



# HAEMOPHILIA CARRIERS

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# WHY BOTHER TESTING?

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- Safe delivery for your baby –  
Incidence of intra or extracranial haemorrhage in newborn with haemophilia is 4%
- 30% of carriers have lower than normal levels.



# What does it mean to be a carrier of haemophilia?

- **HAVING CHILDREN**
- **BLEEDING**

# WHO SHOULD TEST?

## FAMILY HISTORY





# New Mutation

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~30 % of cases of hemophilia are new mutations.  
No family history.



# How to diagnose being a Haemophilia Carrier

GENETIC TESTING



# Clinic

When - above 16 years

- ✓ For education regarding genetics (partners included)
- ✓ Prior to planning a pregnancy
- ✓ When pregnant and unsure about carriership
- ✓ After the unpredicted birth of a haemophilia boy.



# Inheritance of Hemophilia

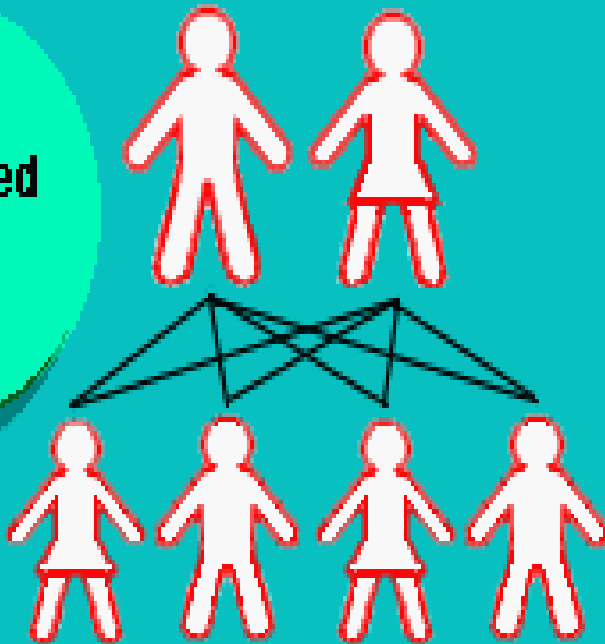
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- Hemophilia A and B are X-linked recessive disorders
- Hemophilia is typically expressed in males (XY) and carried by females (XX)
- Severity level is consistent between family members
- ~30 % of cases of hemophilia are new mutations



