Genetic Counselling

Robyn Shoemark

Clinical Nurse Consultant

The Children's Hospital at Westmead

Sydney, Australia

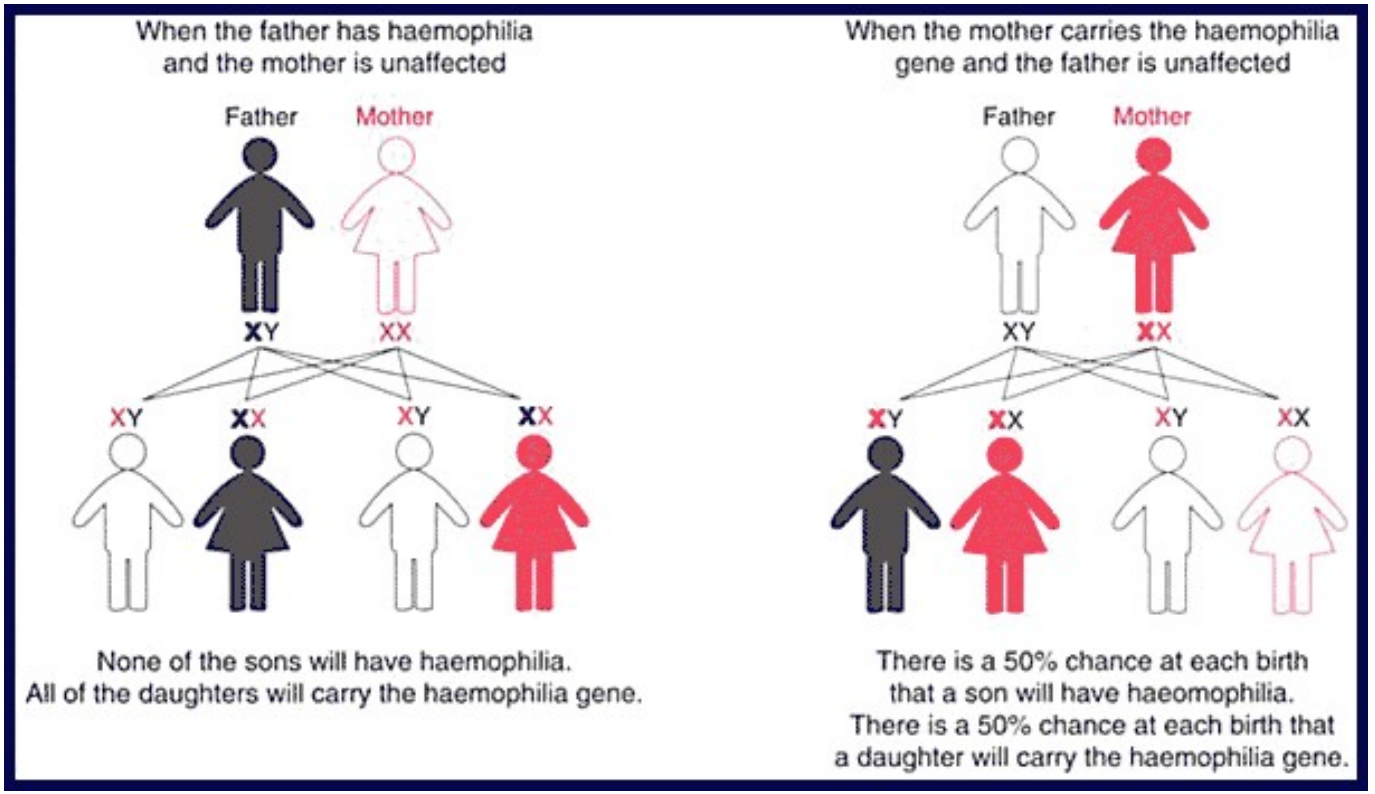
Disclosures

- Nurse Advisory Board – Pfizer, Novonordisk, Biogen Idec
- Sponsorships provided by Novo Nordisk, Pfizer, Baxalta, CSL
- Consultation work with Pfizer, Novo Nordisk, Bayer, Baxalta, CSL, Biogen Idec

Outline

- Haemophilia Genetics
- Carrier testing and age
- HTC's role
- Education
- Ethics

Genetics



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.

http://www.wfh.org/en/abd/carriers/carrier-diagnosis-en

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

Genetic sequence analysis of inherited bleeding diseases F.Peyvandi et al Blood 2013 Nov 14

