Genetic counselling for Haemophilia

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What is Genetic Counselling?

- Information
- Counselling
- Support
- Informed decision-making
Family history of Haemophilia

- What causes haemophilia?
- What about children?
- Am I a carrier?
- What about the rest of my family?
- Who will be told my result?

Is genetic testing relevant to me? What’s involved?
The family tree

Victorian Clinical Genetics Services
Carrier testing

- In the past...
  - Factor VIII/IX levels
  - **but** some carriers have normal FVIII/FIX levels
    - ➡️ not always helpful

- Today...
  - Genetic testing
  - Accurate carrier testing
Mutation detection

- Male with Haemophilia or definite female carrier
- Reasons for testing:
  - Not essential for management
  - Genotype- phenotype correlations
  - Information for other family members
    - Accurate carrier testing in females
    - Confirmatory diagnostic testing in males
Mutation detection

- There are some common mutations
- Many families have “private mutation”
- Time consuming, labour intensive, expensive
- Permission to use for other family members

Mutation identified in ~98% of cases
Accurate carrier testing

- Mutation identified
- Quick, definitive, in-expensive
- Confidential
- Genetic counselling recommended
- Carrier status prior to pregnancy
- Plasma factor measurement in carriers
Decision to have carrier testing

- Personal/family experience with Haemophilia
- Actual & perceived severity in family
- Intention to utilise reproductive technologies
- Beliefs, values & morals
- Culture & religion
Reproductive options

- Accept the 50/50 risk
  - have children (with or without ultrasound)
  - not have children (or adopt)

- Use technologies
  - chorionic villus sampling (CVS)
  - pre-implantation genetic diagnosis (PGD)
  - donor egg
Chorionic villus sampling (CVS)

- Mutation must be known
- ~12 weeks of pregnancy
- 1% risk of miscarriage
- FISH for fetal sex
- Result ~2 weeks
- Can also test for chromosome problems
- Amniocentesis not routine
- Contact genetic counsellor
Pregnancy management plan

- Pregnant patients referred if:
  - At-risk of being a carrier, but status uncertain
  - Known carrier & male baby has Haemophilia
  - Known carrier, but uncertain if the baby has Haemophilia

- Provide advice to Obstetricians regarding:
  - Delivery
  - Possible risks
  - Diagnostic testing of male infant
CASE EXAMPLE
**Peter**
Severe Haemophilia A
Currently 45 years
Inhibitor, joint disease, HIV
Mutation identified

**Harry**
Severe Haemophilia A
Joint disease
d. @ 29 years
**Lyn**

Genetic counselling prior to family planning (many years ago)

Wanted to avoid Haemophilia

Sex selection for females

Carrier status recently revealed
Sarah
Attended with mother, Lyn
Pregnant, requesting carrier testing & PND
For preparation purposes
Lyn has different opinion
**Kate**
Always believed she was a carrier
Very relieved not to be a carrier, but felt bad for her sister
Changed her sense of self
Susan
Increased responsibility
Mixed emotions
Never tested for Haemophilia
Not a carrier

Ruby
Never understood inheritance
Sense of guilt

Harry
Implications for other family members

Happy to communicate information

Family letter provided
Counselling issues

- Individual/family experience
- Factors for pregnant women
- Differing opinions of family members
- Beliefs about carrier status
- Multiple emotions
- Devaluing a life
- Family communication of information