# Genetics

Unaffected  
Family

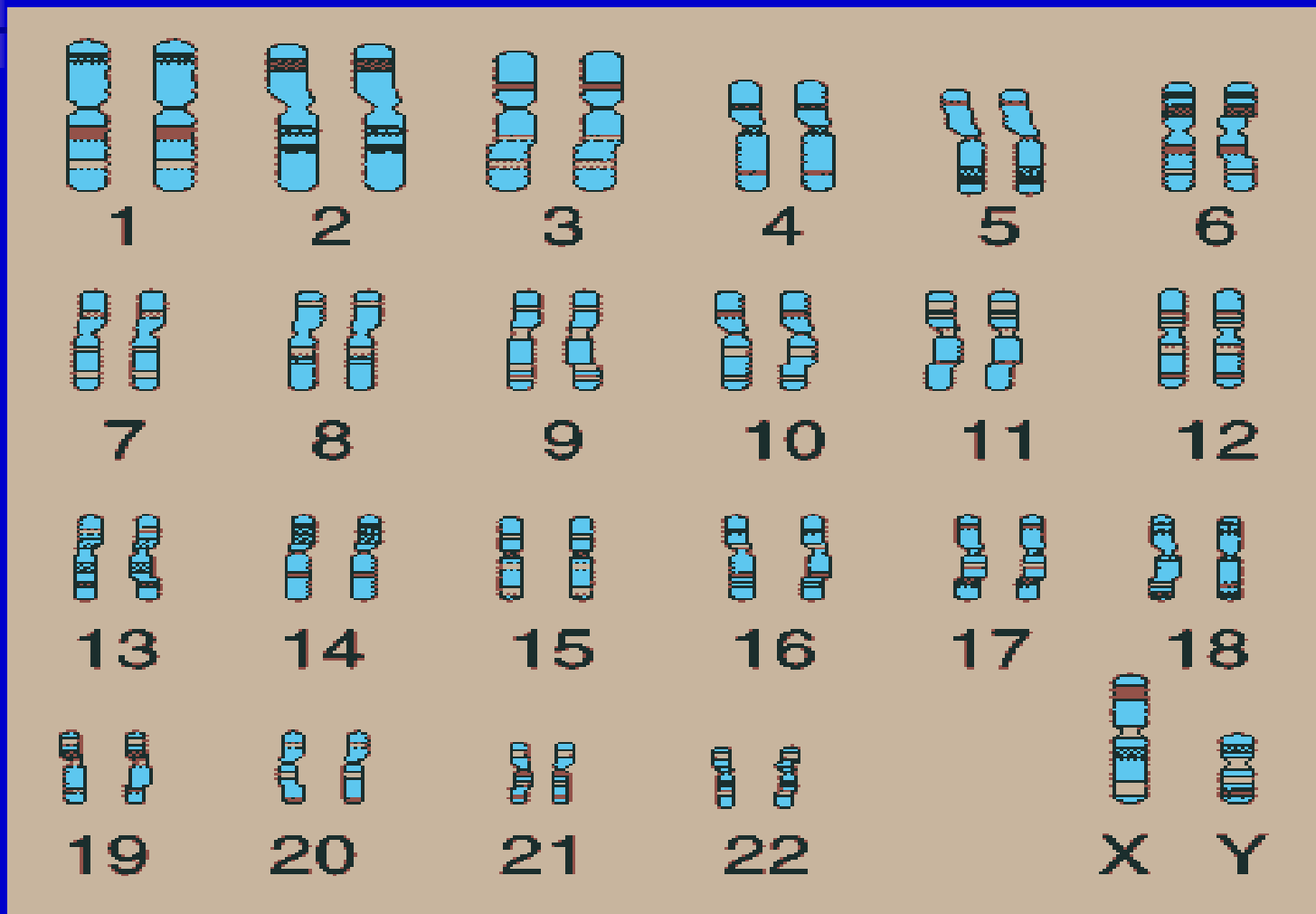


- Two chromosomes determine the sex of an individual, X and Y.

