Genetic Counselling

Robyn Shoemark
Clinical Nurse Consultant
The Children’s Hospital at Westmead
Sydney, Australia
Disclosures

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Outline

• Haemophilia Genetics
• Carrier testing and age
• HTC’s role
• Education
• Ethics
Genetics

When the father has haemophilia and the mother is unaffected

Father  Mother

XY       XX

YY       XX

None of the sons will have haemophilia. All of the daughters will carry the haemophilia gene.

When the mother carries the haemophilia gene and the father is unaffected

Father  Mother

XY       XX

YY       XX

XX       XY

There is a 50% chance at each birth that a son will have haemophilia. There is a 50% chance at each birth that a daughter will carry the haemophilia gene.

www.health.qld.gov.au
Boys vs girls

• Boys – test at young age to determine genetic mutation

• Girls – test factor levels and potentially store DNA
  – If low factor levels – preliminary carrier status determined
Carriers

- Haemophilia – X-linked
- Obligate carriers – daughters of men with haemophilia
- Daughters of carriers
- Sporadic carriers or new mutations
Obligate vs Possible

Obligate carriers are:
• all daughters of a father with hemophilia;
• mothers of one son with hemophilia and who have at least one other family member with hemophilia (a brother, maternal grandfather, uncle, nephew, or cousin);
• mothers of one son with hemophilia and who have a family member who is a known carrier of the hemophilia gene (a mother, sister, maternal grandmother, aunt, niece, or cousin);
• mothers of two or more sons with hemophilia.

Possible carriers are:
• all daughters of a carrier;
• mothers of one son with hemophilia but who do not have any other family members who have hemophilia (or are carriers);
• sisters, mothers, maternal grandmothers, aunts, nieces, and female cousins of carriers.

‘Identification of hemophilic mutations is currently used for two purposes: the definitive diagnosis of the carrier status and prenatal determination of the disease’
Who makes up the HTC

- Haematologist
- Nurse
- Social worker
- Psychologist
- Physiotherapist
- Rheumatologist
- Geneticist
- Other staff members
HTC’s Role

• Inform and educate
  – Genetic testing
  – Inheritance
  – Males with haemophilia
  – Carrier females
  – Potential female carriers
  – Extended family members
Nursing staff need to be informed and well educated as we are often the ones fielding the questions as we have the rapport with the families.
Questions

– How do we identify potential carriers
– At what age do you discuss carrier status
– At what age are potential carriers tested
– How do you broach the subject
– Who talks about it – doctors, nurses, genetics counsellor, psychologist…..
– What information is given
– When to refer
– Who to refer to
Testing

- NSW, NT & Tasmania – send to SA IMVS
- Costs – approx $1500
- Who pays – hospital/local area health
- Age >16 years

- VIC – done locally
- Progeny
- Costs – covered by government
- Age >16 years
Testing

• US – My Life our Future – free genetic mutations. Carriers now tested at all ages

• UK and London – age >16 years

• Amsterdam – weekly meetings to discuss families and family trees to encourage testing
Consent

- Informed
- State
- Country
- Funding
- Counselling
What influences testing?

• Known family history
• New diagnosis
• Severity of haemophilia
• Family planning
• Good or bad memories with relatives
• Peace of mind for young daughters
• Medical
Education

- Mothers/females educated about their own carrier status
- Daughters educated about possible carrier status and what this means
- Haemophilia boys educated about passing on carrier status to daughters
Pre-Testing Education

Education and counselling

– Identify person to be tested
– Do they want to be tested and know results
– What does being a carrier mean
– How is haemophilia inherited
– What does the test involve
– Family planning
– Possible options for pregnancies if a carrier
Post Testing Education

Education and counselling
- Results – carrier or not
- Children or not
- Natural conception or IVF/PGD
- When and options to test the pregnancy
- Keep pregnancy or terminate
When to test?

Test before pregnancy if the female wants to know her carrier status

Should we test minors and if so when?
Pregnancies

• Mother notifies HTC of pregnancy
• Cell free or Free Foetal DNA – sex determination around 10 weeks
• Known carrier – ask to confirm sex at 18-20 week ultrasound
• Female baby – deliver normally
Pregnancies

- Male baby
  - CVS to determine haemophilia or not
  - Assume haemophilia and deliver major hospital
  - No forceps or vacuum delivery
  - Test factor levels from cord
  - No intramuscular Vit K or immunisations
  - No heel pricks for newborn screening
Ethics

- When to test
- Who to test
- When and how do you inform results
- Costs
- Travel and insurance
- How and when to tell partners
- Arranged marriages
- Religious and cultural responses
Thank you
References

• www.health.qld.gov.au
• www.wfh.org/en/abd/carriers/carrier-diagnosis-en
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