Ultrasound Obstet Gynecol 2020; **55**: 474–481
Published online 6 March 2020 in Wiley Online Library (wileyonlinelibrary.com). **DOI:** 10.1002/uog.21938

Diagnosis of fetal defects in twin pregnancies at routine 11–13-week ultrasound examination

A. SYNGELAKI[1], B. CIMPOCA[1], E. LITWINSKA[1], R. AKOLEKAR[2,3] and K. H. NICOLAIDES[1]

[1]*Fetal Medicine Research Institute, King's College Hospital, London, UK;* [2]*Fetal Medicine Unit, Medway Maritime Hospital, Gillingham, UK;* [3]*Institute of Medical Sciences, Canterbury Christ Church University, Chatham, UK*

KEYWORDS: crown–rump-length discordance; dichorionic twins; fetal defects; first-trimester screening; monoamniotic twins; monochorionic twins; nuchal translucency; prenatal diagnosis; twin pregnancy; ultrasound examination

CONTRIBUTION

What are the novel findings of this work?
This study demonstrates that, first, in twins, as in singleton pregnancies, fetal defects essentially fall into three categories in relation to detectability at the 11–13-week scan: always detectable, never detectable or sometimes detectable, and, second, some major fetal defects are more common in monochorionic than in dichorionic twins.

What are the clinical implications of this work?
In twin pregnancies, a routine scan at 11–13 weeks' gestation, carried out according to a standardized protocol, can identify many major fetal defects.

ABSTRACT

Objectives *To examine the performance of the routine 11–13-week scan in detecting fetal defects in twin pregnancies and to examine if, in pregnancies with a fetal defect, compared to those with normal fetuses, there is increased incidence of nuchal translucency thickness (NT) ≥ 95th and ≥ 99th percentiles or intertwin discordance in crown–rump length (CRL) ≥ 10% and ≥ 15%.*

Methods *This was a retrospective analysis of prospectively collected data in twin pregnancies undergoing routine ultrasound examination for fetal anatomy, according to standardized protocols, at 11–13 weeks' gestation between 2002 and 2019. Pregnancies with known chromosomal abnormality were excluded. The final diagnosis of fetal defect was based on the results of postnatal examination in cases of live birth and on the findings of the last ultrasound examination in cases of pregnancy termination, miscarriage or stillbirth. The performance of the 11–13-week scan in the detection of fetal defects was determined.*

Results *The study population of 6366 twin pregnancies with two live fetuses at 11–13 weeks' gestation included 4979 (78.2%) dichorionic (DC) and 1387 (21.8%) monochorionic (MC) twin pregnancies. The main findings were: first, the overall incidence of fetal defects was higher in MC than in DC twins (2.8% vs 1.3%); second, the proportion of defects diagnosed in the first trimester was higher in MC than in DC twins (52.6% vs 27.1%); third, the pattern of defects in relation to detectability at the 11–13-week scan (always detectable, sometimes detectable and never detectable) was similar to that reported previously in singleton pregnancies; fourth, always-detectable defects included acrania, alobar holoprosencephaly, encephalocele, pentalogy of Cantrell, exomphalos, body-stalk anomaly, twin reversed arterial perfusion sequence and conjoined twins; fifth, the incidence of fetal NT ≥ 95th percentile was higher in those with than in those without a defect (16.5% vs 4.5% in DC twins and 19.2% vs 5.9% in MC twins) and this was also true for NT ≥ 99th percentile (8.3% vs 1.0% in DC twins and 15.4% vs 2.0% in MC twins); and sixth, the incidence of CRL discordance ≥ 10% was higher in those with than in those without a defect (20.2% vs 7.9% in DC twins and 33.8% vs 9.3% in MC twins) and this was also true for CRL discordance ≥ 15% (10.1% vs 1.9% in DC twins and 28.2% vs 2.8% in MC twins).*

Conclusions *First, fetal defects are more common in MC than in DC twin pregnancies. Second, first-trimester detection of fetal defects in DC twin pregnancies is similar to that in singleton pregnancies. Third, first-trimester detectability of defects in MC twins is higher than in DC*

Correspondence to: Prof. K. H. Nicolaides, Harris Birthright Research Centre for Fetal Medicine, Fetal Medicine Research Institute, King's College Hospital, 16–20 Windsor Walk, Denmark Hill, London SE5 8BB, UK (e-mail: kypros@fetalmedicine.com)

Accepted: 22 November 2019

twins. Fourth, in twin pregnancies with a fetal defect, there is higher intertwin discordance in CRL and incidence of increased NT, but the predictive performance of screening by these markers is poor.

INTRODUCTION

Ultrasound examination at 11–13 weeks' gestation is used widely for assessment of gestational age[1], diagnosis of multiple pregnancy and chorionicity[2], screening for fetal aneuploidy[3–6] and diagnosis of fetal non-chromosomal defects[7–11]. Two studies in a combined total of 146 188 singleton pregnancies, examined between 2006 and 2019, reported that a routine scan for fetal anatomy at 11–13 weeks' gestation can identify about one-quarter of all defects and that defects can essentially be divided into, first, those that should always be detectable, such as acrania and exomphalos, second, those that are potentially detectable, such as major cardiac defects and spina bifida, and, third, those that are undetectable. The latter group includes, first, defects that develop during the second or third trimester of pregnancy, such as fetal tumors, second, those for which the phenotypic expression becomes apparent later in pregnancy as a result of physiological changes in the fetus, such as increased fetal swallowing unmasking a bowel obstruction, and, third, those that evolve with advancing gestational age, such as short limbs in achondroplasia or pulmonary and aortic stenosis[10,11].

Previous studies on fetal defects in twin pregnancy fall into three categories: first, those that reported the management of pregnancies discordant for specific major fetal abnormalities, such as dichorionic (DC) twins discordant for major trisomies, DC and monochorionic (MC) twins discordant for anencephaly or MC twins with twin reversed arterial perfusion (TRAP) sequence[12–15]; second, studies examining the association between intertwin discordance in crown–rump length (CRL) and incidence of fetal anomalies[16,17]; and, third, screening studies for fetal defects. Sperling *et al.* performed a multicenter study in 421 DC and 74 MC twin pregnancies; fetal defects were identified in 24 (1.9%) cases and 13% of these were detected at the first-trimester scan[18]. D'Antonio *et al.* examined 820 DC and 264 MC twin pregnancies and reported that, in 42 (3.9%) pregnancies, one or more fetuses had a structural defect, 27% of which were detected at the first-trimester scan; increasing discordance in CRL and nuchal translucency thickness (NT) were associated with fetal defects but their predictive performance was only moderately good[19].

The objectives of this study of 6366 twin pregnancies undergoing a routine examination of fetal anatomy were, first, to investigate further the performance of the 11–13-week scan in the detection of fetal defects, and, second, to examine if, in pregnancies with a fetal defect, compared to those with normal fetuses, there is increased incidence of NT $\geq 95^{th}$ and $\geq 99^{th}$ percentile or higher intertwin discordance in CRL.

