



The National Maternity Hospital

Vita Gloriosa Vita – Life Glorious Life

Antenatal Diagnosis and Management of Fetal Abnormalities

Peter McParland



Incidence of congenital abnormalities

- 2% to 3%
- 50% minor
- 50% major (*death, disability or major surgery*)



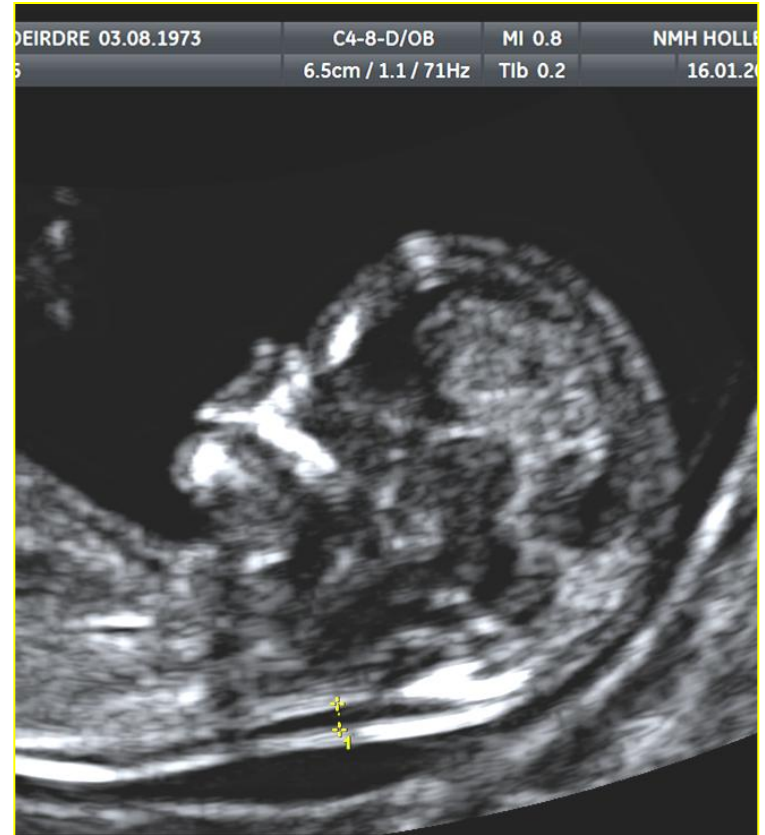
How are abnormalities diagnosed

- Ultrasound
- Invasive procedures
- Non-invasive testing



