Antenatal Diagnosis and Management of Fetal Abnormalities

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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 \textit{(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
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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 48 hours 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