METHODS

Study population

This was a retrospective analysis of prospectively collected data obtained from women with a twin pregnancy undergoing routine ultrasound examination at 11–13 weeks' gestation at King's College Hospital and the Fetal Medicine Centre, London (January 2002 to February 2019), Medway Maritime Hospital, Gillingham (February 2007 to February 2019) or Southend University Hospital, Essex (March 2009 to February 2019), UK. The three participating hospitals are maternity units and offer routine ultrasound examination in all patients. The Fetal Medicine Centre is a private outpatient clinic of self-referred patients who deliver in many different hospitals. The inclusion criteria for this study were DC or MC twin pregnancy with two live fetuses at 11–13 weeks' gestation and known pregnancy outcome; we included pregnancies with TRAP sequence in which the pump twin was alive and there was demonstrable blood flow in the recipient. We excluded pregnancies that were referred from other hospitals for assessment and those with chromosomal defects diagnosed prenatally or postnatally. This study did not require ethics committee approval.

Ultrasound examination at 11–13 weeks

At the 11–13-week visit, we recorded maternal characteristics and performed an ultrasound scan to, first, determine gestational age from the measurement of CRL of the larger twin[1], second, determine chorionicity from the number of placentas and the presence or absence of the lambda sign at the intertwin membrane–placenta junction[2], third, measure fetal NT as part of screening for trisomies 21, 18 and 13[5,6,20], and, fourth, diagnose any fetal defects[11].

All ultrasound examinations were carried out according to standardized protocols by sonographers who had obtained The Fetal Medicine Foundation Certificate of Competence in ultrasound examination for fetal defects or by trainees under the supervision of certified sonographers. The ultrasound examinations were performed transabdominally, using 3–7.5-MHz curvilinear transducers, but, in 2–3% of cases, when there were technical difficulties in obtaining adequate views, a transvaginal scan (3–9 MHz) was also carried out. The time allocated for the ultrasound examination of twin pregnancies was 60 min. All cases of suspected fetal defects were examined, usually on the same day, by a fetal medicine specialist. Likewise, all cases of suspected fetal cardiac defects were examined by a fetal cardiologist.

At 11–13 weeks, we aimed to obtain a transverse section of the fetal head to demonstrate the skull, midline echo and the choroid plexuses, a midsagittal view of the face to demonstrate the nasal bone, a sagittal section of the spine to demonstrate kyphoscoliosis, and transverse

and sagittal sections of the trunk and extremities to demonstrate the stomach, bladder, abdominal insertion of the umbilical cord, all the long bones, hands and feet. In 2006, the protocol was expanded to include Doppler for assessment of blood flow across the tricuspid valve and in the ductus venosus[21–24]. In 2009, we added use of color Doppler to assess the four-chamber view of the heart and outflow tracts, transverse views of the face to demonstrate the orbits, upper lip and palate, a midsagittal view of the head to demonstrate the midbrain and brainstem and a sagittal section of the spine to demonstrate the spine and overlying skin. Examination of the posterior fossa was included in the protocol only after 2011 and this was based on visual assessment rather than measurements of the brainstem and brainstem–occipital bone diameter[25–28]. Fetal echocardiography by a cardiologist was carried out at 11–13 weeks in all cases of fetal NT above the 99th percentile for CRL and at 20 weeks in those with NT between the 95th and 99th percentiles or regurgitation across the tricuspid valve or abnormal flow in the ductus venosus at 11–13 weeks.

Second- and third-trimester scans

At King's College Hospital, the routine first-trimester scan was offered to all women booked for routine pregnancy care from 1992. In the UK, the 11–13-week scan was offered routinely to all pregnant women only after 2007; before this time, several hospitals referred all their patients to King's College Hospital for a routine first-trimester scan but carried out all subsequent second- and third-trimester scans. Similarly, most patients who had their 11–13-week scan at the Fetal Medicine Centre had follow-up scans in their own hospitals.

During the second-trimester scan in the four participating units, we aimed to obtain the following views: a transverse section of the fetal head at the level of the cavum septi pellucidi and lateral ventricles; a suboccipitobregmatic view to examine the midbrain, cerebellum and vermis; a midsagittal view of the face to examine the nasal bone and exclude micrognathia; transverse views of the orbits, upper lip and palate; sagittal, coronal and transverse views of the spine; a sweep through the heart in the transverse plane to include the four-chamber view, outflow tracts and three-vessel view; transverse and sagittal sections of the thorax and abdomen to examine the lungs, diaphragm, liver, stomach, bowel, umbilical cord insertion, kidneys, bladder and ureters; and systematic examination of the upper and lower limbs for length and shape of each bone, position and movement of each joint and examination of both hands and feet, including the digits. Examination of the genitalia was not a compulsory part of the protocol.

The third-trimester scan was aimed primarily at assessing fetal growth, amniotic fluid volume and Doppler measurements in the umbilical and fetal middle cerebral arteries; in MC twins, we also examined flow in the ductus venosus. The sonographers were instructed to assess the fetal anatomy in the same systematic way as in the second trimester, but it was accepted that, depending on the fetal position, examination of the fetal face, sacrum and extremities may not be possible.

Outcome measures

Data on pregnancy outcome were collected from computerized records of delivery wards and neonatal units or the patients' general practitioners or the patients themselves, and all prenatal and postnatal findings were recorded in a fetal database.

The final diagnosis of fetal defect was based on the results of postnatal examination in cases of live birth and on the findings of the last ultrasound examination in cases of pregnancy termination, miscarriage or stillbirth because, in these cases, postmortem examination was not performed systematically. All babies in our hospitals are examined in the neonatal period by a pediatrician, but certain asymptomatic internal defects are inevitably missed. For example, ventricular septal defects or coarctation of the aorta with patent arterial duct may be missed by early neonatal examination, which does not include echocardiography.

Ventriculomegaly was considered only if the atrial width during the second or third trimester was ≥ 15 mm. Hydronephrosis was considered to be present if there was pelvicalyceal dilatation with anteroposterior diameter ≥ 10 mm in the second trimester or ≥ 15 mm in the third trimester. We considered only severe ventriculomegaly and hydronephrosis because the incidence of milder degrees is much higher, and their clinical consequences are questionable. In cases of exomphalos with a sac containing only bowel, megacystis, ventriculomegaly and hydronephrosis, a follow-up scan was carried out, and those with spontaneous resolution of the abnormality were considered to be normal. Polydactyly was considered to be present if the extra digit contained bone, and talipes was considered to be present if the baby required postnatal treatment.

