Fetal Medicine

Trisomy 13 & Holoprosencepaly

Fetal Medicine Centre
University of the Witwatersrand
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2005

- Patient details:
  - 25 year old
  - Primigravida
  - 29 weeks +2 days
  - Referred with CNS & facial anomalies

- Findings:
  - Singleton
  - Cephalic
  - Anterior high placenta
  - Normal liquor
  - 3 vessels cord

Findings:

BPD,HC,AC,FL all small for dates EFW 916g

Head: brachicephaly
Brain: holoprosencephaly
Face: hypotelorism, proboscus
Spine: Sacral spina bifida & myelomenigocoele
Heart: AVSD (septal defect)
Kidneys: Bilaterally enlarged, normal echopattern
Extremities: Bilaterally hyperextended knees, postaxial polydacytly and hyperextended wrists in both hands.

Management:

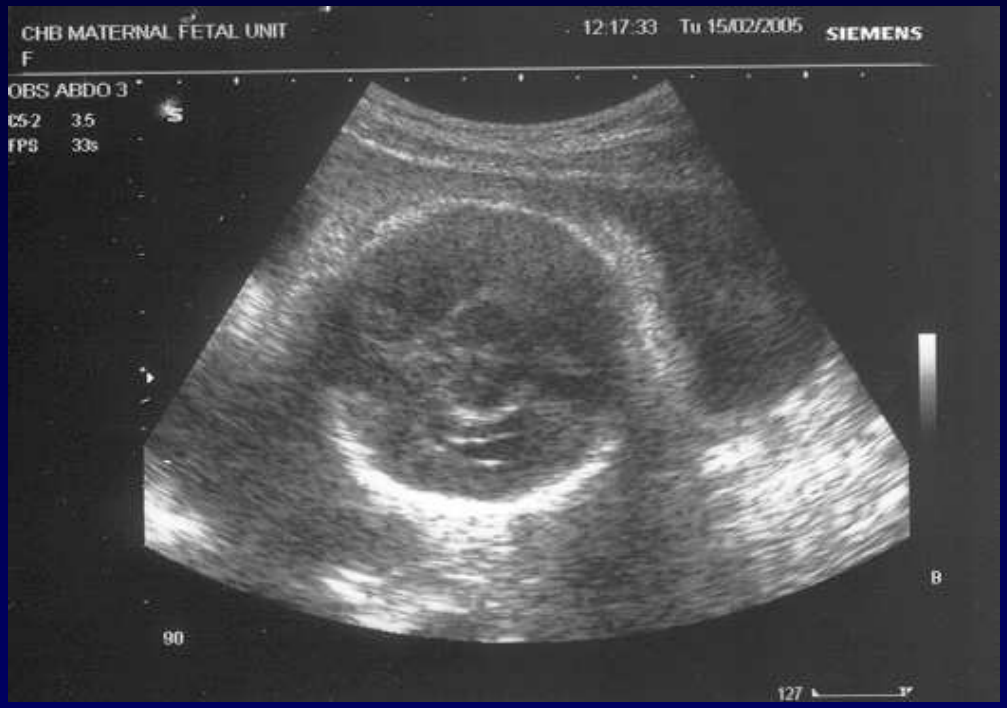
Patient was counselled about the findings and offered fetal karyotyping.

The patient opted for karyotyping and a cordocentesis was performed.

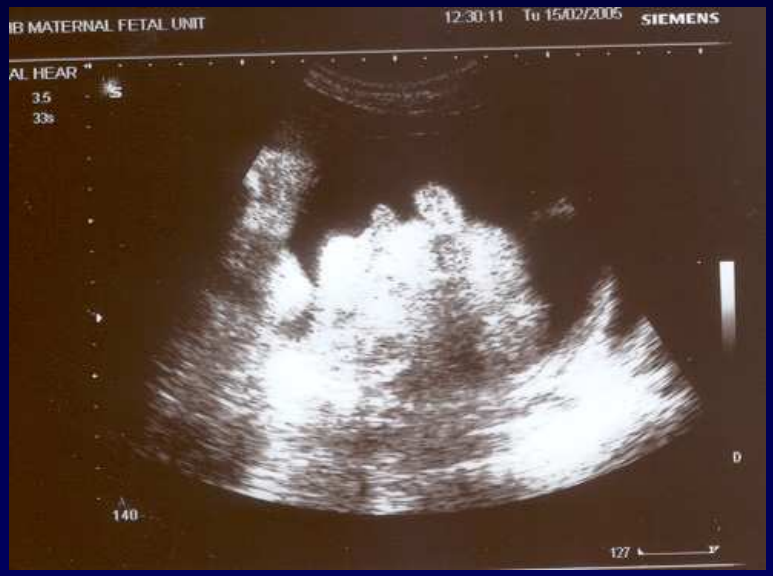
Karyotype was found to be – Trisomy 13

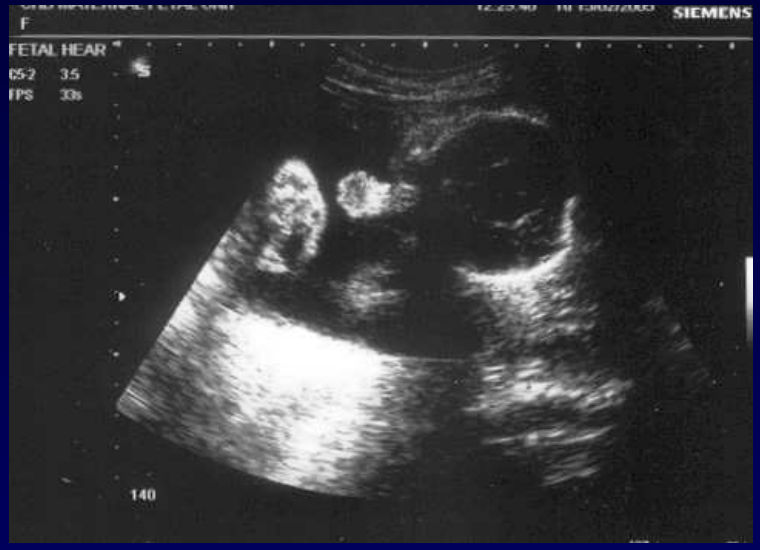
The patient was offered termination of pregnancy or continuation of the pregnancy.