- Female XX

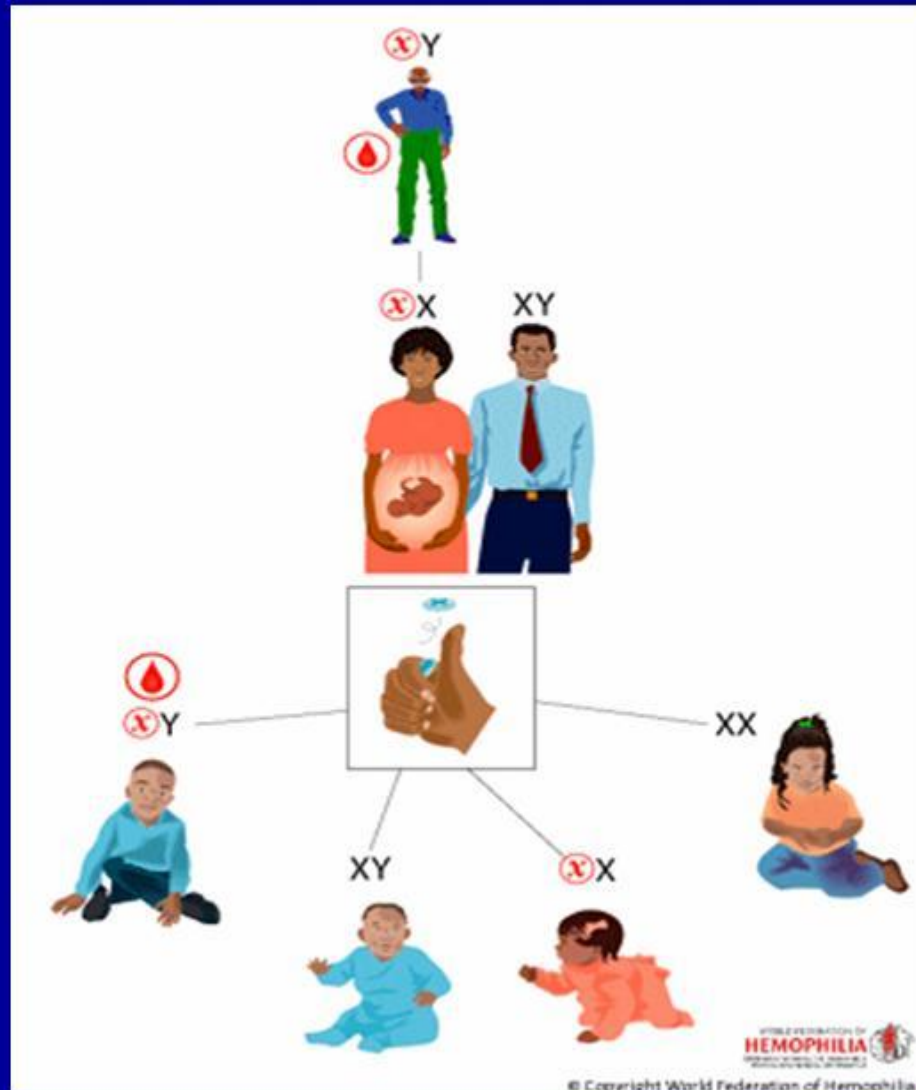
- Male XY

# 23 Pairs of Chromosomes



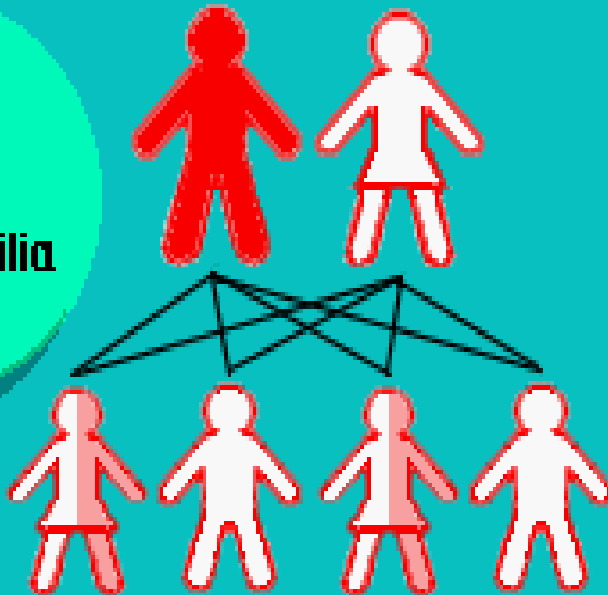
# Hemophilia

## Genetic Transmission



# Father with Haemophilia

Father  
with  
haemophilia



- Genetic defect causing haemophilia on X chromosome
- Daughter of man with haemophilia must inherit his X chromosome, therefore she is an obligatory carrier.
- Sons of a man with haemophilia will not be affected as they inherit fathers Y chromosome which does not carry haemophilia gene.

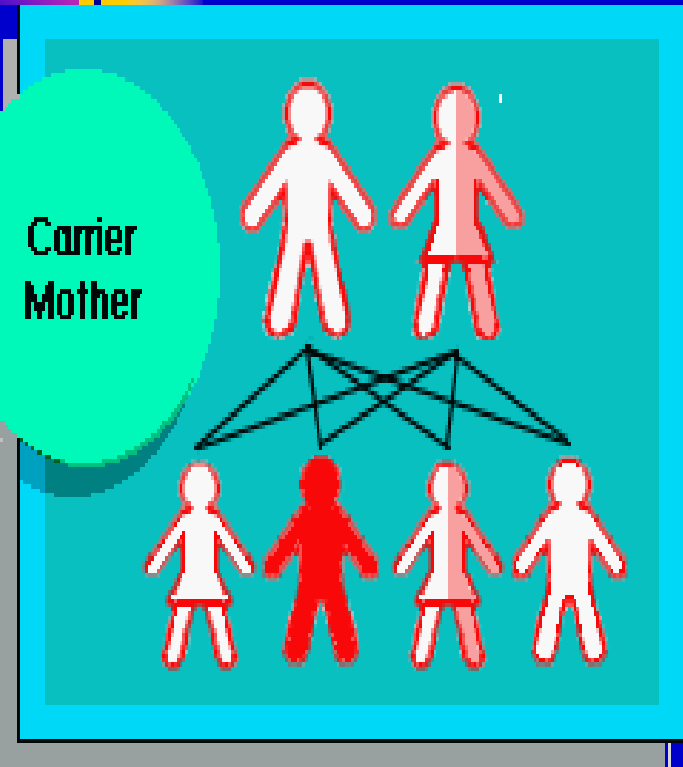


# Obligate carriers

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- Father has haemophilia ( they must have inherited an affected X chromosome from him)
- Two sons with haemophilia ( two spontaneous mutations are not likely to happen)
- One son with haemophilia and a blood relative who is known to have or to carry haemophilia.