We included all cases of defects of the heart and great vessels but excluded cases of persistent left superior vena cava and aberrant right subclavian artery because these are variants of normal rather than true defects. Cases with coarctation of the aorta, aortic arch hypoplasia and interrupted aortic arch were classified as arch defects. Cases with at least two different major heart defects were classified as complex.

Association of fetal defects with increased NT and high CRL discordance

The incidence of fetal NT $\geq 95^{th}$ and $\geq 99^{th}$ percentiles for CRL in each fetus with a defect was determined and compared to that in fetuses without a defect using the chi-square test with Yates' correction for large sample sizes[29,30]. Similarly, the incidence of CRL discordance $\geq 10\%$ and $\geq 15\%$ in pregnancies with at least one abnormal fetus was compared to that in pregnancies with two normal fetuses[31].

RESULTS

Study population

The study population of 6366 twin pregnancies with two live fetuses at 11–13 weeks' gestation and known pregnancy outcome was composed of 4979 (78.2%) DC and 1387 (21.8%) MC twin pregnancies, including 67 (4.8%) that were MC monoamniotic. Median maternal age was 33.7 (interquartile range (IQR), 29.8–37.2) years, median maternal weight was 67.6 (IQR, 60.0–77.1) kg, and racial origin of the women was white in 5242 (82.3%), black in 660 (10.4%), South Asian in 273 (4.3%), East Asian in 91 (1.4%) and mixed in 100 (1.6%). At the time of the first-trimester scan, median CRL of the larger twin was 65.3 (IQR, 60.2–71.2) mm and median gestational age was 12.9 (IQR, 12.5–13.3) weeks.

Termination of the whole pregnancy because of a fetal defect or miscarriage before 20 weeks' gestation occurred in 119 pregnancies. Of the remaining 6247 pregnancies, follow-up scans were carried out in the four participating units in 69.0% (4308/6247).

Fetal defects

A fetal defect was identified either prenatally or postnatally in 1.7% (211/12 732) of twins, including in 2.8% (78/2774) of MC twins, which was higher than in DC twins (1.3%; 133/9958; $P < 0.0001$).

In the 131 of 4979 DC twin pregnancies with a fetal defect, one fetus was affected in 129 and both were affected in two. In the 71 of 1387 MC twin pregnancies with a fetal defect, one fetus was affected in 64 and both were affected in seven. At the 11–13-week scan, we diagnosed 36.5% (77/211) of defects, including 27.1% (36/133) of those in fetuses from DC twin pregnancies and 52.6% (41/78) of those in fetuses from MC twin pregnancies (Table 1).

At 11–13 weeks, we diagnosed: first, all cases of acrania, alobar holoprosencephaly and encephalocele, and 40% of cases of open spina bifida, but none of severe ventriculomegaly, absent corpus callosum, hypoplastic cerebellum and/or vermis; second, 33% of cases of cleft lip and palate, but none of cleft lip only; third, 50% of cases of congenital diaphragmatic hernia, but none of congenital pulmonary airway malformation; fourth, 52% (14/27) of cases of tetralogy of Fallot, hypoplastic left heart syndrome, arch defects, tricuspid atresia or complex heart defect, but none of atrioventricular or ventricular septal defect, transposition of the great arteries, aortic or pulmonary stenosis/atresia, double or right aortic arch, arrythmia or rhabdomyoma; fifth, all cases of exomphalos, but none of duodenal atresia or bladder exstrophy; sixth, 71% of cases of lower urinary tract obstruction, but none of other urogenital defects; seventh, 67% (6/9) of cases of lethal skeletal dysplasia, fetal akinesia deformation sequence or absence of extremities, but none of hemivertebra, defects of digits, deformities of wrists or talipes; and eighth, all cases of body-stalk anomaly, pentalogy of Cantrell, TRAP sequence and conjoined twins, but none of lymphangioma or sacrococcygeal teratoma.

Association of fetal defects with increased NT

Of the total population of 12 732 fetuses, 638 (5.0%) had fetal NT $\geq 95^{th}$ percentile and 172 (1.4%) had NT $\geq 99^{th}$ percentile. In fetuses from both DC and MC twin pregnancies, the incidence of NT $\geq 95^{th}$ and $\geq 99^{th}$ percentiles was higher in those with than in those without a defect (Table 2). In the total population of fetuses from twin pregnancies, the percentage with a fetal defect was 5.8% (37/638) in those with NT $\geq 95^{th}$ percentile and 1.4% (174/12 094) in those with NT $< 95^{th}$ percentile (relative risk, 4.031 (95% CI, 2.853–7.902); $P < 0.0001$). The percentage with a fetal defect was 13.4% (23/172) in those with NT $\geq 99^{th}$ percentile and 1.5% (188/12 560) in those with NT $< 99^{th}$ percentile (relative risk, 8.934 (95% CI, 5.953–13.407); $P < 0.0001$).

Association of fetal defects with high intertwin CRL discordance

In all twin pregnancies, median intertwin discordance in CRL was 3.3% (IQR, 1.4–6.0%); intertwin discordance in CRL was $\geq 10\%$ in 8.7% (557/6366) of pregnancies and $\geq 15\%$ in 2.5% (162/6366). In both DC and MC twin pregnancies, the incidence of CRL discordance $\geq 10\%$ and $\geq 15\%$ was higher in those with than in those without defects (Table 2). In the total population of twin pregnancies, the percentage with a fetal defect was 9.0% (50/557) in those with CRL discordance $\geq 10\%$ and 2.6% (150/5809) in those with CRL discordance $< 10\%$ (relative risk, 3.476 (95% CI, 2.555–4.730); $P < 0.0001$). The percentage with a fetal defect was 20.4% (33/162) in those with CRL discordance $\geq 15\%$ and 2.7% (167/6204) in those with CRL discordance $< 15\%$ (relative risk, 7.568 (95% CI, 5.390–10.624); $P < 0.0001$).

DISCUSSION

Main findings

This study of 6366 twin pregnancies with two live fetuses at 11–13 weeks' gestation and known pregnancy outcome involved systematic examination of the fetal anatomy according to standardized protocols and in an allocated period of 60 min; however, the protocol evolved over the study period, with the requirement for examination of additional structures. Fetuses with transient anomalies, including exomphalos with a sac containing only bowel, megacystis, ventriculomegaly and hydronephrosis, were considered to be normal. The main findings of this study are: first, the overall incidence of fetal defects was higher in MC than in DC twins (2.8% *vs* 1.3%); second, the proportion of defects diagnosed in the first trimester was higher in MC than in DC twins (52.6% *vs* 27.1%); third, the pattern of defects in relation to detectability at the 11–13 weeks scan (always detectable,

Table 1 Diagnosis of fetal defects on routine ultrasound examination at 11–13 weeks

