Genetic counselling for Haemophilia

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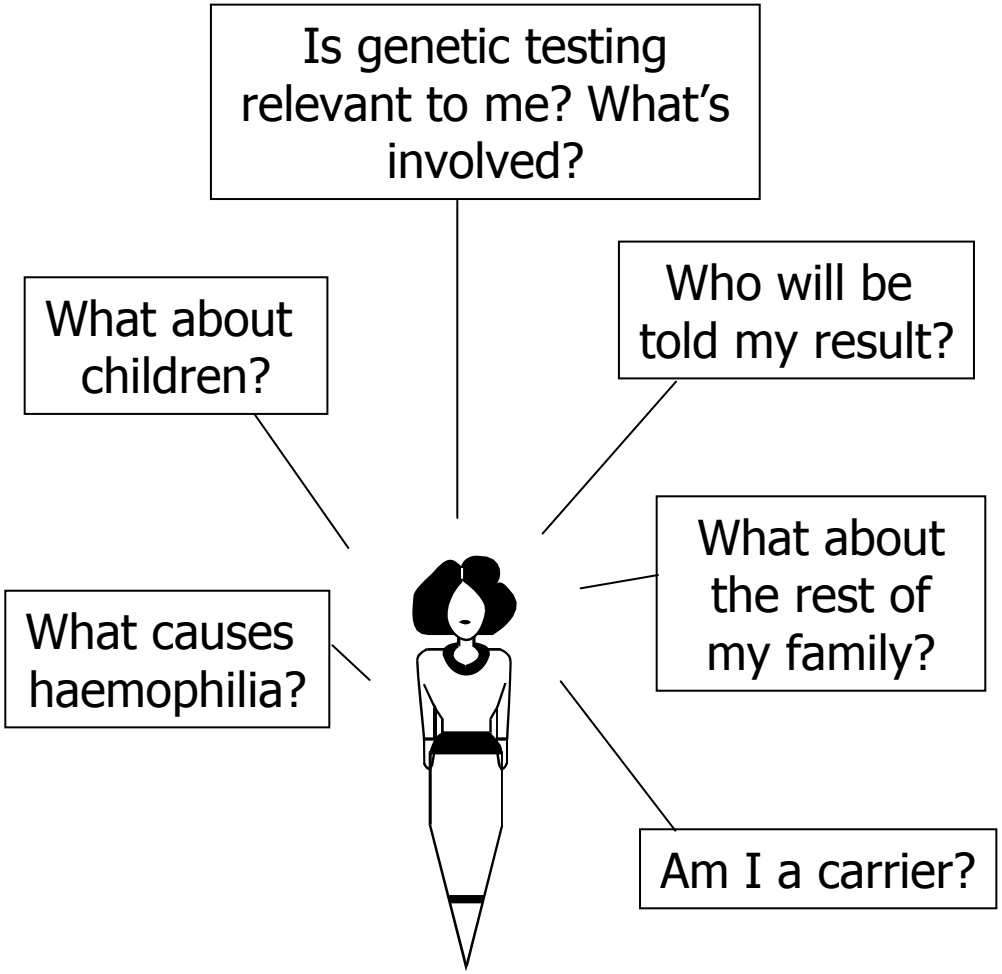
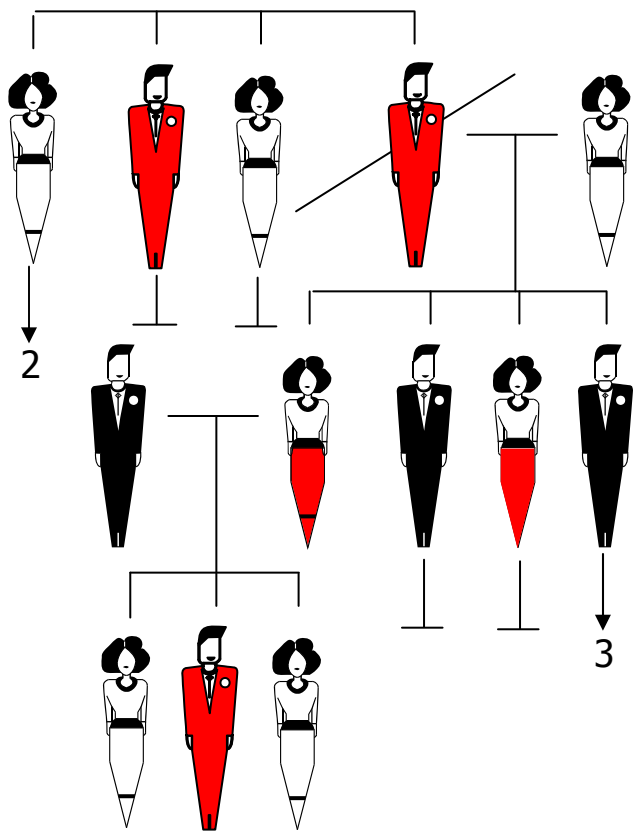
What is Genetic Counselling?