# Carrier Mother



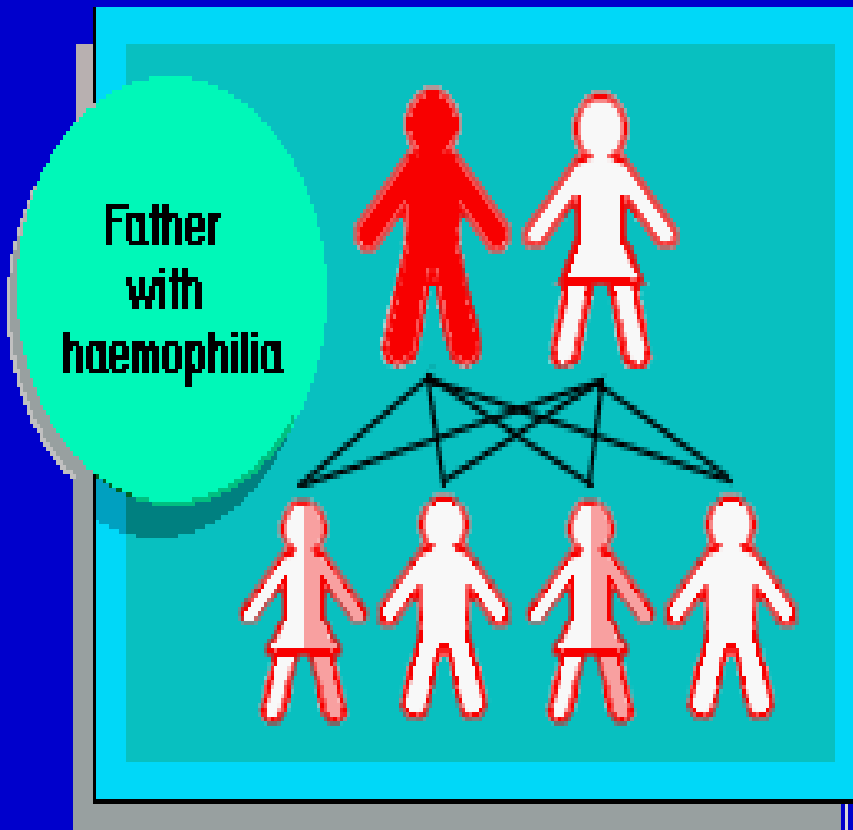
- Chances of carrier mother passing defective gene to a child are 50:50.
- Each daughter has 50:50 chance being a carrier
- Each son has 50:50 chance of having haemophilia.
- Possible for defective gene to remain hidden through several generations of family.

# Possible Carriers



- All daughters of a carrier
- Sisters, mothers, maternal grandmothers, aunts, nieces and female cousins of carriers. of an affected male
- Mothers of one son with haemophilia who have no other members who either have or are carriers of haemophilia.

# CARRIERS



- In about 1/3 carriers bleeding problems can occur with tooth extractions, surgery, severe accident or following pregnancy.
- Carriers should have FVIII/FIX levels measured.
- NOTE: If your factor levels are normal this does not outrule carriership.





# Haemophilia

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- Normal VIII > 60. IX > 80
- Low Level - > 40%
- Mild Haemophilia 5-40%
- Moderate Haemophilia 1-5%
- Severe Haemophilia > 1%



# Bleeding in haemophilia carriers

- 30 % have lower than normal levels
- Treatment necessary for:
  - Dental work
  - Surgical procedures
  - After trauma
  - Pregnancy (FIX)
- Tendency to bruise
- Epistaxis

**If you don't know your level you cant prevent bleeding**



# Management of pregnant carriers

- Before getting pregnant, carriers need clear and accurate information about:
- the chance of transmitting hemophilia to the child. Carriers of hemophilia have a 50 per cent chance of passing the disorder on to their children;
- the consequences, to both a female and a male child, of inheriting hemophilia;
- how hemophilia is treated, what care is available at a local level, and at what cost;
- how pregnancy, labour, and delivery should be managed to reduce risks to both mother and child;
- the options available for conception, prenatal diagnosis, and fetal sex determination.



# Pregnancy

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- **Monitor and correct at delivery mums factor levels**
- **Sex of baby**
- **Communication and planning**

**Incidence of intra or extracranial haemorrhage in newborn with haemophilia is 4%**



# Genetic Counselling

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*Counselling provides support throughout the process of undergoing investigations, receiving results and making decisions appropriate to the individual's situation for their future.*

Lee et.al. (2006).