			Twin pregnancies					
	*Singleton pregnancies**		*All* (n = *12 732*)		*Dichorionic* (n = *9958*)		*Monochorionic* (n = *2774*)	
Defect	*Prevalence* (*1 in* N)	*DR* (%)	*Prevalence* (n (*1 in* N))	*DR* (n (%))	*Prevalence* (n (*1 in* N))	*DR* (n (%))	*Prevalence* (n (*1 in* N))	*DR* (n (%))
Central nervous system	1 in 437	47.6	26 (1 in 490)	15 (57.7)	18 (1 in 553)	9 (50.0)	8 (1 in 347)	6 (75.0)
Acrania	1 in 2104	100	10 (1 in 1273)	10 (100)	5 (1 in 1992)	5 (100)	5 (1 in 555)	5 (100)
Encephalocele	1 in 6733	100	2 (1 in 6366)	2 (100)	1 (1 in 9958)	1 (100)	1 (1 in 2774)	1 (100)
OSB	1 in 1712	59.3	5 (1 in 2546)	2 (40.0)	5 (1 in 1992)	2 (40.0)	0	—
Alobar holoprosencephaly	1 in 10 100	100	1 (1 in 12 732)	1 (100)	1 (1 in 9958)	1 (100)	0	—
ACC	1 in 3885	0	2 (1 in 6366)	0 (0)	2 (1 in 4979)	0 (0)	0	—
Severe ventriculomegaly	1 in 5611	0	1 (1 in 12 732)	0 (0)	0	—	1 (1 in 2774)	0 (0)
Hypoplastic cerebellum/vermis	1 in 6733	13.3	5 (1 in 2546)	0 (0)	4 (1 in 2490)	0 (0)	1 (1 in 2774)	0 (0)
Face	1 in 962	18.1	13 (1 in 979)	3 (23.1)	6 (1 in 1660)	2 (33.3)	7 (1 in 396)	1 (14.3)
Cleft lip and palate	1 in 1942	34.6	9 (1 in 1415)	3 (33.3)	4 (1 in 2490)	2 (50.0)	5 (1 in 555)	1 (20.0)
Cleft lip only	1 in 3607	0	4 (1 in 3183)	0 (0)	2 (1 in 4979)	0 (0)	2 (1 in 1387)	0 (0)
Thorax	1 in 1403	9.7	11 (1 in 1157)	2 (18.2)	9 (1 in 1106)	1 (11.1)	2 (1 in 1387)	1 (50.0)
CPAM	1 in 2349	0	7 (1 in 1819)	0 (0)	6 (1 in 1660)	0 (0)	1 (1 in 2774)	0 (0)
CDH	1 in 4208	29.2	4 (1 in 3183)	2 (50.0)	3 (1 in 3319)	1 (33.3)	1 (1 in 2774)	1 (100)
Heart	1 in 260	30.1	58 (1 in 220)	14 (24.1)	38 (1 in 262)	11 (28.9)	20 (1 in 139)	3 (15.0)
TOF	1 in 3607	39.3	8 (1 in 1592)	3 (37.5)	3 (1 in 3319)	1 (33.3)	5 (1 in 555)	2 (40.0)
TGA	1 in 6733	13.3	2 (1 in 6366)	0 (0)	1 (1 in 9958)	0 (0)	1 (1 in 2774)	0 (0)
HLHS	1 in 2525	92.5	7 (1 in 1819)	6 (85.7)	7 (1 in 1423)	6 (85.7)	0	—
ASD	1 in 9182	90.9	2 (1 in 6366)	0 (0)	2 (1 in 4979)	0 (0)	0	—
VSD	1 in 743	0	11 (1 in 1157)	0 (0)	6 (1 in 1660)	0 (0)	5 (1 in 555)	0 (0)
Arch abnormality	1 in 2658	31.6	7 (1 in 1819)	3 (42.9)	5 (1 in 1992)	3 (60.0)	2 (1 in 1387)	0 (0)
Double/right aortic arch	1 in 3156	15.6	7 (1 in 1819)	0 (0)	6 (1 in 1660)	0 (0)	1 (1 in 2774)	0 (0)
Tricuspid atresia	1 in 14 428	100	2 (1 in 6366)	1 (50.0)	1 (1 in 9958)	0 (0)	1 (1 in 2774)	1 (100)
Pulmonary atresia	1 in 9182	100	2 (1 in 6366)	0 (0)	2 (1 in 4979)	0 (0)	0	—
Pulmonary stenosis	1 in 10 100	0	2 (1 in 6366)	0 (0)	1 (1 in 9958)	0 (0)	1 (1 in 2774)	0 (0)
Aortic stenosis	1 in 16 833	0	1 (1 in 12 732)	0 (0)	1 (1 in 9958)	0 (0)	0	—
Complex heart defect	1 in 4040	60	3 (1 in 4244)	1 (33.3)	1 (1 in 9958)	1 (100)	2 (1 in 1387)	0 (0)
Rhabdomyoma	1 in 16 833	0	1 (1 in 12 732)	0 (0)	1 (1 in 9958)	0 (0)	0	—
Arrhythmia	1 in 33 666	0	3 (1 in 4244)	0 (0)	1 (1 in 9958)	0 (0)	2 (1 in 1387)	0 (0)
Gastrointestinal/abdominal wall	1 in 727	63.3	8 (1 in 1592)	6 (75.0)	5 (1 in 1992)	4 (80.0)	3 (1 in 925)	2 (66.7)
Exomphalos with bowel or liver	1 in 2295	100	6 (1 in 2122)	6 (100)	4 (1 in 2490)	4 (100)	2 (1 in 1387)	2 (100)
Bladder exstrophy	1 in 50 499	0	1 (1 in 12 732)	0 (0)	0	—	1 (1 in 2774)	0 (0)
Duodenal atresia	1 in 11 222	0	1 (1 in 12 732)	0 (0)	1 (1 in 9958)	0 (0)	0	—
Urogenital	1 in 196	8.4	30 (1 in 424)	5 (16.7)	22 (1 in 453)	2 (9.1)	8 (1 in 347)	3 (37.5)
LUTO	1 in 1942	71.2	7 (1 in 1819)	5 (71.4)	4 (1 in 2490)	2 (50.0)	3 (1 in 925)	3 (100)
Severe hydronephrosis	1 in 1278	0	2 (1 in 6366)	0 (0)	2 (1 in 4979)	0 (0)	0	—
Bilateral polycystic kidneys	1 in 7214	7.1	1 (1 in 12 732)	0 (0)	1 (1 in 9958)	0 (0)	0	—
Bilateral MK	1 in 25 249	0	1 (1 in 12 732)	0 (0)	1 (1 in 9958)	0 (0)	0	—
Unilateral MK	1 in 1741	0	5 (1 in 2546)	0 (0)	4 (1 in 2490)	0 (0)	1 (1 in 2774)	0 (0)
Bilateral renal agenesis	1 in 7769	15.4	1 (1 in 12 732)	0 (0)	1 (1 in 9958)	0 (0)	0	—
Unilateral pelvic kidney/agenesis	1 in 814	2.4	6 (1 in 2122)	0 (0)	4 (1 in 2490)	0 (0)	2 (1 in 1387)	0 (0)
Duplex kidney	1 in 1161	0	4 (1 in 3183)	0 (0)	2 (1 in 4979)	0 (0)	2 (1 in 1387)	0 (0)
Unilateral dilated ureter	1 in 16 833	0	1 (1 in 12 732)	0 (0)	1 (1 in 9958)	0 (0)	0	—
Hypospadias	1 in 3885	0	1 (1 in 12 732)	0 (0)	1 (1 in 9958)	0 (0)	0	—
Ambiguous genitalia	1 in 20 199	0	1 (1 in 12 732)	0 (0)	1 (1 in 9958)	0 (0)	0	—
Skeleton/extremities	1 in 449	29.8	35 (1 in 364)	6 (17.1)	29 (1 in 343)	5 (17.2)	6 (1 in 462)	1 (16.7)
Lethal skeletal dysplasia	1 in 7214	71.4	2 (1 in 6366)	1 (50.0)	2 (1 in 4979)	1 (50.0)	0	—
FADS	1 in 9182	18.2	3 (1 in 4244)	2 (66.7)	2 (1 in 4979)	1 (50.0)	1 (1 in 2774)	1 (100)
Absent hand, arm, leg or foot	1 in 4208	75.0	4 (1 in 3183)	3 (75.0)	4 (1 in 2490)	3 (75.0)	0	—
Hemivertebra	1 in 8416	33.3	1 (1 in 12 732)	0 (0)	1 (1 in 9958)	0 (0)	0	—
Abnormal digits	1 in 1712	42.4	4 (1 in 3183)	0 (0)	4 (1 in 2490)	0 (0)	0	—
Deformity of wrists			3 (1 in 4244)	0 (0)	1 (1 in 9958)	0 (0)	2 (1 in 1387)	0 (0)
Talipes	1 in 1086	2.2	18 (1 in 707)	0 (0)	15 (1 in 664)	0 (0)	3 (1 in 925)	0 (0)

