 2, a marked difference in the uptake of the two screening methods has been noted. The combined test (CT) has an uptake of around 30% and the 20-week scan an uptake of around 95%. The CT is not free of charge and is offered exclusively as a screening test for aneuploidies3. The low uptake is thought to reflect a general mistrust in the performance of the test and a high acceptance of Down syndrome (trisomy 21), which is not considered by many parents as a reason to terminate a pregnancy3, 4. Recently, the Dutch Ministry of Health agreed to offer non-invasive whole-genome sequencing (cell-free DNA (cfDNA) testing) to all pregnant women as first-tier screening for trisomies5, as an alternative to the CT. This raises the issue as to whether an early scan should also be offered to all pregnant women. Although early scans are performed for dating, location of pregnancy and the diagnosis of multiple pregnancy6-8, there is compelling evidence that diagnosis of structural anomalies is also possible from the late first trimester9. It is, therefore, necessary to reappraise the role of an early scan in screening for congenital anomalies10. The primary aim of this study was to examine the proportion and type of congenital anomalies, both chromosomal and structural, detected by an early scan (at 12–13 weeks' gestation; new strategy), compared with the 20-week scan (current strategy). Secondary aims were to evaluate the incidence of false-positive findings and ultrasound markers at both scans, and parental choice regarding termination of pregnancy (TOP). Two referral centers (University Medical Centre in Groningen and Isala Hospital in Zwolle) and six ultrasound practices in the provinces of Groningen and Overijssel participated in the study. Only sonographers accredited for nuchal translucency (NT) measurement and performing at least 100 NT scans per year were eligible to participate in the study. A special license was obtained for the study from the Ministry of Health, within the Dutch Population Screening Act11, regulating screening for incurable diseases. Women opting for the CT were asked by their referring midwife to participate in the study, and received an information leaflet and an informed consent form. Participants were recruited between November 2012 and December 2015. The scans were performed transabdominally, unless the transvaginal route was preferred or considered necessary. Prior to commencement of the study, all sonographers underwent training to improve their skills in late first-trimester fetal anatomy examination. The anatomical survey was performed following a structured protocol, aimed at excluding or diagnosing severe anomalies considered 'not to be missed' along with other severe, but less obvious, anomalies (Table 1). Table 1. Protocol followed for early anatomical survey indicating views that should be obtained, structures that should be investigated and measurements that should be taken in order to exclude or detect all anomalies that should be seen at an early scan Longitudinal view/examined structure [measurement] Axial/tangential view [measurement] Anomaly Head Evaluation of skull contour Visualization of midline Acrania/anencephaly* [Nuchal translucency] [BPD, HC] Holoprosencephaly (alobar)* Profile Symmetry of choroid plexus Exencephaly containing brain tissue* Nasal bone Visualization of orbits Micrognathia, cleft lip and palate† Intracranial translucency Retronasal triangle, lips Micro-anophthalmia† Large nuchal translucency/cystic hygroma* Body Diaphragm Umbilical cord insertion Large omphalocele* Stomach (below diaphragm) Umbilical arteries along bladder Gastroschisis† Bladder filling Diaphragmatic hernia (stomach in thorax)† Megacystis > 7 mm* Abdominal cysts Spine Shape, closure Scoliosis† Open spina bifida with myelomeningocele† Sacral agenesis† Sacro-occygeal teratoma† Limbs Count of long-bone segments [Femur length] Part of or whole limb missing† Position of feet and hands Clubfoot† , syndactyly† , polydactyly† Heart