Down's Syndrome

It is difficult to generalise since there are more differences between people with Down's syndrome than there are similarities...

Most children with Down's syndrome will walk and talk. Many will read and write. Many go to ordinary schools, and look forward to a semi-independent life, away from the family home...

Just as there is a wide spread of abilities in the general population, the ability range of people with Down's syndrome is very wide.

Extracts from leaflet by the DSA

Aims and Objectives

- To understand the range of conditions covered by the term 'Neural Tube Defects' (NTD), their significance and incidence
- To understand the role of ultrasound in diagnosis of NTD's
- To identify factors that may affect detection
- To gain knowledge on other possible diagnoses
- To inform the practitioner on appropriate surveillance of pregnancies with no detected structural anomaly

Neural Tube Defects

- Neural tube defects occur in early embryonic life, between the 4-7th week

Abnormalities can occur anywhere along the neural tube; at the upper end they involve the brain and at the lower end the spinal column

Anencephaly

Spina bifida

Incidence and Recurrence

- Overall incidence is 1-2/1000 *(ONS 2002: MB3 No: 17)*
- 2% risk of recurrence
- Incidence is higher in women taking anti-convulsants *(Crawford 2002)*

Folic acid 400 mcgs is recommended 12 weeks prior to conception, and for the first 12 weeks of pregnancy to reduce the risk of NTD. (Higher doses recommended to prevent recurrence or for women on anti-convulsants) *(MRC)*

Diagnosis of Neural Tube Defects

- Ultrasound is the diagnostic test for NTD
- Detection of NTD by ultrasound depends on
  - Gestation
  - Visualisation
  - Sonographer skill
  - Differential diagnosis of other causes of raised AFP

Diagnosis of Neural Tube Defects

- Gestation
  - Ideally 18-20 weeks
  - Views of fetal spine may be limited by uterine position before 18 weeks gestation
- Visualisation
  - Maternal habitus may prevent good views
  - Fetal position may cause difficulty in getting required views

Diagnosis of Neural Tube Defects

Normal fetal spine in sagittal view

Examination of the head, brain and spine will be part of a full fetal anomaly scan

Diagnosis of Neural Tube Defects

Cranial Defects (Cranium Bifida)

- Anencephaly and exencephaly

  (complete or partial absence of the cranial vault and malformed brain) are the most serious NTD's and incompatible with life

- Iniencephaly

  Anencephaly and hyper-extended neck

- Craniorachischisis

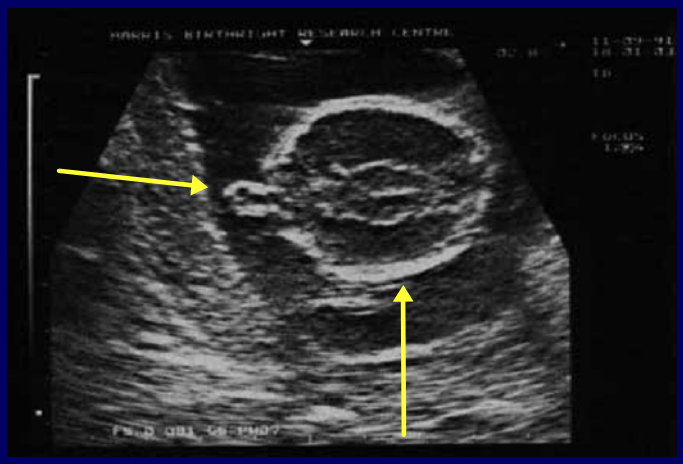
  Anencephaly and spina bifida
  Serum AFP >5.0 MoMs

Diagnosis of Neural Tube Defects

Cranial Defects (Cranium Bifida)

- Encephalocele

  Herniation of brain tissue and CSF through the skull usually covered in skin

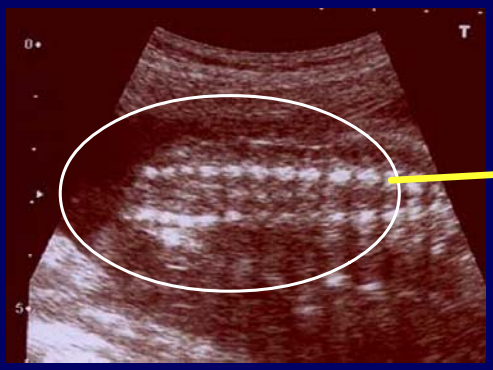


Cranial outline

Diagnosis of Neural Tube Defects
Spina Bifida

- Myelocele

  Complete failure of formation.
  The spinal cord has not sunk below the surface and lies opened out with little or no meningeal sac



Widened posterior arch of vertebrae

Diagnosis of Neural Tube Defects
Spina Bifida

- Meningocele

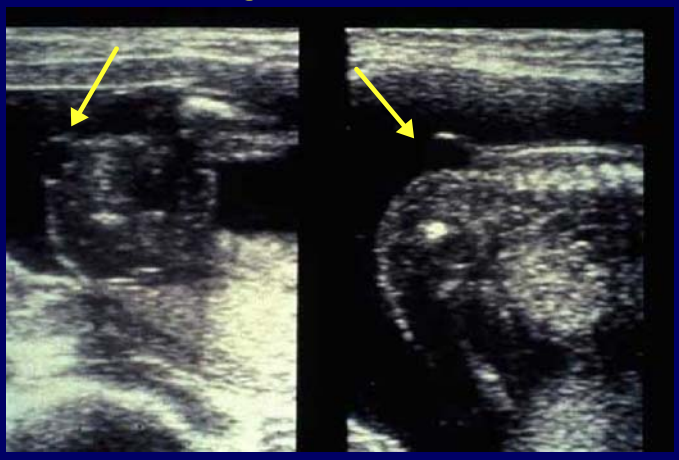
  Protrusion of the meninges through the spinal column