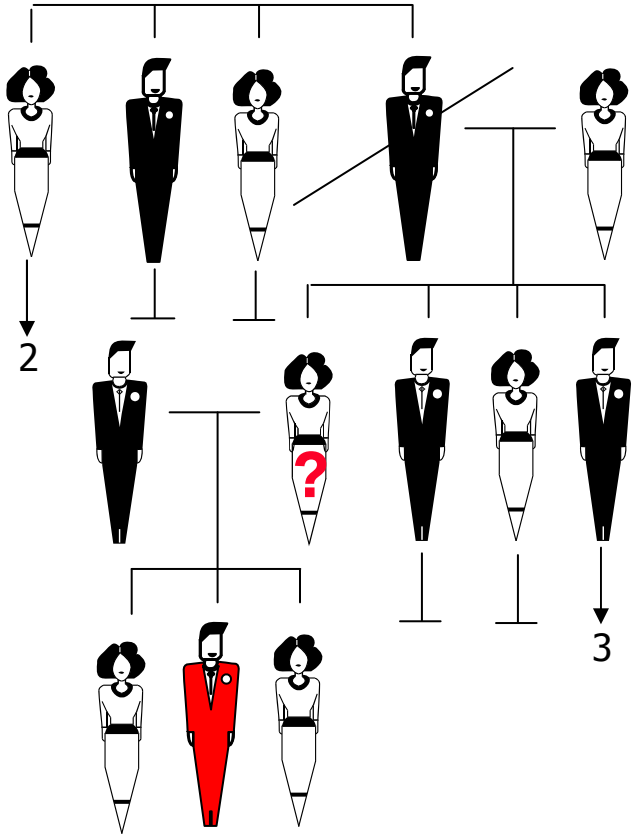
- Information
- Counselling
- Support
- Informed decision-making