*'Genetic disorders give rise to complex issues about parenting (guilt, anger, fear and resentment)'*

Miller, (1999:77).

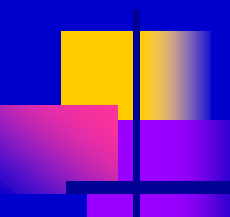


# Issues

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- To test or not to test?
- What age to test?
- Who do I tell?
- What implications for me?

# Issues. Decisions



The decision to undergo genetic testing is shaped by the family's perceptions and cultural concerns. Some families delay testing as a form of denial, or to protect the child and themselves from what they perceive as bad news.

Cultural issues, such as arranged marriages or the possibility of the daughter having an affected child of her own, may discourage some families from having a daughter tested.

Others test routinely as a matter of course, letting the child grow up with the knowledge of their carrier status. Knowing their status early can also help girls come to terms gradually with the complex reality of being a carrier.



# Issues

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- Poor communication regarding genetic inheritance.  
feelings of stigmatisation, guilt, discrimination, denial.

*'...over half of the women interviewed first became aware that they were likely carriers of the haemophilia gene after the birth of a son with haemophilia'.*

Thomas, (2007)





# Issues

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- Guilt

“If I didn’t know then I can’t feel guilty about the result”  
Having passed on the condition or risk of the condition

- Joy

‘Better and closer’ family by facing the common threat together. Value life more.

Todd, 2007



# Quality of Life

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- Being a carrier of hemophilia can have a significant impact on a woman's health and her academic, professional, and social life.
- Excessive or prolonged menstrual bleeding can be especially difficult for young girls, who may isolate themselves from family and friends, miss days from school, or avoid social events due to pain, discomfort, or the fear of staining clothing.
- A girl's self-image and confidence can be negatively affected if she experiences shame or embarrassment because of heavy menstrual bleeding.



# Quality of life issues

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- Many carriers are not aware that their symptoms are abnormal and do not seek medical advice. Even when they do, caregivers are not always well informed about bleeding disorders and the right diagnosis may be overlooked. Furthermore, medical care for women is lacking in many countries around the world. There may be cultural taboos and obstacles preventing women from seeking help, particularly for menstrual problems.
- Heavy and prolonged menstrual bleeding and pain can affect a woman's sexuality and may cause problems in her marriage. Women may also need to take time off work each month because of heavy bleeding, which can impact their career choices or professional success.



# Quality of Life

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- Many carriers of hemophilia, like others at risk of passing on a genetic disease, also experience guilt. They may feel as though they should not have children because of the possibility of passing on a bleeding disorder, or having a daughter who must face this possibility in turn.
- The prospect of marriage may be affected because men, or their families, may not accept the risk of having an affected child. If they do have children with hemophilia, the needs of that child can put pressure on all family members, including siblings.



# Support.

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- Hemophilia treatment centres provide carriers with skilled and sensitive counselling providing information and support to work through these complex feelings.
- Building a support network of other women who are facing the same issues, through the hemophilia treatment centre or Haemophilia Societies , can be a great source of empowering women to take charge of their condition and advocate for proper treatment.



# Client's needs

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- Each individual determines what is right for themselves.
- Be sure to have your questions answered in a way you understand.



# What do I do?

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- Phone NCHCD for carrier clinic appointment. (E O Shea)
- A member of your family with haemophilia or carrying Haemophilia must initially give blood samples.
- Following consultation, consent to proceed with DNA analysis.
- Blood samples X 5 .
- Results available routinely 1 month. (Emergency 2 days.)
- Attend in person for results. (never given over phone)
- A confirmatory sample is requested when initial results are available.

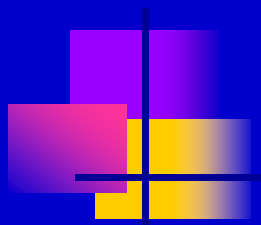


# Conclusion

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- Important to know your carrier status
- Important to know your factor level
- Inform centre when any surgery planned or procedures which may require treatment.
- Make appointment when planning a pregnancy.
- Inform centre when pregnant
- Be informed and educated
- Don't take risks





I

II

III

IV

V

VI

VII

VIII

Albert

Victoria

Elizabeth II

Charles

Alexei

■ = hemophilia

# = age at death

● = carrier for hemophilia

