

Specific Antenatal Ultrasound Findings

Guidelines
for Health
Professionals
in Wales

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Summary

The Wales Antenatal Screening Project baseline study identified there were different policies between Trusts in Wales on some specific ultrasound findings seen on a routine fetal anomaly scan. These findings are referred to within this guidance as ultrasound ‘markers’.

Two all Wales multiprofessional workshops were organised and a working group established by the Wales Antenatal Screening Project in response to the request of obstetricians, radiologists, sonographers and midwives for assistance in reaching a consensus on how these features should be interpreted.

This guidance, which has been consulted on widely, concludes that:

- The following ultrasound findings may have an association with a number of conditions and require follow up or further investigation, suggestions of which are given in this guidance:
 - **Mild Ventriculomegaly**
 - **Nuchal Thickening** (also known as Nuchal Oedema or Nuchal Pad)
 - **Pelvi-calyceal Dilatation (PCD)**
 - **Echogenic Bowel** (also known as Echogenic Gut)
- Other ultrasound ‘markers’ discussed in this guidance (i.e. shortened¹ femur, cardiac echogenic focus, sandal gap toes and choroid plexus cysts) are of uncertain significance, and do not need to be reported when seen together or in isolation if they are seen in the absence of other structural abnormalities.
- Routinely offering women a separate ultrasound ‘marker’ scan as part of the Down’s syndrome screening programme is not recommended (NICE 2003) and should only be undertaken within the context of a formally consented research study.

¹ Other short limb conditions may require reporting and investigation.

1.0 Introduction

Many fetuses with a major chromosomal abnormality have isolated or multiple structural abnormalities which can be recognised on the 18 to 20 week ultrasound scan and will always require reporting and appropriate follow-up. Guidance on major structural abnormalities is not discussed further in this guidance.

Certain features that are associated with an increased risk of a range of chromosomal or anatomical problems can sometimes be seen on a routine fetal anomaly ultrasound scan and are referred to in this guidance as ultrasound ‘markers’. Ultrasound ‘markers’ can be defined as mainly non-permanent structural changes within the fetus which, in the absence of underlying serious pathology, appear to have very little, if any, long term significance.

Whilst there is an association between the presence of multiple ultrasound ‘markers’ and aneuploidy, in most cases the increased risk is relatively small and less significant than other factors.

The Wales Antenatal Screening Project baseline study identified different policies between Trusts in Wales on reporting ultrasound ‘markers’. The relationship between ultrasound ‘markers’ and Down’s syndrome screening has been the cause of considerable debate.

Two all Wales multiprofessional workshops and a working group were organised by the Wales Antenatal Screening Project in response to the desire of obstetricians, radiologists, sonographers, and midwives for assistance in reaching a consensus on how these features should be interpreted and reported.

2.0 Baseline Fetal Anomaly Scan

It is recommended that all women in Wales should be offered a fetal anomaly scan at 18 to 20 weeks of pregnancy as described in the Royal College of Obstetricians Ultrasound Guidance (RCOG 1997).

Scanning specifically for ultrasound ‘markers’ is not part of the recommended routine baseline fetal anomaly scan, but ultrasound ‘markers’ are occasionally, unavoidably, seen during the scan.

The potential for maternal anxiety and difficulty in counselling women when ultrasound ‘markers’ or features are reported and the need for a consistent approach across Wales are the principal reasons for the development of these guidelines.

3.0 **Abnormal findings which should be reported to the woman and further investigations offered**

3.1 **Echogenic Bowel (or Echogenic Gut)**

Description

Areas of increased echogenicity in the bowel (as bright as bone) are found in about 0.5% of fetuses. The cause is unknown but echogenic bowel has been linked with chromosomal anomalies, fetal viral infections and cystic fibrosis. A number of theories, such as increased proteinaceous secretions in the gut, have been postulated as a cause of Echogenic Bowel but the exact aetiology is still unclear.

Further Investigations

- Careful counselling with an experienced team to discuss the implications of the finding and the options available to the couple
- **Investigations** – Parental CF, consider maternal CMV IgM.
- **Diagnostic options** – Amniocentesis for karyotype, CMV, PCR and cystic fibrosis

There is insufficient evidence (NICE 2003) to offer karyotyping for Down's syndrome if echogenic bowel is present on the anomaly ultrasound.

In the presence of echogenic bowel of unknown cause, as there may be an increased risk of intra-uterine growth restriction (Carroll and Maxwell 1996), consideration should be given by Trusts to developing a local ultrasound policy of offering serial growth scans.

3.2 **Mild Ventriculomegaly**

Description

A mild dilatation of the cerebral ventricles at the atrium, measuring from 10.1 mm up to 14.9mm. Mild ventriculomegaly may be associated with fetal viral infections but also has an association with chromosomal anomaly (Russell 2000). The woman should be referred for specialist ultrasound scanning, either at a tertiary centre or a more detailed scan at a District General Hospital, and further multidisciplinary management including paediatric referral, depending on the clinical situation. Mild ventriculomegaly can be associated with a normal outcome.

- **Further investigations** – viral studies for toxoplasmosis, parvovirus and CMV
- **Diagnostic options** – amniocentesis for karyotype
- **Monitoring options** – may include follow up antenatal ultrasound scans

A measurement of the cerebral ventricles at the atrium of 15.0mm and above is classified as hydrocephalus.