Patient opted for a TOP and a fetocide was performed.

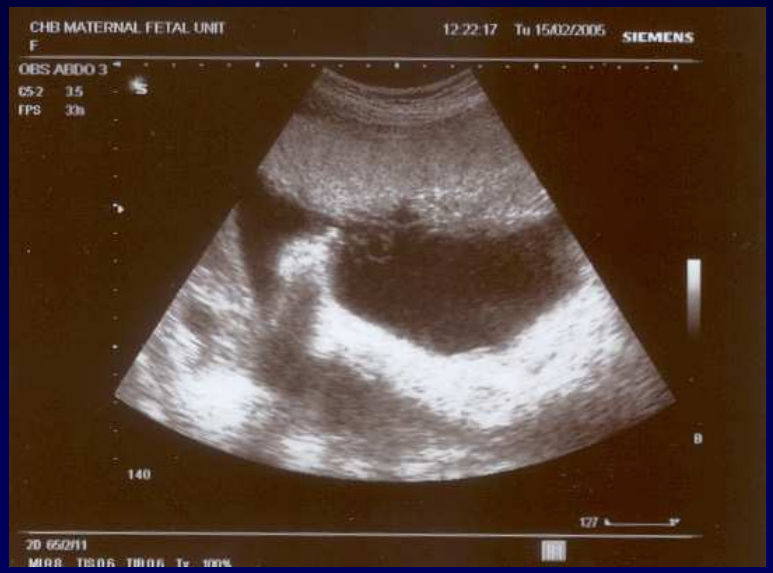


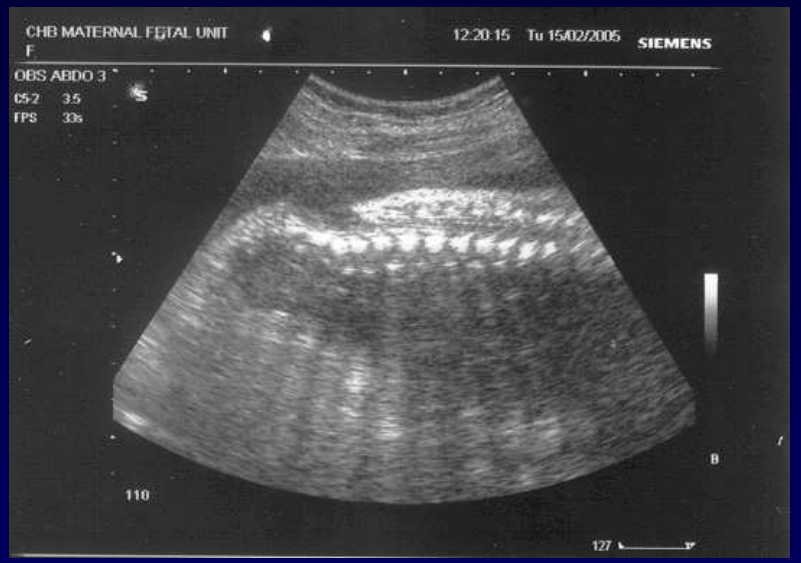
Brachichephaly & Holoprosencephaly





Proboscus on lateral and transverse facial views





Hyperextended knees, sacral NTD

- Holoprosencephaly is a spectrum of anomalies as a result of incomplete cleavage of the prosencephalon or forebrain during week 6 – 8 LMP.
- There are 3 types (depending on the degree of cleavage):
  - Alobar(most severe form)
    - Monoventricular cavity + fusion of thalami. Severe facial anomalies: hypotelorism, cyclops, proboscus.
  - Semilobar
    - Small brain rudimentary lobes, partial segmentation of ventricles and cerebral hemispheres posteriorly + incomplete fusion of thalami, absent corpus callosum. Facial anomalies.
  - Lobar
    - Normal separation of ventricles and thalami but absence of septum pellucidum. Ventricles communicate anteriorly, absent corpus callosum.
    - Mild facial deformities.

- Prevalence:
  - 1 in 10 000 births

- Etiology:
  - Chromosomal: usually Trisomy 13, also Trisomy 18, 13q-, 18p-.
  - Genetic disorders with autosomal dominant/ recessive inheritance mode.
  - Unknown.

- Recurrence:
  - For sporadic non- chromosomal holoprosencephaly the empiricl recurrence risk is 6%.

- Diagnosis:

- Alobar:
  - No Mildine (absent falx).
  - Single dilated mid- line ventricle replacing the two lateral ventricles or partial segmentation of the ventricles
  - Facial defects, hypotelorism, cyclopia, facial cleft,nasal hypoplasia, proboscus

- Semilobar:

- Small brain rudimentary lobes, partial segmentation of ventricles and cerebral hemispheres posteriorly + incomplete fusion of thalami, absent corpus callosum – (no cavum septum pellucidum). Facial anomalies.

- Lobar:
- Normal separation of ventricles and thalami but absence of septum pellucidum.Ventricles communicate anteriorly, absent corpus callosum.
- Mild facial deformities.

- Prognosis:
- Alobar and semilobar are lethal.
- Lobar: associated with mental retardation.