Continued over.

Table 1 *Continued*

			Twin pregnancies					
	*Singleton pregnancies**		*All* (n = *12 732*)		*Dichorionic* (n = *9958*)		*Monochorionic* (n = *2774*)	
Defect	*Prevalence (1 in* N*)*	*DR (%)*	*Prevalence* (n *(1 in* N*))*	*DR* (n *(%))*	*Prevalence* (n *(1 in* N*))*	*DR* (n *(%))*	*Prevalence* (n *(1 in* N*))*	*DR* (n *(%))*
Other	1 in 449	29.8	24 (1 in 531)	22 (91.7)	3 (1 in 3319)	1 (33.3)	21 (1 in 132)	21 (100)
Body-stalk anomaly	1 in 6312	100	2 (1 in 6366)	2 (100)	0	—	2 (1 in 1387)	2 (100)
Pentalogy of Cantrell	1 in 50 499	100	1 (1 in 12 732)	1 (100)	1 (1 in 9958)	1 (100)	0	—
Lymphangioma	1 in 25 249	0	1 (1 in 12 732)	0 (0)	1 (1 in 9958)	0 (0)	0	—
Sacrococcygeal teratoma	1 in 50 499	50.0	1 (1 in 12 732)	0 (0)	1 (1 in 9958)	0 (0)	0	—
TRAP sequence	—	—	13 (1 in 979)	13 (100)	0	—	13 (1 in 213)	13 (100)
Conjoined twins	—	—	6 (1 in 2122)	6 (100)	0	—	6 (1 in 462)	6 (100)
Multiple	1 in 12 625	25.0	6 (1 in 2122)	4 (66.7)	3 (1 in 3319)	1 (33.3)	3 (1 in 925)	3 (100)
CoA, clenched hands, talipes	—	—	1 (1 in 12 732)	0 (0)	1 (1 in 9958)	0 (0)	0	—
CoA, bilateral MK	—	—	1 (1 in 12 732)	1 (100)	0	—	1 (1 in 2774)	1 (100)
DWM, micrognathia, cleft palate	—	—	1 (1 in 12 732)	0 (0)	1 (1 in 9958)	0 (0)	0	—
Dextrocardia, phocomelia, bilateral renal agenesis	—	—	1 (1 in 12 732)	1 (100)	0	—	1 (1 in 2774)	1 (100)
HLHS, exomphalos bowel	—	—	1 (1 in 12 732)	1 (100)	1 (1 in 9958)	1 (100)	0	—
OSB, bladder exstrophy	—	—	1 (1 in 12 732)	1 (100)	0	—	1 (1 in 2774)	1 (100)
Total	—	—	211 (1.7)	77 (36.5)	133 (1.3)	36 (27.1)	78 (2.8)	41 (52.6)

*Data from previous study in singleton pregnancies[11]. ACC, agenesis of corpus callosum; ASD, atrioventricular septal defect; CDH, congenital diaphragmatic hernia; CoA, coarctation of the aorta; CPAM, congenital pulmonary airway malformation; DR, detection rate; DWM, Dandy–Walker malformation; FADS, fetal akinesia deformation sequence; HLHS, hypoplastic left heart syndrome; LUTO, lower urinary tract obstruction; MK, multicystic kidney; OSB, open spina bifida; TGA, transposition of the great arteries; TOF, tetralogy of Fallot; TRAP, twin reversed arterial perfusion; VSD, ventricular septal defect.

Table 2 Incidence of increased nuchal translucency thickness (NT) and high crown–rump length (CRL) discordance in 6366 dichorionic and monochorionic twin pregnancies, according to presence of fetal defect

Variable	*No fetal defect*	*Fetal defect*	P
NT ≥ 95th percentile*			
Dichorionic	441/9825 (4.5)	22/133 (16.5)	< 0.0001
Monochorionic	160/2696 (5.9)	15/78 (19.2)	< 0.0001
NT ≥ 99th percentile*			
Dichorionic	96/9825 (1.0)	11/133 (8.3)	< 0.0001
Monochorionic	53/2696 (2.0)	12/78 (15.4)	< 0.0001
CRL discordance ≥ 10%†			
Dichorionic	384/4850 (7.9)	26/129 (20.2)	< 0.0001
Monochorionic	123/1316 (9.3)	24/71 (33.8)	< 0.0001
CRL discordance ≥ 15%†			
Dichorionic	92/4850 (1.9)	13/129 (10.1)	< 0.0001
Monochorionic	37/1316 (2.8)	20/71 (28.2)	< 0.0001

Data are given as *n*/*N* (%). *Per fetus. †Per pregnancy.

sometimes detectable and never detectable) was similar to that reported previously in singleton pregnancies[10,11]; fourth, always-detectable defects included acrania, alobar holoprosencephaly, encephalocele, pentalogy of Cantrell, exomphalos, body-stalk anomaly, TRAP sequence and conjoined twins; fifth, the incidence of fetal NT ≥ 95th percentile was higher in those with than in those without a defect (16.5% *vs* 4.5% in DC twins and 19.2% *vs* 5.9% in MC twins) and this was also true for NT ≥ 99th percentile (8.3% *vs* 1.0% in DC twins and 15.4% *vs* 2.0% in MC twins); and sixth, the incidence of CRL discordance ≥ 10% was higher in those with than in those without a defect (20.2% *vs* 7.9% in DC twins and 33.8% *vs* 9.3% in MC twins) and this was also true for CRL discordance ≥ 15% (10.1% *vs* 1.9% in DC twins and 28.2% *vs* 2.8% in MC twins).