Family history of Haemophilia



The family tree





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

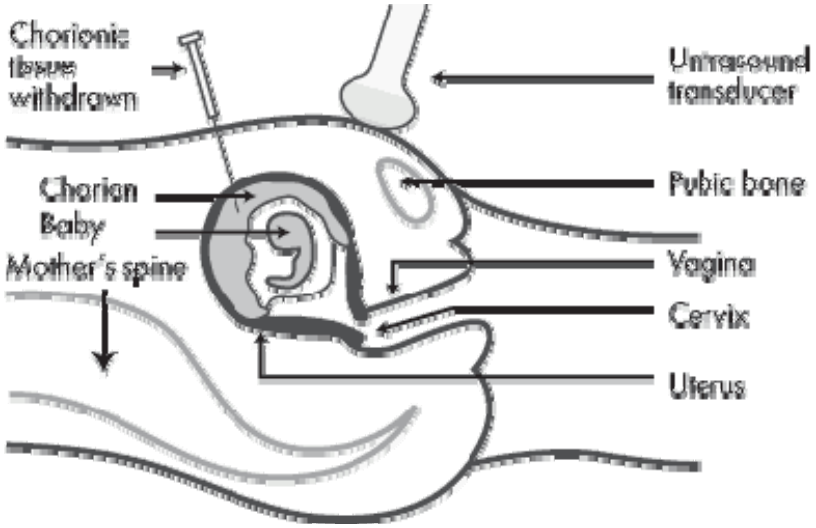
Decision to have carrier testing

- 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