Heart on same side of stomach Equal size of chambers and heart-chamber filling (color/power Doppler) Crossing of outflow tracts (color/power Doppler) Single ventricle* Persisting bradycardia (heart block) Abnormal chambers, outflows† Other Excessive fluid: generalized hydrops/severe hydrothorax/severe pericardial effusion* Severe anomalies* : Severe skeletal dysplasia Syrenomelia Conjoined twins Amniotic band syndrome Body-stalk anomaly * Severe anomaly considered 'not to be missed' at early scan. † Anomaly potentially detectable at early scan. BPD, biparietal diameter; HC, head circumference. In addition to women opting for CT, all women with an increased a-priori risk of congenital anomalies (e.g. positive family history, diabetes, use of teratogenic drugs), who are usually referred to a fetal medicine unit for detailed ultrasound examination, were offered the early 12–13-week scan and enrolled in the study. All women with a continuing pregnancy were offered, as part of the 'usual care', a 20-week anomaly scan. All study data were extracted from local databases (Astraia or Mosos-U). Pregnancy outcomes and information on additional investigations (e.g. genetic investigations, delivery or pathology reports) were obtained from the hospital databases, returned follow-up forms provided to patients or the ultrasound practices, well-baby clinics and referring midwives or physicians. All sonographers involved in the study kept a logbook documenting the start and end times of all early scans. For each early scan, an extra 15 min was scheduled, in addition to the 30 min allocated for the CT. Descriptive statistics on patients' characteristics and study findings, including frequencies, means, median and ranges were calculated in Excel (Microsoft Corp., Redmond, WA, USA). A total of 5534 women underwent the early scan, including 5237 women who had opted for CT and 297 with an increased a-priori risk of fetal anomaly based on their history. The early scans were performed at a mean gestational age of 12 + 5 (range, 11 + 0 to 13 + 6) weeks. Mean maternal age was 32 (range, 17–53) years. An overview of the number of early scans, 20-week scans, findings and pregnancy outcomes is given in Figure 1. The average additional time required to perform the anatomical survey in women undergoing the CT was 12 (range, 9–18) min. Flowchart showing findings and pregnancy outcome of 5534 pregnant women undergoing early scan at 12–13 weeks' gestation and 5014 attending 20-week anomaly scan. IUD, intrauterine death; NND, neonatal death; NT, nuchal translucency; TAPVR, total anomalous pulmonary venous return; TOP, termination of pregnancy. A total of 51 structural anomalies were diagnosed prenatally in chromosomally normal fetuses, 23 (45.1%) at the early scan and 28 (54.9%) at the 20-week scan. Details of the structural anomalies detected on the 12–13-week scan and the associated NT measurements are provided in Table S1. NT was increased in 12/23 (52.2%) of the cases diagnosed with a structural anomaly. Details of structural anomalies detected prenatally, time of diagnosis and pregnancy outcome are shown in Table 2. Overall, 103 (18.6/1000 participants) anomalies were diagnosed during pre- or postnatal life, comprising 34 (32.4%) chromosomal and 69 (67.0%) structural anomalies. All 34 aneuploidies in the study population were diagnosed prenatally. With respect to structural anomalies, 51 (73.9%) were diagnosed before and 18 (26.1%) after birth (Tables 2, S2 and S3). Of the 12 cases of heart defect in the study population, four (33.3%) were detected at the 12–13-week scan, five (41.7%) at the 20-week scan and three (25.0%) after birth. Table 2. Structural anomalies detected prenatally in euploid fetuses, at early scan (12–13 weeks; n = 5534 women), 20-week scan (n = 5014 women) and after birth, and pregnancy outcome Anomaly 12–13-week scan (TOP) 20-week scan (TOP) After birth IUD NND LB CNS (5) Acrania/anencephaly 3 (2) 1 SB 1 (1) SB + acrania/anencephaly 