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# Attitudes towards and beliefs about genetic testing in the haemophilia community: a qualitative study

*Monash Centre for Ethics in Medicine and Society*

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# Objectives

- Haemophilia is not only a disorder of blood clotting factors
  - Genetic testing is a big deal in the haemophilia community
  - The community context is important
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# Overview

- Genetic counselling and genetic testing
    - Technical issues
    - Social and ethical issues
  - What do we know?
  - What do we not know?
  - What can we do?
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# What do we know about genetic testing?

Genetic testing is a method to screen, select and identify the gene mutation that causes a disorder.

- Several ways to perform genetic testing
    - Carrier testing
    - Pre-implantation genetic diagnosis
    - Prenatal
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# What do we not know about genetic testing?

- Limited knowledge about:
    - Testing for carrier and antenatal purposes
    - Utilisation of genetic counselling
  - Attitudes towards predictive/genetic and carrier testing have been studied for other conditions
    - Huntington's disease, cystic fibrosis, deafness and breast cancer
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# Aims

1. Clarify the social and ethical implications of genetic testing in the haemophilia community
  2. Assess the knowledge, attitudes, beliefs and needs of this community with respect to genetic testing
  3. Identify issues and problems that may arise as a result of the introduction of genetic testing into the haemophilia community
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# Methods

- Sites
    - The Royal Children's Hospital, Melbourne
    - The Alfred, Melbourne
  - Individual interviews
    - Face to face
    - Telephone
  - 39 participants
  - Analysis of interview transcripts
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# Results

## Themes

1. Experiences relating to having a condition of genetic origin
  2. Experiences relating to haemophilia itself, including its effects and treatments
  3. Issues relating to genetic counselling and testing
  4. Outcomes and implications of genetic testing
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# 1. Experiences relating to having a condition of genetic origin

- **The genetic condition as a mark of uniqueness and identity**

*“It doesn’t feel...it feels normal, like mum said, you kind of feel special” (Carrier)*

- **Carrier guilt**

*“Very very hard for me because I blamed myself terribly, even though my head knew that I wasn’t responsible but my heart wouldn’t let it go...” (Carrier)*

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## 2. Experiences relating to haemophilia itself, including its effects and treatments

- **Influence of haemophilia on education and growing up**

*“I guess it really started to impact on me once I sort of started going to school not being able to participate in a lot of activities that the so called normal kids were able to take part in...”*

(Person with haemophilia)

- **The positive outlook within families**

*“[her husband] He became Mr Mum when I went back to work...he treats him at home and is really good...[the treatments have become] a whole family affair and it is like he [my son] has his treatment and he is off”*

(Non-carrier)

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### 3. Issues relating to genetic counselling and testing

- **Obstetric purposes**

*“...as a safety precaution I think that needs to be known”*  
(Person with haemophilia)

- **Genetic counselling**

*“They didn’t really give us a lot of information to be honest...we could have been told a little bit more”* (Father of a son with haemophilia)

- **Strong cultural influences**

*“You just can’t make that decision in isolation of everything else it has come from you...your values and your culture.”*  
(Carrier)

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## 4. Outcomes and implications of genetic testing

- **Abortion and embryo selection**

*“I chose not to find out before he was born whether he has haemophilia because like termination wasn’t an option for me”  
(Carrier)*

- **Severity of haemophilia**

*“I have seen kids with inhibitors...it can affect their lives, that would be my concern. [It] is manageable for us because [my son] doesn’t present as severe but if he did present as severe I think that it would change the dynamic of it” (Non-carrier)*

- **Financial implications**

*“...I cant afford to pay for it...I am quite happy to have a child and to keep going down that path that I am a carrier lets just forget about the testing...keep going assuming that I am a carrier”  
(Non-carrier)*

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# Conclusions

- There is strong support in the community for genetic testing
  - Proposed uses of testing are very variable
    - Familial or Spontaneous haemophilia
    - Carrier status
    - Severity
  - Issues with respect to genetic counselling need to be addressed
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## Next phase...

**The social and ethical dimensions of genetic testing: a longitudinal study of the haemophilia community.**

- Invitation to participate
    - The Alfred hospital, Melbourne
    - The Royal Children's Hospital, Melbourne
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