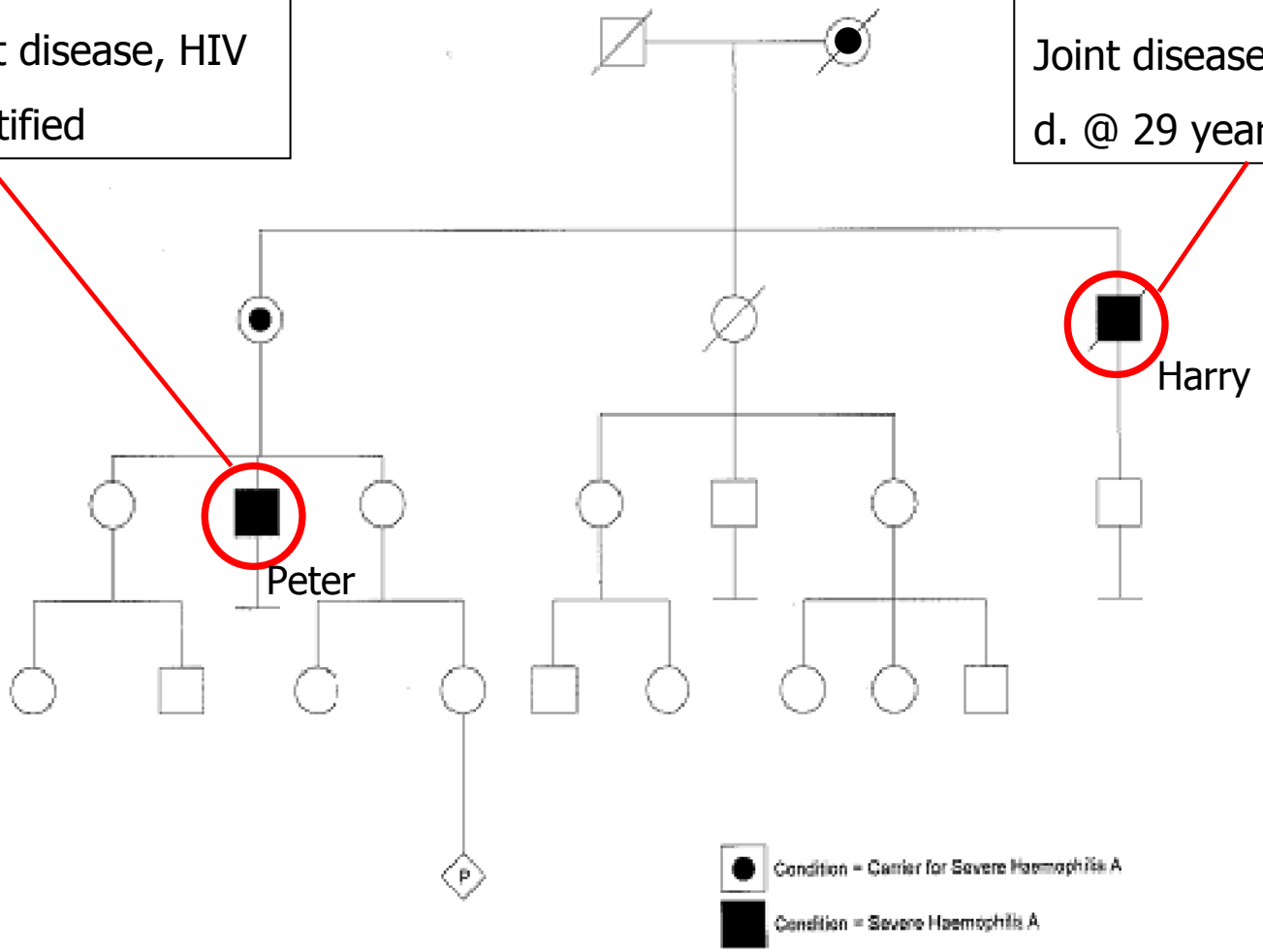
Mutation identified

Harry

Severe Haemophilia A

Joint disease

d. @ 29 years



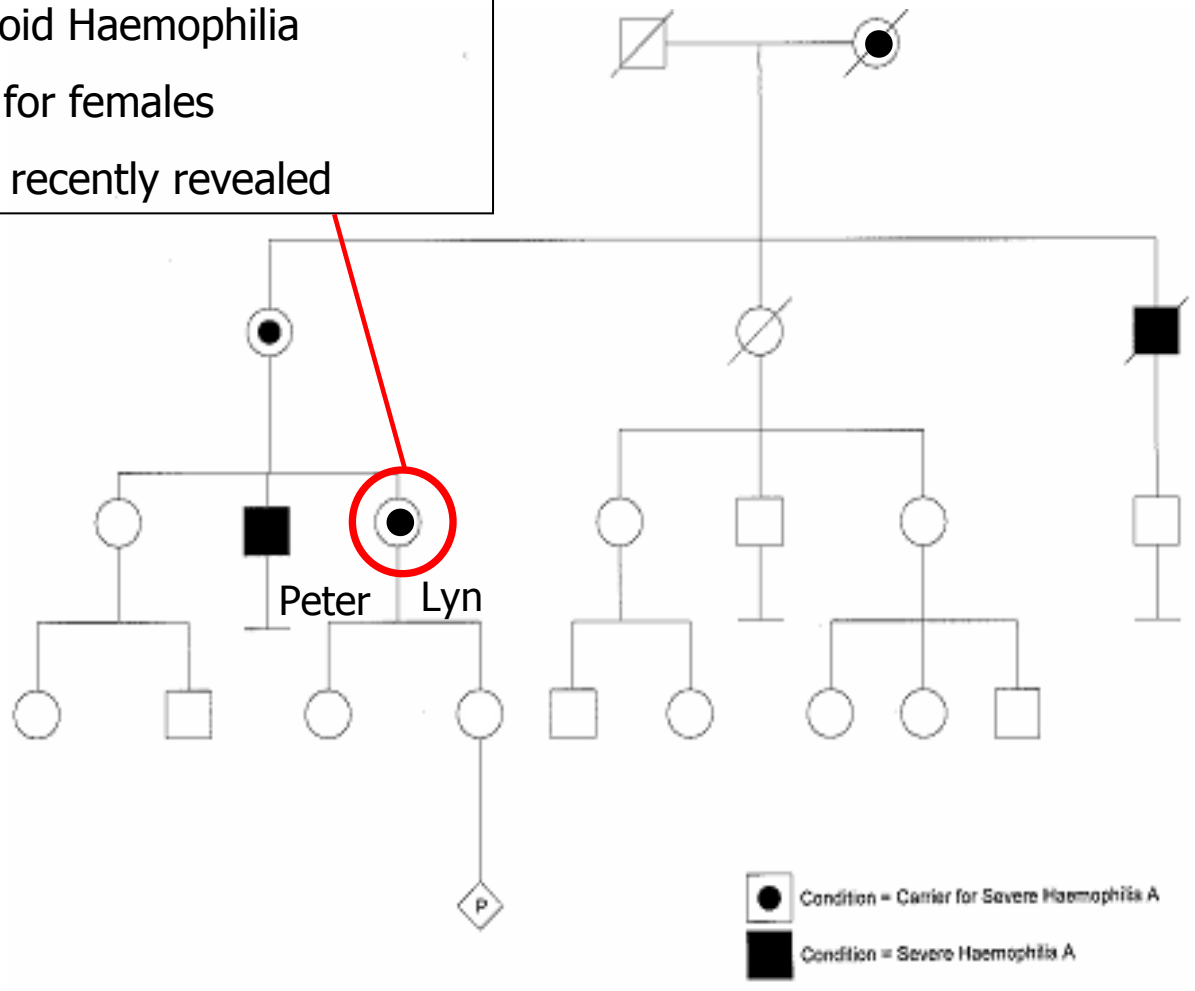
Lyn

Genetic counselling prior to family planning (many years ago)