1 (1) Heart (12) Complex 1 (1) 1 (1) TGA 1 1 1 3 ToF 1 (1)* 2 (1) 1 TAPVR 1 1 VSD 1 1 1 3 Abdominal wall (3) Omphalocele 1 1 Gastroschisis 2 (1) 1 Intestinal (3) 1 2 3 MCA (4) 4 (3) 1 Skeletal (4) Severe 1 (1) 1 (1) Less severe 1 1 2 Hydrops (3) 1 (1) 2 (1) 1 Kidneys/high urinary tract (7) 1 (1) 6 (2) 4 Bladder/low urinary tract (3) 3 (2) 1 Clubfoot (8) 6 2 8 Schisis (5) 4 1† 5 Genetic syndrome (5) 2 (1) 3‡ 1 3 Other (7) 7 7 Total 23 (15) 28 (7) 18 2 3 42 * 22q11 deletion. † Only lip. ‡ One case of Klippel–Trenaunay syndrome. CNS, central nervous system; IUD, intrauterine death; LB, live birth; MCA, multiple congenital anomalies; NND, neonatal death; SB, spina bifida; TAPVR, total anomalous pulmonary venous return; TGA, transposition of great arteries; ToF, tetralogy of Fallot; TOP, termination of pregnancy; VSD, ventricular septal defect. Although care was taken in tracing all severe anomalies observed after birth, under-reporting of minor anomalies by parents or of anomalies (such as minor cardiac ones) detected after the follow-up form had been returned, cannot be excluded. Overall, the anomalies detected at the early scan were more severe than those detected at the 20-week scan. Of the anomalies 'not to be missed', all were detected at the early scan. A total of 34 chromosomal anomalies (6.1/1000 participants) were diagnosed in the study population, all diagnosed prenatally (Figure 1). In 33/34 (97.1%) of these, either the NT or the CT risk was increased (≥ 1:200), and in 31 of these 33 (91.2% of the total) the diagnosis was made in the first trimester. In the first case not diagnosed in the first trimester, the parents opted for cfDNA analysis instead of invasive testing; the anomaly (triploidy) was not identified by cfDNA analysis and was diagnosed later in pregnancy, owing to abnormal ultrasound findings prompting an amniocentesis. In the second such case, the couple had declined early invasive prenatal diagnosis, and trisomy 21 was diagnosed after an amniocentesis performed at 29 weeks' gestation, following the detection of structural anomalies. In the remaining only (1/34) case of true late diagnosis (deletion/duplication), NT was not increased and CT risk was not assessed and structural anomalies were seen only at the 20-week scan. Details of the chromosomal anomalies, observed associated structural anomalies and time of diagnosis are reported in Table 3. Table 3. Chromosomal anomalies diagnosed in study population, time of diagnosis and reason for testing (increased nuchal translucency (NT) measurement, increased risk on combined test (CT), carriership, suspicious findings on ultrasound) Reason for testing Chromosomal anomaly n NT > p95 High-risk CT Anomalies at ultrasound 12–13-week scan (n = 31) 47,XXX 1 (LB) Y N NT > 3.5 mm Trisomy 13 1 N Y Median cleft Mosaic 45,X0/46,XY 1 Y Y Large NT, hydrops Mosaic trisomy 16 1 Y Y Omphalocele, large NT, abnormal DV Unbalanced translocation chr 4/15 (parent carrier) 1 Y N Increased NT very early Triploidy 1 N Y Intracardiac focus, growth restriction Triploidy 1 N Y Asymmetrical growth restriction Trisomy 18 5 Y Y Hydrops, omphalocele, VSD, tachycardia, abnormal DV (n = 1); hydrops (n = 2); omphalocele (n = 1); omphalocele, abnormal four-chamber view (n = 1) Trisomy 18 1 Not measured Not calculated Abnormal four-chamber view Trisomy 21 16 Y Y Only increased NT (n = 4); increased NT + markers (n = 12)* Trisomy 21 2 Y Not calculated Tricuspid regurgitation (n = 1); hydrothorax (n = 1) After 20-week scan (n = 3) Deletion 2q37.3, duplication 11p15.5p15.2 1 (NND) N Not calculated Macrosomia, hypertelorism, polyhydramnios Triploidy 1 N Y Growth restriction, VSD, rocker-bottom feet† Trisomy 21 1 (LB) N Y Growth restriction, echogenic bowel‡ Total 34 LB (n = 2) Increased (n = 27) High risk (n = 28) NND (n = 1) Not increased (n = 6) Normal NT (n = 2) TOP (n = 31) Not measured (n = 1) Not calculated (n = 4) Outcome was termination of pregnancy (TOP) unless indicated otherwise. * Including two