Comparison with findings of previous studies

At the 11–13-week scan, we diagnosed 36.5% of defects, including 27.1% of those in fetuses from DC twin pregnancies and 52.6% of those in fetuses from MC twin pregnancies. In two previous studies in 495 and 1084 twin pregnancies, the proportion of defects detected in the first trimester was 13% and 27%, respectively[18,19]. In relation to the types of defects that are detected at 11–13 weeks, the results are consistent with those of our previous studies in singleton pregnancies, which highlighted that defects can essentially be divided into those that should be always detectable, those that are potentially detectable and those that are undetectable[10,11]. The higher detectability of defects in MC than in DC twin pregnancies is to a great extent the consequence of, first, TRAP sequence and conjoined twins which are unique to MC twinning and accounted for 24% of defects in such pregnancies, and, second, easily detectable defects which were more common in MC than in DC twins, such as

acrania, exomphalos, lower urinary tract obstruction and body-stalk anomaly. Successful first-trimester diagnosis of the potentially detectable defects depends on, first, the objectives set for such a scan and, consequently, the time allocated for the fetal examination, the expertise of the sonographer and the quality of the equipment used, and, second, the presence of an easily detectable marker for an underlying abnormality, such as increased NT or abnormal flow across the tricuspid valve and in the ductus venosus for cardiac defects and posterior fossa defects for open spina bifida.

We found that, in pregnancies with a fetal defect, there is higher intertwin discordance in CRL and incidence of increased NT, but the predictive performance of screening by CRL discordance $\geq 10\%$ or $\geq 15\%$ or fetal NT $\geq 95^{th}$ or $\geq 99^{th}$ is poor. These findings are consistent with the results of previous smaller studies. Kalish *et al.* examined 159 DC twin pregnancies at 11–14 weeks' gestation, including nine with a fetal structural or chromosomal defect, and reported that the incidence of CRL discordance $> 10\%$ was higher in those with than in those without a defect (22.2% *vs* 2.8%)[16]. Harper *et al.* examined 594 DC twin pregnancies at 7–14 weeks' gestation, including 111 with a fetal defect, and reported that the incidence of CRL discordance $\geq 11\%$ was higher in those with than in those without a defect (27.3% *vs* 17.4%)[17]. D'Antonio *et al.* examined 820 DC and 264 MC twin pregnancies at 11–14 weeks' gestation, including 42 with a fetal structural defect, and reported that increased intertwin discordance in either CRL or NT was associated with an increased risk for fetal defect, but the predictive accuracy was only moderately good (CRL discordance: detection rate of 76%, false-positive rate of 45%; NT discordance: detection rate of 71%, false-positive rate of 40%)[19].

Strengths and limitations

The main strength of our study is the examination of a large number of twin pregnancies attending for a routine first-trimester scan performed using standardized protocols and by appropriately trained sonographers in units with expertise in fetal medicine and fetal cardiology. However, the overall number of cases is still small for meaningful conclusions to be drawn concerning detectability of individual defects. This problem of small numbers of individual defects cannot be overcome by reporting overall detection rates because these are inherently dependent on the distribution of different types of defects within a given study population. During this study, which spanned over a period of 17 years, there was evolution in the detail of the first-trimester ultrasound scan and incorporation of new easily recognizable markers of underlying defects.

The main limitation of this and most previous studies investigating the effectiveness of routine ultrasound examination in the prenatal diagnosis of fetal defects relates to ascertainment of such defects. For example, although in our participating centers all neonates are examined by pediatricians, it is possible that asymptomatic defects of internal organs could be missed. Similarly, we assumed that all morphologically normal neonates were chromosomally normal and, in cases of pregnancy termination, miscarriage or stillbirth, we assumed that the findings of the last ultrasound examination were correct because, in these cases, postmortem examination was not performed systematically. Furthermore, in this study, about 30% of patients delivered in hospitals other than the ones in which the first-trimester scan was carried out.

Conclusions

First, fetal defects are more common in MC than in DC twin pregnancies. Second, first-trimester detection of fetal defects in DC twin pregnancies is similar to that in singleton pregnancies. Third, detectability of defects in MC twins is higher than in DC twins. Fourth, in twin pregnancies with high intertwin discordance in CRL and increased NT, the incidence of fetal defects is increased, but the predictive performance of screening by these markers is poor.

ACKNOWLEDGMENT

This study was supported by a grant from The Fetal Medicine Foundation (Charity No: 1037116).