Wanted to avoid Haemophilia

Sex selection for females

Carrier status recently revealed



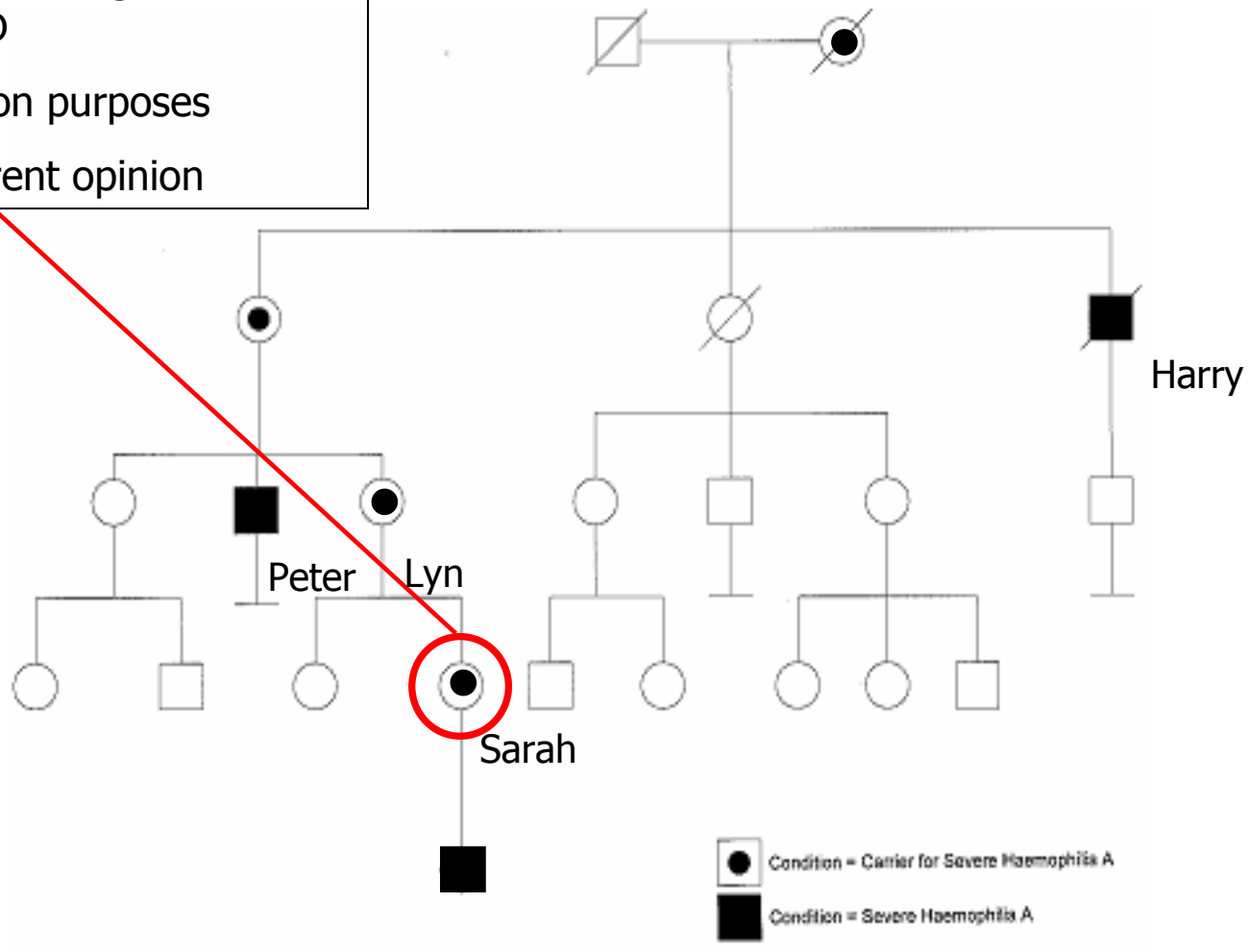
Sarah

Attended with mother, Lyn