cases with additional structural anomaly (clubfoot (n = 1), atrioventricular septal defect (n = 1)). † Cell-free DNA analysis showed no increased risk. ‡ Diagnosed at 29 weeks. chr, chromosome; DV, ductus venosus; LB, live birth; N, No; NND, neonatal death; p95, 95th percentile; VSD, ventricular septal defect; Y, yes. Of the 5534 early scans and 5014 20-week scans performed, anomalies were detected but not confirmed at subsequent scans (false-positives) in five (0.1%) and 30 (0.6%) cases, respectively (Tables 4 and 5). Isolated ultrasound markers were observed at the early scan (excluding enlarged NT) and at the 20-week scan in 31 (0.6%) and in 152 (3.0%) cases, respectively (Table 6). Table 4. False-positive structural anomaly findings on 12–13-week scan in 5534 pregnant women Finding Time of initial (false) diagnosis (weeks) Time of final (normal) diagnosis (weeks) n Outcome Echogenic choroid plexus 12 + 3 13 + 2 1 LB Omphalocele 11 + 1* 15 + 2 1 LB Discrepancy in size of great vessels 12 + 5 15 + 3 1 LB Megacystis 12 + 2 14 + 2 1 LB Intra-abdominal cyst 12 + 1 19 + 3 1 LFU Total 5 * Physiological gut herniation. LB, live birth with no anomaly; LFU, lost to follow-up. Table 5. False-positive structural anomaly findings on 20-week scan in 5014 pregnant women Finding n Outcome Anal/bowel 1 LB Uncertain cardiac findings 8 LB Asymmetric brain ventricles 1 LB Miscellaneous* 16 LB Urogenital 1 LB Spine 1 LB Echogenic lungs 2 LB Total 30 * Abnormal biometry, abnormal amniotic fluid. LB, live birth with no anomaly. Table 6. Markers for structural anomalies observed at 12–13-week scan (n = 5534 women) and at 20-week scan (n = 5014 women) Marker n Outcome 12–13-week scan Single umbilical artery 19 15 LB, 4 LFU Absent nasal bone 1 1 LB Pyelectasis 3 3 LB Echogenic bowel 2 2 LB Other first-trimester markers* 6 6 LB Total 31 20-week scan Single umbilical artery 21 1 IUD, 20 LB Ventriculomegaly 8 8 LB Pyelectasis 27 27 LB† Echogenic bowel 12 12 LB Choroid plexus cyst 6 6 LB Echogenic heart focus 17 17 LB‡ Multiple markers 4 1 TOP, 3 LB Short femur 5 5 LB Marker not specified 52 11 LB, 41 LFU Total 152 * Abnormal ductus venosus flow, tricuspid regurgitation. † Pyelectasis (n = 2), congenital heart defect (n = 1). ‡ Hand anomaly (n = 1). LB, liveborn with no anomaly; LFU, lost to follow-up after birth; TOP, termination of pregnancy. In 4661 fetuses, no anomalies were observed at the 20-week scan and the pregnancy outcome was uneventful. Spontaneous pregnancy loss occurred in 37 cases (0.7%); in 21 cases, intrauterine death (IUD) occurred before the 20-week scan and involved two fetuses with structural anomaly and 19 without anomaly detected on the early scan, while in 16 cases, IUD occurred after the 20-week scan and included four fetuses with anomaly and 12 without detected anomaly (Figure 1). In 53 cases the pregnancy was terminated. This occurred in 31/34 (91.2%) cases with chromosomal anomalies and in 22/51 (43.1%) pregnancies with structural anomalies. The majority of TOP for structural anomalies followed a first-trimester diagnosis (15/22, 68.2%) of the anomaly. Overall, parents opted for TOP in 15/23 (65.2%) cases of early-diagnosed structural anomaly, and in 7/28 (25.0%) of those diagnosed at the 20-week scan. Neonatal death occurred in six cases. This study shows that an early scan impacts on the time of detection of congenital anomalies and on parental decisions. In fact, 45% of the prenatally diagnosed anomalies, including all the lethal ones, in the population could be diagnosed in the first trimester. Parents opted for TOP in 83.3% of the early-diagnosed anomalies, as opposed to in 25.8% of those diagnosed late. The incidence of false-positive results leading to unnecessary parental anxiety was 0.1% at the early scan, six times lower than that at the 20-week scan. The prevalence of anomalies detected early (1.0%) is similar to that found in another large study, suggesting no diagnostic bias6. Interestingly, of the 34 chromosomal anomalies, only