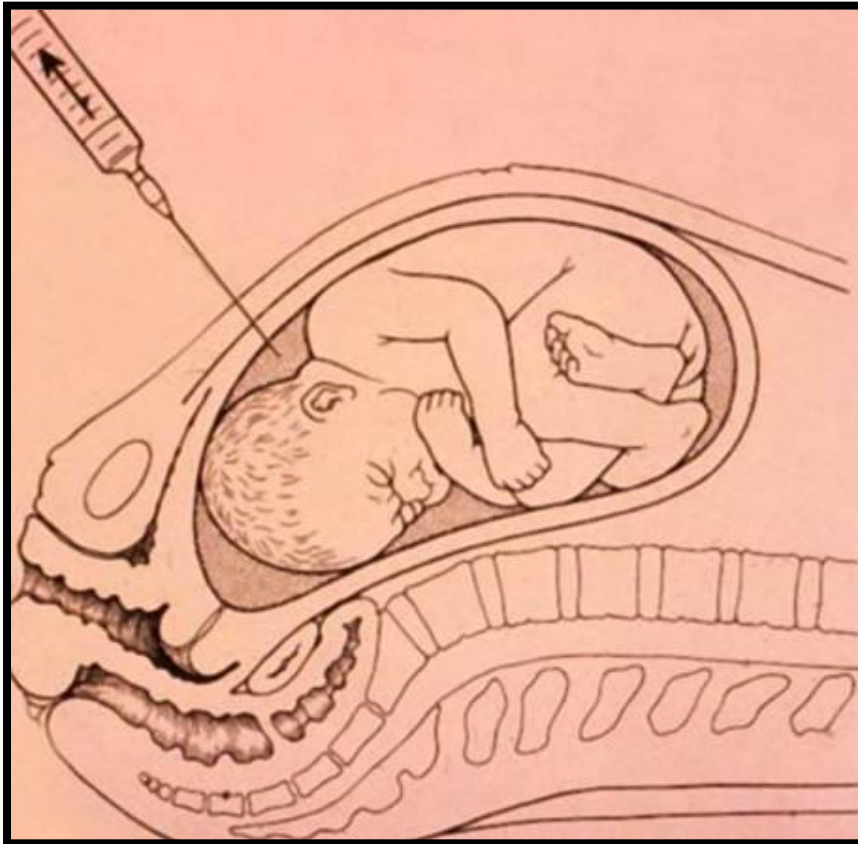
Ultrasound

- Any time during pregnancy
- Aim for 18 to 22 weeks
- This scan not available in all units
- Diagnosis depends
 - operator,
 - machine,
 - size of mother,
 - position of fetus,
 - gestation)

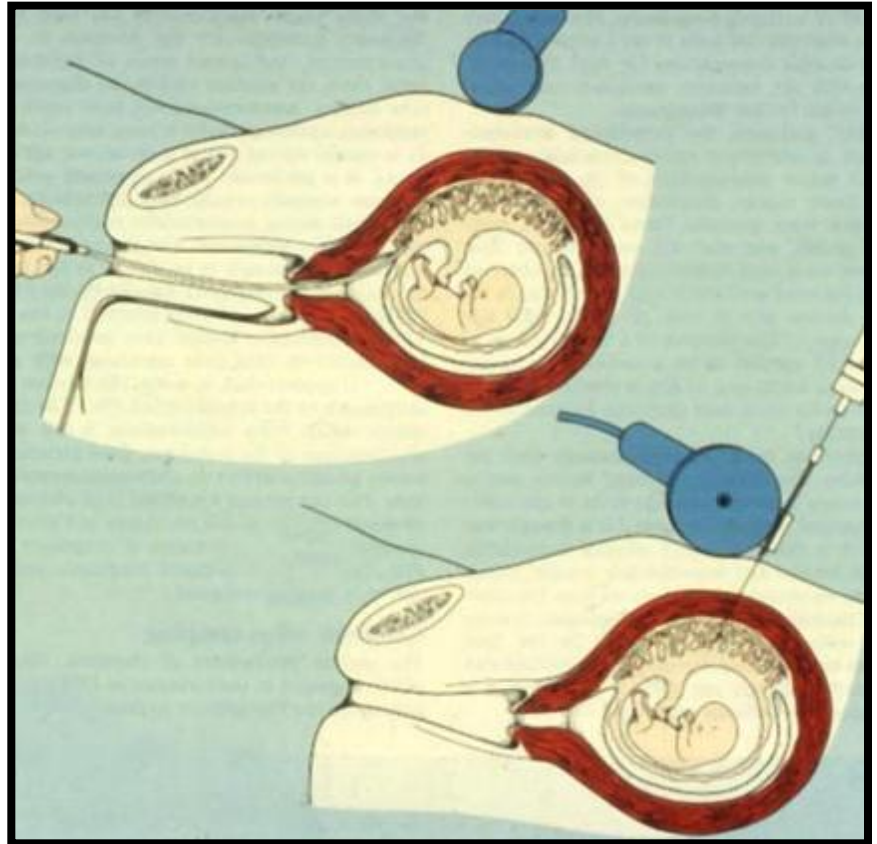


Invasive procedures

Amniocentesis
(from 15 weeks)

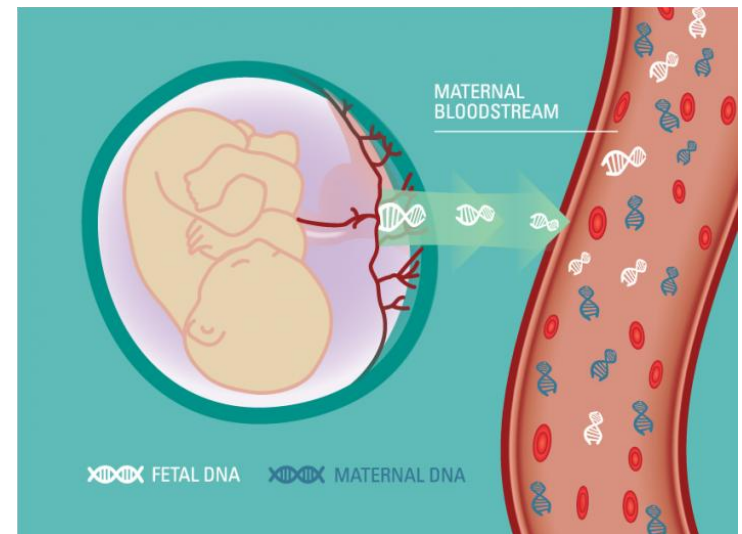


CVS (11 to 14 wks)