Pregnant, requesting carrier testing & PND

For preparation purposes

Lyn has different opinion

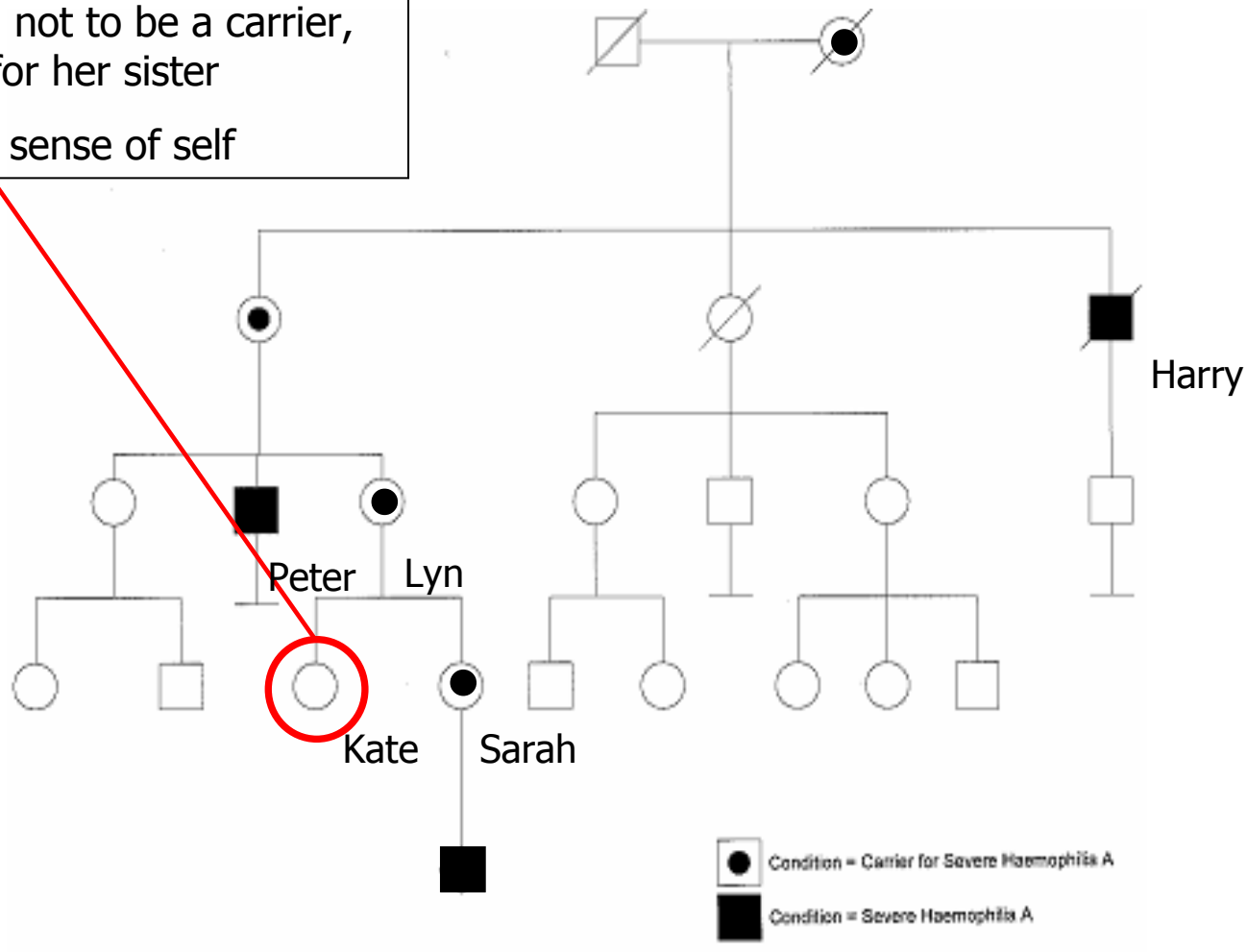


Kate