one (deletion/duplication without obvious anomalies) was not diagnosed by either an enlarged NT and/or increased risk on CT. The detection rate achieved confirms that early scans performed by trained sonographers can detect around 40–50% of structural anomalies in an unselected population5. In line with previous reports, we have shown that, in particular, severe anomalies are amenable to early diagnosis, as can be inferred by the high number of TOPs, fetal and neonatal losses and low number of live births in the early-diagnosis group5, 13-15. Early TOP was carried out on average at 15 weeks' gestation, after additional investigations and repeat scans had been performed. There is evidence that early TOP is less traumatic for the mother than when carried out at a stage at which fetal movements are already felt, or at a time when legal limits for TOP (usually 20–24 weeks) may push parents into making a rushed decision16. Moreover, the number of false positives and markers is much lower at the early scan, limiting parental anxiety11, 17. Hence, the study confirms the value of an increased NT, observed in over 50% of the early-diagnosed anomalies, as a marker of abnormal development18, 19. cfDNA analysis is offered increasingly as a second- or first-tier screening test5. The question arises as to whether an early 'anomaly' scan should also be part of an up-to-date screening policy20. All pregnant women undergo dating scans; however, in order to maximize detection of anomalies, these should be carried out after 12 weeks' gestation, when ossification of the skull is complete18 and physiologic bowel herniation is resolved19. The scan should be performed by sonographers experienced in first-trimester ultrasound and adhering to a structured protocol9, 21. Policy makers may argue on the cost-effectiveness of this additional scan10, 22. Besides psychological aspects and patient preference17, 23, an early (transvaginal) scan may be preferred, especially for heart examination, to a highly unsatisfactory 20-week scan, in women with high body mass index and at higher risk of congenital anomalies24, 25. However, there are arguments that cfDNA screening (and the associated scan) should be postponed for 1–2 weeks, from 10 to 12 weeks' gestation. First, younger women – the majority of those at reproductive age – have a higher chance of structural rather than chromosomal anomalies; second, this may save unnecessary costs in case of fetal demise; third, it may reduce the number of test failures due to low fetal fraction; fourth, the coincidental finding of a large NT would call for a more advanced genetic examination26, 27; and fifth, it may prevent false-negative cfDNA results in cases of trisomy 18, trisomy 13, Turner syndrome and triploidy9, 28, 29. If the early scan becomes standard practice, the remaining question is whether the 20-week scan should be revisited to include a detailed re-examination of only organs, such as brain and heart, that are still developing or can be better visualized later in pregnancy. In conclusion, policy-makers faced with the challenge of devising an up-to-date screening strategy aimed at maximizing anomaly detection and reproductive choices of parents, should balance the costs of an additional early scan with the advantages of early diagnosis of fetal anomalies. 1 Gezondheidsraad. Wet bevolkingsonderzoek: prenatale screening op downsyndroom en neuralebuisdefecten. Publicatienr. 2007/05WBO. Gezondheidsraad: Den Haag, Netherlands, 2007. 2 Health Council of the Netherlands. Prenatal Screening (2); Down's syndrome, neural tube defects. Publication no. 2004/06. Health Council of the Netherlands: The Hague, the Netherlands, 2004. 3Bakker M, Birnie E, Pajkrt E, Bilardo CM, Snijders RJ. Low uptake of the combined test in The Netherlands – which factors contribute? Prenat Diagn 2012; 32: 1305– 1312. 4Crombag NM, Boeije H, Iedema-Kuiper R, Schielen PC, Visser GH, Bensing JM. 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