Best Practice & Research Clinical Obstetrics and Gynaecology 28 (2014) 341–353

Contents lists available at ScienceDirect

Best Practice & Research Clinical Obstetrics and Gynaecology

journal homepage: www.elsevier.com/locate/bpobgyn

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Detection of structural abnormalities in the first trimester using ultrasound

Harm-Gerd Karl Blaas, MD, PhD, Professor Dr. med. [a,b,*]

[a] Department of Laboratory Medicine, Children's and Women's Health, Norwegian University of Science and Technology, National Center for Fetal Medicine, Norway
[b] Department of Obstetrics and Gynecology, University Hospital Trondheim, Norway

Keywords:
structural abnormalities
embryo
foetus
first trimester
sonoembryology

During the past 25 years, embryonic and early fetal ultrasound and diagnosis have increasingly gained attention in pregnancy care. Modern high-frequency ultrasound transducers make it possible to obtain detailed images of the early conceptus and its organs, and thus move part of the anatomy and anomaly scan from the second to the first trimester. Today, detection of embryonic and fetal structural abnormalities in the first trimester has frequently been reported. One has to distinguish between diagnosis during the early period until about 10 weeks when the embryo or early fetus is small and transvaginal ultrasound is applied, and diagnosis during the late period at the nuchal translucency screening, usually carried out using transabdominal ultrasound. Early first-trimester abnormalities are often diagnosed by chance on clinical indications, whereas late first-trimester diagnoses are the result of systematic screening using ultrasound markers.

Introduction

The aims of any early pregnancy ultrasound scan should be to determine viability and age of the embryo or fetus, to detect multiples and describe chorionicity and amnionicity, and to detect gross abnormalities. At 10 postmenstrual weeks, an embryo is less than one-half the length of an adult thumb, but possesses already several thousands of identified structures, practically any of which may be subject to developmental deviations [1]. Holoprosencephaly, for example, develops early in

* Tel.: +47 72 57 59 25.
E-mail addresses: harm-gerd.blaas@ntnu.no, harm_gerd.blaas@mac.com.

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http://dx.doi.org/10.1016/j.bpobgyn.2013.11.004

embryonic life, and has been diagnosed by light-microscope in 201 cases of 44,000 early pregnancy terminations and miscarriages before 10 weeks [2]. Thus, the embryonic period proper is of importance because most congenital anomalies make their appearance during that time, and it should be possible to detect major structural anomalies by ultrasound.

Over the past 25 years, embryonic and early fetal ultrasound and diagnosis has gained increased attention in pregnancy care. It started with the general introduction of transvaginal ultrasound transducers, which by its improved resolution made it possible to develop the research field 'sonoembryology' [3–8]. At the same time, the first first-trimester diagnoses of structural anomalies were reported [9–13].

When we talk about detection of first-trimester structural abnormalities, the definition of gestational age is important: a trimester corresponds to a period of 3 months, and trimesters are used to divide pregnancies into three periods of approximately equal length. An exact definition of the transition between the trimesters is not possible, because no specific developmental stages would indicate such borders [1]. The introduction of the 11–13+6 weeks scan has established a practical break of the end of the first trimester at 13+6 to 14+0 weeks. In addition, we have to make a distinction between diagnoses made during the 11–13+6 week scan in a screening situation, and those made before this period during the embryonic period on clinical indications. The prerequisite for any early diagnosis and the understanding of how and when anomalies can be detected later in pregnancy is the knowledge of the normal development of the human embryo [14] and the corresponding sonographic appearance [3–5]. The transvaginal approach should be preferred for examinations before 11–12 weeks gestation.

In this chapter, we present an overview of normal development and review the relevant literature on early diagnoses of structural abnormalities. All statements of gestational age are based on the last menstrual period, expressed in completed weeks and completed days, assuming a regular cycle with ovulation at 2 weeks 0 days.

Normal development in the first trimester

The human embryo develops from the fertilised ovum, through the bilaminar and three-laminar disc, into a cylindrical body, and only at the end of the embryonic period does it look like an immature human being. Except for the physiological herniation of bowel into the umbilical coelom from 7–11 weeks, the body wall is already established after the folding process during weeks 4 and 5.

Measurement of the conceptus is important, because healthy embryos and their associated structures, such as the amniotic cavity, show virtually identical growth velocities [15]. In addition, embryos of the same size (crown-lump length [CRL]) look identical. The development of the size of embryos and the shape of the body and the brain cavities [15] are in accordance with images from the embryological literature [14]. All these facts can be used as the basis for early sonographic assessment. The synopsis in Table 1 shows the normal development of the embryo based on sonoanatomic descriptions of longitudinal two-dimensional studies [3–5,16] and on three-dimensional ultrasound studies [17,18] week by week (Table 1) [19–21].

Structural abnormalities during embryonic period

There are many clinical indications for an early ultrasound examination, such as threatened abortions (e.g. bleeding, pain), suspicion of ectopic pregnancy, uncertain pregnancy length, survey after assisted fertilisation, and targeted examination of anatomy because of maternal disease (e.g. diabetes), hereditary disease, known exposition to teratogens (e.g. anti-epileptica and lithium), and previous pregnancy with fetal anomaly. The patient-in-the-patient (the embryo), is the centre of attention. The sonographer's professional aim should be to diagnose abnormal development, if it is present, to prevent unnecessary harm to the woman, embryo or fetus. Any significant deviation from the developmental schedule described in Table 1, including abnormal biometry, can be the expression of a structural abnormality.

All first-trimester miscarriages usually show sonographic abnormalities, such as the yolk sac being too large or too small, the amniotic cavity being too large, or the embryonic pole being too small, abnormal, or the heart rate being too slow. Of interest, are those abnormalities in which the embryo