Non-invasive procedures (NIPT)

- New(ish) technology
- Blood test from mother
- Screening test
- >99% accuracy for Down syndrome (*and other chromosomal*)
- Not paid for by state
- Can tell gender



Medical Council Guidelines

- **8.2:** patients have the right to be offered all treatment options....
- **11.1:** you must give patients enough information, in a way that they can understand, to enable them to exercise their right to make informed decisions about their care
- **48.4:** it is lawful to give information on abortion. It is not lawful to promote or advocate...



Anencephaly

- 26yo
- 1st pregnancy
- Fit and well
- Anatomy scan at 21 weeks
- 75% survive to birth
- All die within hours/days

Anencephaly

- 26yo
- 1st pregnancy
- Fit and well
- Anatomy scan at 21 weeks
- 75% survive to birth
- All die within hours/days



Who informs the patient?

- Most frequently midwife ultrasonographer
- Referral to a fetal medicine specialist
- Consultant obstetrician with an interest
- Midwives (Prenatal diagnosis)
- Geneticist /Paediatrician



What happens next?

- Full explanation of diagnosis
- Prognosis
- Options discussed
- Non-directive counselling
- GP informed

(Many unaware of the law)



Environment

- ✓ Ultrasound room
- ✓ Quiet non-clinical room
- ✓ Involve partner
- ✓ Clear simple language
- ✓ Access to telephone
- ✓ Give time on their own/further discussion
- ✓ Offer to see/again in 48hours again



Case study

- 41yo
- 3rd pregnancy
- 1 baby with Down syndrome
- NIPT blood test at 11 weeks
- > 99% chance of Edwards syndrome
- Amniocentesis at 15 weeks confirms Edwards



Edwards syndrome

- An extra chromosome 18 (47 vs 46)
- Physical and mental handicap
- Median survival is 5 to 14 days
- 20% will live until 3 months
- 8% will survive beyond 1 year
- 1% will live to age 10



Fatal/life limiting conditions

- What is fatal? Mins? Days? Weeks? Months?
- Ultrasound looks at anatomy not function
- Can be fairly certain most of the time - but not always...



Options - Continue pregnancy

- Vast majority of overall
- Consultant provided care with specialised midwifery care
- Meet with neonatologist or specialist
- Multidisciplinary meeting (*weekly*)
- Direct telephone contact with midwife specialist
- Social work/chaplaincy/bereavement
- Plan for delivery and subsequent care



Options - Termination of pregnancy

- Patient given phone numbers (*Liverpool*)
- Patient needs to make phone call
- We provide all the information to UK unit
- Arrangement made by patient
- Cost of flights, accommodation, medical procedure borne by patient (*€1000 to €5000*)
- Post-mortem not always available
- Appointment given for postnatal follow up



Termination of pregnancy

- Early
 - Medical i.e. tablets
 - Surgical i.e. like a D and C
- Late
 - Surgical
 - Stopping heart beat and induction of labour



UK figures for Irish women 2015

- Total in UK: 185,824 – 92% less than 13 wks
- 3,451 Irish women (*every county*)
- 135 (4%) Irish women under section E (*serious physical or mental handicap*)
- 112/135 over 20 weeks



Abnormalities undergoing TOP

Chromosomal

- N = 69
 - 40 with Down syndrome
 - 13 with Edward syndrome
 - 7 with Patau syndrome
 - 9 others



Abnormalities undergoing TOP

Anatomical

- N = 66
 - Brain/spine 23
 - Anencephaly 7
 - Spina bifida 9
 - Heart 10
 - Kidney/bladder 4
 - Musculoskeletal system 13
 - Others cystic fibrosis/hydrops/SMA 15



The Future

- ✓ Better access to ultrasound
- ✓ Improved ultrasound performance
- ✓ Increased non-invasive testing – increased no of chromosomal abnormalities diagnosed
- ✓ Increased no of conditions diagnosable with a blood test



Summary

- Prenatal diagnosis widely available in Ireland
- No. of prenatal diagnosis increasing (especially NIPT)
- Options discussed non-directively
- Many continue pregnancy, 135 did not in 2015