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

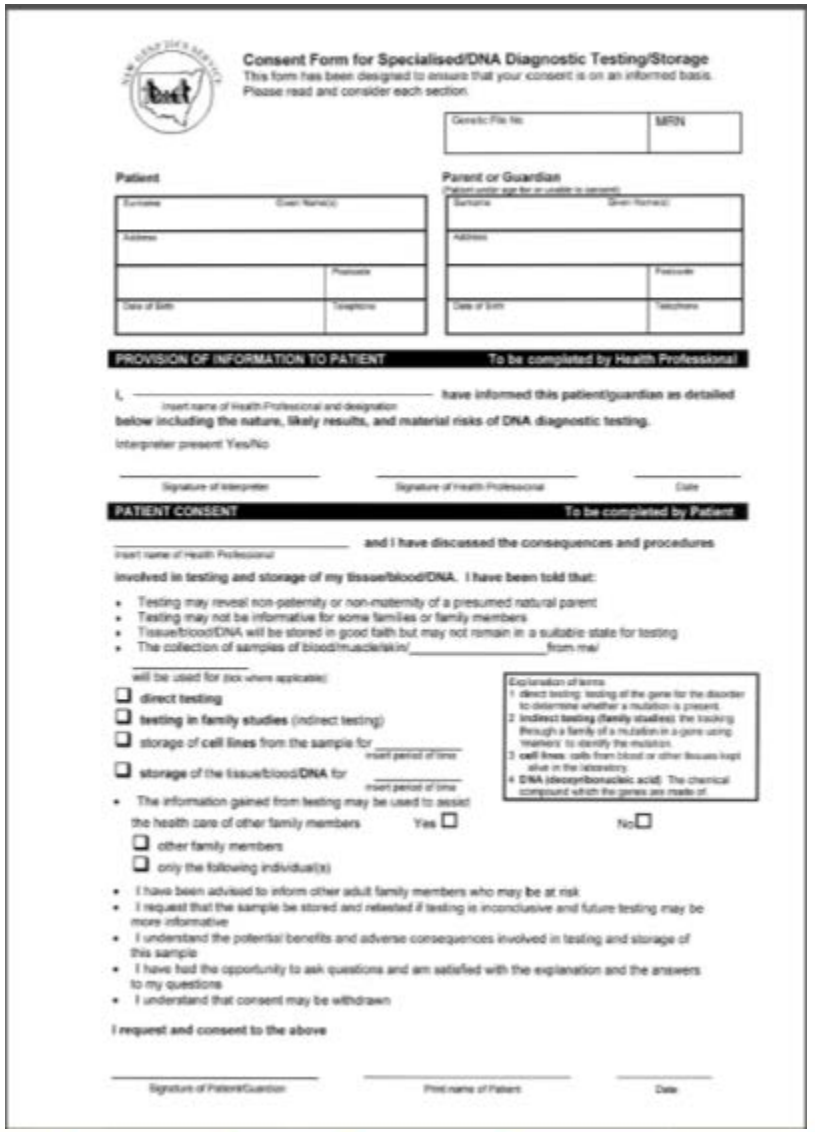
- VIC – done locally
- Progeny
- Costs – covered by government
- Age >16 years

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



Consent Form for Specialised/DNA Diagnostic Testing/Storage

This form has been designed to ensure that your consent is on an informed basis. Please read and consider each section.

Genetic File No. | MRN

Patient

Surname | Given Name(s)
Address
Postcode
Date of Birth | Telephone

Parent or Guardian

Surname | Given Name(s)
Address
Postcode
Date of Birth | Telephone

PROVISION OF INFORMATION TO PATIENT — To be completed by Health Professional

I, _____ (Insert name of Health Professional and designation) have informed this patient/guardian as detailed below including the nature, likely results, and material risks of DNA diagnostic testing.

Interpreter present Yes/No

Signature of Interpreter | Signature of Health Professional | Date

PATIENT CONSENT — To be completed by Patient

_____ (Insert name of Health Professional) and I have discussed the consequences and procedures involved in testing and storage of my tissue/blood/DNA. I have been told that:

- Testing may reveal non-paternity or non-maternity of a presumed natural parent
- Testing may not be informative for some families or family members
- Tissue/blood/DNA will be stored in good faith but may not remain in a suitable state for testing
- The collection of samples of blood/muscle/skin/_____ from me/_____ will be used for (tick where applicable)

☐ **direct testing**
☐ **testing in family studies** (indirect testing)
☐ storage of **cell lines** from the sample for _____ (insert period of time)
☐ **storage** of the tissue/blood/**DNA** for _____ (insert period of time)

Explanation of terms
1 direct testing: testing of the gene for the disorder to determine whether a mutation is present.
2 **indirect testing (family studies)**: the tracking through a family of a mutation in a gene using markers to identify the mutation.
3 **cell lines**: cells from blood or other tissues kept alive in the laboratory.
4 **DNA (deoxyribonucleic acid)**: The chemical compound which the genes are made of.

- The information gained from testing may be used to assist the health care of other family members Yes ☐ No ☐
  - ☐ other family members
  - ☐ only the following individual(s)
- I have been advised to inform other adult family members who may be at risk
- I request that the sample be stored and retested if testing is inconclusive and future testing may be more informative
- I understand the potential benefits and adverse consequences involved in testing and storage of this sample
- I have had the opportunity to ask questions and am satisfied with the explanation and the answers to my questions
- I understand that consent may be withdrawn

I request and consent to the above

Signature of Patient/Guardian | Print name of Patient | Date

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

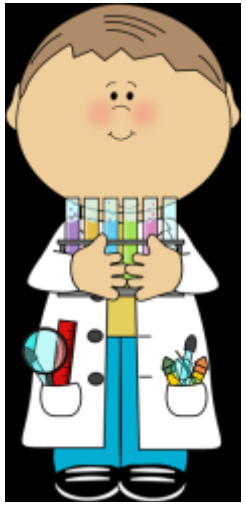
- Identify person to be tested
- Do they want to be tested and know results
- What does being a carrier mean
- How is haemophilia inherited
- What does the test involve
- Family planning
- Possible options for pregnancies if a carrier

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?



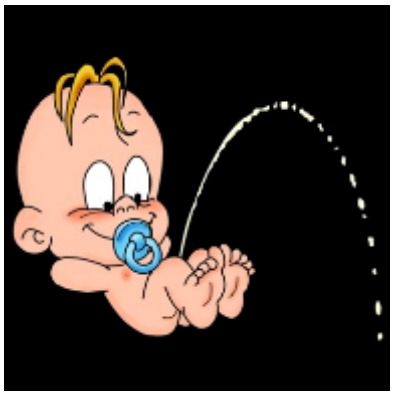
Test before pregnancy if the female wants to know her carrier status

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
- 'Genetic sequence analysis of inherited bleeding diseases' F.Peyvandi, T.Kunicki, D.Lillicrap. Blood 2013 Nov 14
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