- Myelomeningocele

  Spinal cord and meninges protrude through the bony defect

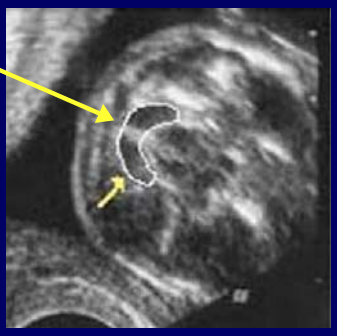
  AFP >2.5 MoM's

Meningocele



Diagnosis of Neural Tube Defects
Spina Bifida

- Other indications sometimes seen include:
  - Dilated ventricles in brain (hydrocephalus)
  - Abnormal 'lemon' shaped skull
  - Abnormal 'banana' shaped cerebellum
  - Club foot, talipes



Diagnosis of Neural Tube Defects

- Sonographer skill

  - Likelihood of detection *(RCOG 2000)*
    - Anencephaly 99%
    - Spina Bifida 90%

Spina Bifida lesions vary so smaller lesions may be harder to detect. This has an effect on the overall detection rate *(Boyd et al 2000)*

NB: Spina Bifida Occulta cannot be detected

Diagnosis of Neural Tube Defects

What does the diagnosis mean?

- The outcome of NTD's is variable
- It is not always possible to tell the degree of disability from the ultrasound findings
- 58.6% survival rate at 1 year *(Rankin et al 2000)*
- Quality of life is influenced by the general effect of the condition severity and family resources *(Cate 2002)*
- Women and their partners will need professional and specialist support following a diagnosis

ASBAH (Association of Spina Bifida and Hydrocephalus)

Problems associated with Spina Bifida

- Hydrocephalus develops in 80% of cases
- Paralysis of the lower limbs
- Joint deformities or contractures 70%
- Scoliosis 50%
- Bowel and urinary incontinence
- Renal impairment 48%
- Infections: Urinary Tract and Shunt
- Increased blood pressure 15%
- Epilepsy 9%
- Anaesthesia of the skin

(McDonnell and McCann 2000)

Differential Diagnosis

If no NTD can be found there may be other reasons for a raised AFP. These include:

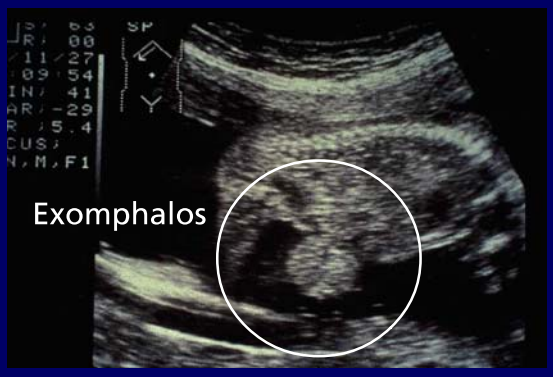
- Incorrect dates
- Multiple pregnancy
- Bleeding (frank or concealed APH)

NB: These reasons should have been excluded by a scan prior to the screening test!

Differential Diagnosis

- Abdominal wall defect
  - Body stalk anomaly
  - Exomphalos
  - Gastroschisis
- Fetal death
- Fetal nephropathy

(Chisholm et al 2001)



Differential Diagnosis

When no fetal abnormality is detected a raised AFP may indicate an increases risk of:

- Placental abruption
- Fetal growth restriction
- Pre-eclampsia
- Spontaneous miscarriage, stillbirth and infant death *(Krause et al 2001)*

First Trimester Detection of NTD's

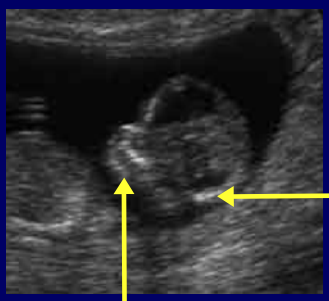
Depends on:

- Quality of visualisation and timing of dating scan - skull bones should be visible after 11 weeks
- Site and size of lesion
- Understanding of natural course of development of ultrasonographic findings
- Certainty or uncertainty of diagnosis

First Trimester Detection of NTD's

Fetal head views at 12 weeks

Normal appearance

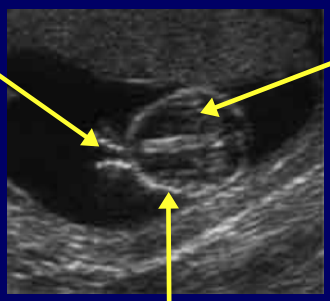


Occiput

Jaw bones

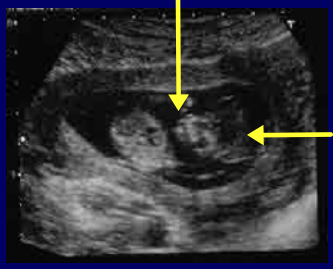
Hands

Brain

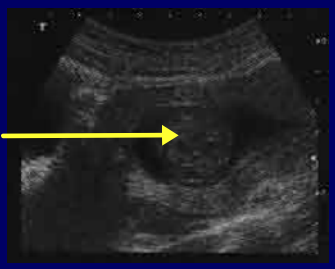


Cranial outline

Exencephaly



Brain tissue without bony covering



Summary

- Ultrasound is the diagnostic test for NTD following raised AFP
- Ultrasound scans are not specific for NTD and other conditions may be detected
- The prognosis for some abnormalities is variable
- There may still be increased risk of poor outcome for some pregnancies despite normal fetal structure
- NTD's can be detected in the first trimester