3.3 Nuchal Thickening (sometimes known as Nuchal Oedema or Nuchal Pad) of 6mm or more² at the time of the second trimester anomaly scan

Description

Nuchal thickening is assessed by measuring the gap between the skin and occipital bone at the posterior aspect of the neck.

Nuchal thickening is found in about 0.5% of fetuses at 20 weeks and may be of no pathological significance (Snijders & Nicolaides 1996). It is however associated with chromosomal defects, (particularly Down's syndrome), cardiac anomalies, infection such as parvovirus, Rhesus incompatibility or genetic syndromes. Nuchal thickening may also be present in hydrops fetalis, along with pleural and pericardial effusions, ascites and generalised skin oedema.

- **Diagnostic options** - amniocentesis for karyotype
- **Other investigations** - rhesus testing, parvovirus and CMV

Pelvi-calyceal Dilatation (PCD)

3.4 Description

The presence of mild unilateral or bilateral PCD (an anterior/posterior diameter from 5mm up to 9.9mm) occurs in about one in 50 normal babies.

² Using a measurement of 6mm is the current practice in Wales

Management Plan

A follow up scan should be offered in the third trimester between 28 and 32 weeks and if the dilatation is >7 or 8mm (depending on local protocol), a scan will be indicated in the neonatal period (Bleakney & Duncan 2001) and should follow local protocol.

Antenatal diagnosis of PCD is useful to ensure appropriate early treatment is offered. Long-term antibiotics are often commenced following birth to prevent urinary tract infections. There is no evidence to support the offer of karyotyping to a woman whose fetus has an isolated PCD on ultrasound scanning.

4.0 Ultrasound ‘Markers’ of Uncertain Significance

4.1 Choroid Plexus Cyst (CPC)

Description

Small fluid filled spaces in the choroid plexus are seen in at least 1 to 2% of normal pregnancies and can be observed on scan as black echo-free round areas. Data currently suggests that neither size, number, or laterality of cysts influence risk (Walkinshaw 2001). CPCs do not cause any local damage or effect and will resolve (Russell 2000). They are usually of no pathological significance but, when other defects are present, there is a high risk of chromosomal abnormalities, usually Edwards syndrome (trisomy 18). In the absence of other structural findings, the chance of aneuploidy is very low and does not require follow up.

Management Plan

When CPCs are seen, as with all anomaly scans, a careful examination of the fetal anatomy should be completed to look for specific abnormalities of Edwards syndrome. If other abnormalities are not seen, the CPC is considered to be a normal finding and does not need to be reported. When other defects are present, there is a high risk of chromosomal abnormality, usually Edwards syndrome, and the woman should be offered a diagnostic test.

4.2

Other Ultrasound ‘Markers’

A variety of other ultrasound findings or ‘markers’ have been described and discussed by the all Wales workshops and working group. As a result of the two all Wales multiprofessional workshops and discussion in the working group, a consensus has been reached that there is currently insufficient evidence to support offering a woman an amniocentesis solely on the basis of the ultrasound ‘markers’ listed below:

- shortened femur (below two standard deviations)³
- cardiac echogenic focus
- sandal gap toes

Consequently it is recommended that the reporting of these ultrasound findings should not form part of the second trimester anomaly scan in Wales as it will lead to an increase in the number of amniocenteses offered or performed but will not necessarily achieve a higher sensitivity in the detection of Down’s syndrome. Where a fetus is found to have multiple abnormalities, including one or more ultrasound markers, all the findings should be reported.

4.3

Down’s syndrome serum screening

Second trimester Down’s syndrome serum screening is offered to most women in Wales. Women identified by the screening test as having an increased chance of having a child with Down’s syndrome are offered an amniocentesis. All women should be informed that if they decide to have an 18 to 20 week fetal anomaly scan, structural abnormalities might be seen which may lead to the offer of amniocentesis. This includes women who decline Down’s syndrome screening and also women who decline an amniocentesis following a Down’s syndrome screening result.

There is insufficient evidence on ultrasound ‘markers’ to support a practice of offering women a separate ultrasound ‘marker’ scan and this practice should only be undertaken within the context of a research study to which the woman has formally consented. The woman should be offered a fetal anomaly scan at the usual time (18 to 20 weeks) when the opportunity for identifying major fetal abnormalities is optimised.

³ Other short limb conditions may require reporting and investigation.

The National Screening Committee antenatal sub-group will be considering ultrasound screening (including ultrasound ‘markers’) during 2004 and this guidance will be reviewed following their report.

5.0 Information for Women

All pregnant women receiving care in Wales should receive written and verbal information on ultrasound prior to a second trimester anomaly scan being performed. The written information that women receive should state that ultrasound ‘markers’ of uncertain significance will not usually be reported.

If Echogenic Bowel, Mild Ventriculomegaly, or Nuchal thickening is seen, the woman should be informed and appropriate counselling and investigations arranged, supported by additional written information.

Pelvi-calyceal dilatation may be an indication of potential vesico-uterine reflux and the woman should be informed that further antenatal monitoring and neonatal follow up may be advised.

Choroid plexus cyst(s), shortened femur, cardiac echogenic focus and sandal gap toes should not be reported as there is insufficient evidence to directly link these findings with aneuploidy.

A population based all Wales study on ultrasound findings and their relationship to fetal anomalies, reported via CARIS, is currently being discussed.

This guidance has been developed and written by a working group following consultation with other working groups who had representation from members of the Obstetric Service and Ultrasound Services from Trusts in Wales, CARIS and the Antenatal Screening Project and the final draft has been consulted on with all NHS Trusts in Wales.

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