REFERENCES

1. Robinson HP, Fleming JE. A critical evaluation of sonar crown rump length measurements. *Br J Obstet Gynaecol* 1975; **82**: 702–710.
2. Sepulveda W, Sebire NJ, Hughes K, Odibo A, Nicolaides KH. The lambda sign at 10–14 weeks of gestation as a predictor of chorionicity in twin pregnancies. *Ultrasound Obstet Gynecol* 1996; **7**: 421–423.
3. Nicolaides KH, Azar GB, Byrne D, Mansur CA, Marks K. Nuchal translucency: ultrasound screening for chromosomal defects in the first trimester of pregnancy. *BMJ* 1992; **304**: 867–869.
4. Snijders RJ, Noble P, Sebire N, Souka A, Nicolaides KH. UK multicentre project on assessment of risk of trisomy 21 by maternal age and fetal nuchal-translucency thickness at 10–14 weeks of gestation. Fetal Medicine Foundation First Trimester Screening Group. *Lancet* 1998; **352**: 343–346.
5. Sebire NJ, Snijders RJM, Hughes K, Sepulveda W, Nicolaides KH. Screening for trisomy 21 in twin pregnancies by maternal age and fetal nuchal translucency thickness at 10–14 weeks of gestation. *Br J Obstet Gynaecol* 1996; **103**: 999–1003.
6. Vandecruys H, Faiola S, Auer M, Sebire N, Nicolaides KH. Screening for trisomy 21 in monochorionic twins by measurement of fetal nuchal translucency thickness. *Ultrasound Obstet Gynecol* 2005; **25**: 551–553.
7. Hyett J, Moscoso G, Papapanagiotou G, Perdu M, Nicolaides KH. Abnormalities of the heart and great arteries in chromosomally normal fetuses with increased nuchal translucency thickness at 11–13 weeks of gestation. *Ultrasound Obstet Gynecol* 1996; **7**: 245–250.
8. Johnson SP, Sebire NJ, Snijders RJ, Tunkel S, Nicolaides KH. Ultrasound screening for anencephaly at 10–14 weeks of gestation. *Ultrasound Obstet Gynecol* 1997; **9**: 14–16.
9. Souka AP, Snijders RJ, Novakov A, Soares W, Nicolaides KH. Defects and syndromes in chromosomally normal fetuses with increased nuchal translucency thickness at 10–14 weeks of gestation. *Ultrasound Obstet Gynecol* 1998; **11**: 391–400.
10. Syngelaki A, Chelemen T, Dagklis T, Allan L, Nicolaides KH. Challenges in the diagnosis of fetal non-chromosomal abnormalities at 11–13 weeks. *Prenat Diagn* 2011; **31**: 90–102.
11. Syngelaki A, Hammami A, Bower S, Zidere V, Akolekar R, Nicolaides KH. Diagnosis of fetal non-chromosomal abnormalities at routine ultrasound examination at 11–13 weeks' gestation. *Ultrasound Obstet Gynecol* 2019; **54**: 468–476.
12. Sebire NJ, Snijders RJ, Santiago C, Papapanagiotou G, Nicolaides KH. Management of twin pregnancies with fetal trisomies. *Br J Obstet Gynaecol* 1997; **104**: 220–222.
13. Sebire NJ, Sepulveda W, Hughes KS, Noble P, Nicolaides KH. Management of twin pregnancies discordant for anencephaly. *Br J Obstet Gynaecol* 1997; **104**: 216–219.
14. Vandecruys H, Avgidou K, Surerus E, Flack N, Nicolaides KH. Dilemmas in the management of twins discordant for anencephaly diagnosed at 11 + 0 to 13 + 6 weeks of gestation. *Ultrasound Obstet Gynecol* 2006; **28**: 653–658.

15. Chaveeva P, Poon LC, Sotiriadis A, Kosinski P, Nicolaides KH. Optimal method and timing of intrauterine intervention in twin reversed arterial perfusion sequence: case study and meta-analysis. *Fetal Diagn Ther* 2014; **35**: 267–279.
16. Kalish RB, Gupta M, Perni SC, Berman S, Chasen ST. Clinical significance of first trimester crown–rump length disparity in dichorionic twin gestations. *Am J Obstet Gynecol* 2004; **191**: 1437–1440.
17. Harper LM, Roehl KA, Odibo AO, Cahill AG. First-trimester growth discordance and adverse pregnancy outcome in dichorionic twins. *Ultrasound Obstet Gynecol* 2013; **41**: 627–631.
18. Sperling L, Kiil C, Larsen LU, Brocks V, Wojdemann KR, Qvist I, Schwartz M, Jørgensen C, Espersen G, Skajaa K, Bang J, Tabor A. Detection of chromosomal abnormalities, congenital abnormalities and transfusion syndrome in twins. *Ultrasound Obstet Gynecol* 2007; **29**: 517–526.
19. D'Antonio F, Familiari A, Thilaganathan B, Papageorghiou AT, Manzoli L, Khalil A, Bhide A. Sensitivity of first-trimester ultrasound in the detection of congenital anomalies in twin pregnancies: population study and systematic review. *Acta Obstet Gynecol Scand* 2016; **95**: 1359–1367.
20. Madsen HN, Ball S, Wright D, Tørring N, Petersen OB, Nicolaides KH, Spencer K. A reassessment of biochemical marker distributions in trisomy 21-affected and unaffected twin pregnancies in the first trimester. *Ultrasound Obstet Gynecol* 2011; **37**: 38–47.
21. Maiz N, Plasencia W, Dagklis T, Faros E, Nicolaides K. Ductus venosus Doppler in fetuses with cardiac defects and increased nuchal translucency thickness. *Ultrasound Obstet Gynecol* 2008; **31**: 256–260.
22. Chelemen T, Syngelaki A, Maiz N, Allan L, Nicolaides KH. Contribution of ductus venosus Doppler in first-trimester screening for major cardiac defects. *Fetal Diagn Ther* 2011; **29**: 127–134.
23. Kagan KO, Valencia C, Livanos P, Wright D, Nicolaides KH. Tricuspid regurgitation in screening for trisomies 21, 18 and 13 and Turner syndrome at 11 + 0 to 13 + 6 weeks of gestation. *Ultrasound Obstet Gynecol* 2009; **33**: 18–22.
24. Pereira S, Ganapathy R, Syngelaki A, Maiz N, Nicolaides KH. Contribution of fetal tricuspid regurgitation in first-trimester screening for major cardiac defects. *Obstet Gynecol* 2011; **117**: 1384–1391.
25. Chaoui R, Benoit B, Mitkowska-Wozniak H, Heling KS, Nicolaides KH. Assessment of intracranial translucency (IT) in the detection of spina bifida at the 11–13-week scan. *Ultrasound Obstet Gynecol* 2009; **34**: 249–252.
26. Chaoui R, Nicolaides KH. From nuchal translucency to intracranial translucency: towards the early detection of spina bifida. *Ultrasound Obstet Gynecol* 2010; **35**: 133–138.
27. Chaoui R, Nicolaides KH. Detecting open spina bifida at the 11–13-week scan by assessing intracranial translucency and the posterior brain region: mid-sagittal or axial plane? *Ultrasound Obstet Gynecol* 2011; **38**: 609–612.
28. Lachmann R, Chaoui R, Moratalla J, Picciarelli G, Nicolaides KH. Posterior brain in fetuses with open spina bifida at 11 to 13 weeks. *Prenat Diagn* 2011; **31**: 103–106.
29. Wright D, Kagan KO, Molina FS, Gazzoni A, Nicolaides KH. A mixture model of nuchal translucency thickness in screening for chromosomal defects. *Ultrasound Obstet Gynecol* 2008; **31**: 376–383.
30. Cimpoca B, Syngelaki A, Litwinska E, Muzaferovic A, Nicolaides KH. Increased nuchal translucency at 11–13 weeks' gestation and pregnancy outcome in twin pregnancy. *Ultrasound Obstet Gynecol* 2020; **55**: 318–325.
31. Litwinska E, Syngelaki A, Cimpoca B, Sapantzoglou I, Nicolaides KH. Intertwin discordance in fetal size at 11–13 weeks' gestation and pregnancy outcome. *Ultrasound Obstet Gynecol* 2020; **55**: 189–197.

This article has been selected for Journal Club.

A slide presentation prepared by Dr Fiona Brownfoot, one of UOG's Editors for Trainees, is available online.

Spanish translation by Dr Rubén D. Fernández Jr.
Chinese translation by Dr Xiaofei Li and Prof. Qingqing Wu, ISUOG China Task Force.

Ultrasound Obstet Gynecol 2020; **55**: 474–481
Published online 6 March 2020 in Wiley Online Library (wileyonlinelibrary.com). **DOI:** 10.1002/uog.21938

Diagnóstico de defectos del feto en embarazos de gemelos en el examen ecográfico de rutina de las 11–13 semanas

RESUMEN

Objetivos Examinar la eficacia del examen rutinario de 11–13 semanas para detectar defectos fetales en embarazos de gemelos y examinar si, en los embarazos con un defecto fetal, en comparación con los de fetos normales, hay una mayor incidencia del grosor de la translucencia nucal (TN) ≥ percentil 95º y ≥ percentil 99º o una discordancia entre gemelos en la longitud céfalo-caudal (LCC) ≥10% y ≥15%.

Métodos Este estudio fue un análisis retrospectivo de datos recogidos prospectivamente de embarazos de gemelos sometidos a exámenes ecográficos de rutina entre 2002 y 2019 para determinar la anatomía del feto, según protocolos estándar a las 11–13 semanas de gestación. Se excluyeron los embarazos con anomalías cromosómicas conocidas. El diagnóstico final de la anomalía fetal se basó en los resultados del examen posnatal en los casos de nacimientos vivos y en los hallazgos del último examen ecográfico en los casos de interrupción del embarazo, aborto o éxitus fetal. Se determinó la eficacia de la exploración de las 11–13 semanas en la detección de anomalías fetales.

Resultados La población de estudio fue de 6366 embarazos de gemelos con dos fetos vivos a las 11–13 semanas de gestación e incluyó 4979 (78,2%) embarazos bicoriales (BC) y 1387 (21,8%) monocoriales (MC). Los principales hallazgos fueron: primero, la prevalencia total de defectos fetales fue mayor en los gemelos MC que en los gemelos BC (2,8% vs. 1,3%); segundo, la proporción de defectos diagnosticados en el primer trimestre fue mayor en los gemelos MC que en los gemelos BC (52,6% vs. 27,1%); tercero, la pauta de defectos en relación con la detectabilidad en la exploración de 11–13 semanas (siempre detectable, a veces detectable y nunca detectable) fue similar a la reportada previamente para los embarazos con feto único; cuarto, entre los defectos siempre detectables estaban la acrania, la holoprosencefalia alobar, el encefalocele, la pentalogía de Cantrell, el onfalocele, la anomalía del pedículo embrionario, la secuencia de perfusión arterial inversa de los gemelos y los gemelos unidos; quinto, la frecuencia del percentil de la TN fetal ≥95º fue mayor en los que tenían un defecto que en los que no lo tenían (16,5% vs 4,5% en los gemelos BC y 19,2% vs 5,9% en los gemelos MC) y esto también fue cierto para el percentil de la TN ≥99º (8,3% vs 1,0% en gemelos BC y 15,4% vs 2,0% en gemelos MC); y sexto, la frecuencia de una discordancia de la LCC ≥10% fue mayor en los que tenían un defecto que en los que no lo tenían (20,2% vs 7,9% en los gemelos BC y 33,8% vs 9,3% en los gemelos MC) y esto también fue cierto para la discordancia de la LCC ≥15% (10,1% vs 1,9% en los gemelos BC y 28,2% vs 2,8% en los gemelos MC).

Conclusiones Primero, los defectos fetales son más comunes en embarazos de gemelos MC que en los de gemelos BC. Segundo, la detección en el primer trimestre de defectos fetales en los embarazos de gemelos BC es similar a la de los embarazos con feto único. Tercero, la detectabilidad en el primer trimestre de los defectos en los gemelos MC es mayor que en los gemelos BC. Cuarto, en los embarazos de gemelos con un defecto fetal, hay mayor discordancia entre los gemelos en la LCC y prevalencia de una mayor TN, pero la eficacia predictiva del cribado mediante estos marcadores es escasa.

妊娠11-13周常规超声检测中双胎胎儿缺陷诊断

摘要

目标：审核评估妊娠11-13周常规检测在双胎胎儿缺陷诊断中的表现，审核评估在发现胎儿缺陷的孕妇中，（对照正常胎儿）宫颈半透明厚度（NT）≥95%和≥99%或冠臀长（CRL）脐带缠绕≥10%和≥15%的机率是否增加了。

方法：这是一项根据标准协议，针对2002至2019年间妊娠11-13周接受常规超声胎儿解剖学检测双胎孕妇的前瞻性收集数据回顾分析。排除了已知染色体异常的妊娠孕妇。胎儿缺陷最终诊断依据是（活产孕妇）产后检查结果，以及（终止妊娠、流产或死产孕妇）最后一次超声检查结果。明确了妊娠11-13周检测在胎儿缺陷诊断中的表现。

结果：研究对象为6366名双胎妊娠孕妇（两个为妊娠11-13周活胎孕妇），包括4979（78.2%）个双绒毛膜（DC）和1387（21.8%）个单绒毛膜（MC）双胎孕妇。主要研究结果：第一，MC双胎孕妇胎儿缺陷总发生率高于DC双胎孕妇（2.8%对 1.3%）；第二，孕早期MC双胎孕妇胎儿缺陷检出率高于DC双胎孕妇（52.6%对27.1%）；第三，妊娠11-13周检测胎儿缺陷检出模式（总是可以检出、有时可以检出、总是无法检出）类似于之前报告的单胎妊娠检出模式；第四，总是可以检出的缺陷包括无颅、无前脑畸形、脑膨出、坎特雷尔五联症、脐凸出、体柄异常、胎动脉反向灌注综合征和连体双胞胎；第五，缺陷胎儿NT≥95%发生机率高于正常胎儿（DC双胎孕妇，16.5%对4.5%；MC双胎孕妇，19.2%对5.9%），NT≥99%也是如此（DC双胎孕妇，8.3%对1.0%；MC双胎孕妇，15.4%对2.0%）；第六，缺陷胎儿CRL紊乱≥10%发生机率高于正常胎儿（DC双胎孕妇，20.2%对7.9%；MC双胎孕妇，33.8%对9.3%），CRL紊乱≥15%也是如此（DC双胎孕妇，10.1%对1.9%；MC双胎孕妇，28.2%对2.8%）。

结论：第一，相比DC双胎孕妇，MC双胎孕妇出现胎儿缺陷的机率更高。第二，DC双胎孕妇孕早期发现胎儿缺陷的机率与单胎妊娠相当。第三，MC双胎孕妇孕早期胎儿缺陷检出率高于DC双胎孕妇。第四，在检出胎儿缺陷的双胎孕妇中，CRL脐带缠绕机率较高，NT增加机率较高，但基于这些标志物的筛检预测性能较差。