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

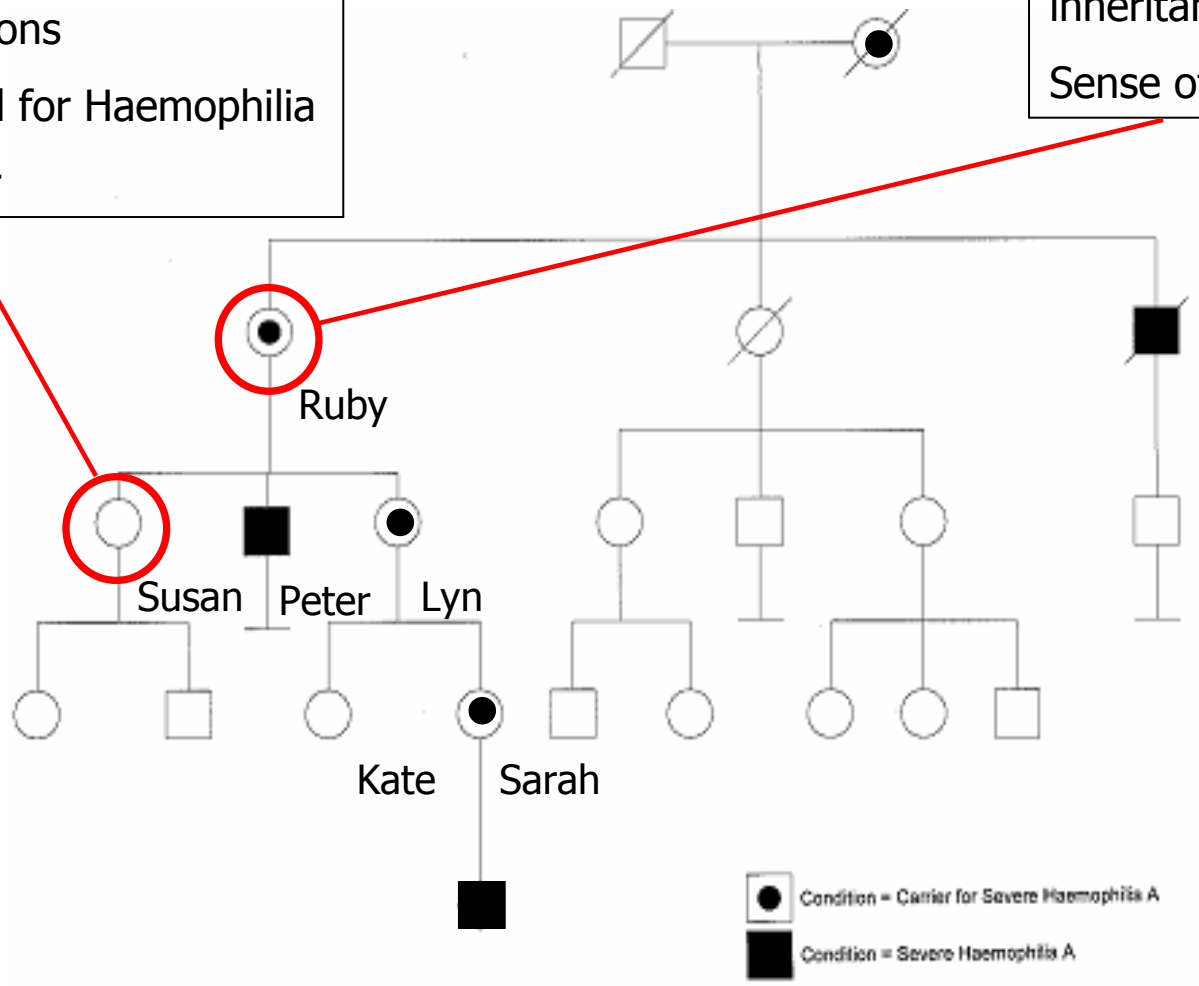
Never tested for Haemophilia

Not a carrier

Ruby

Never understood inheritance

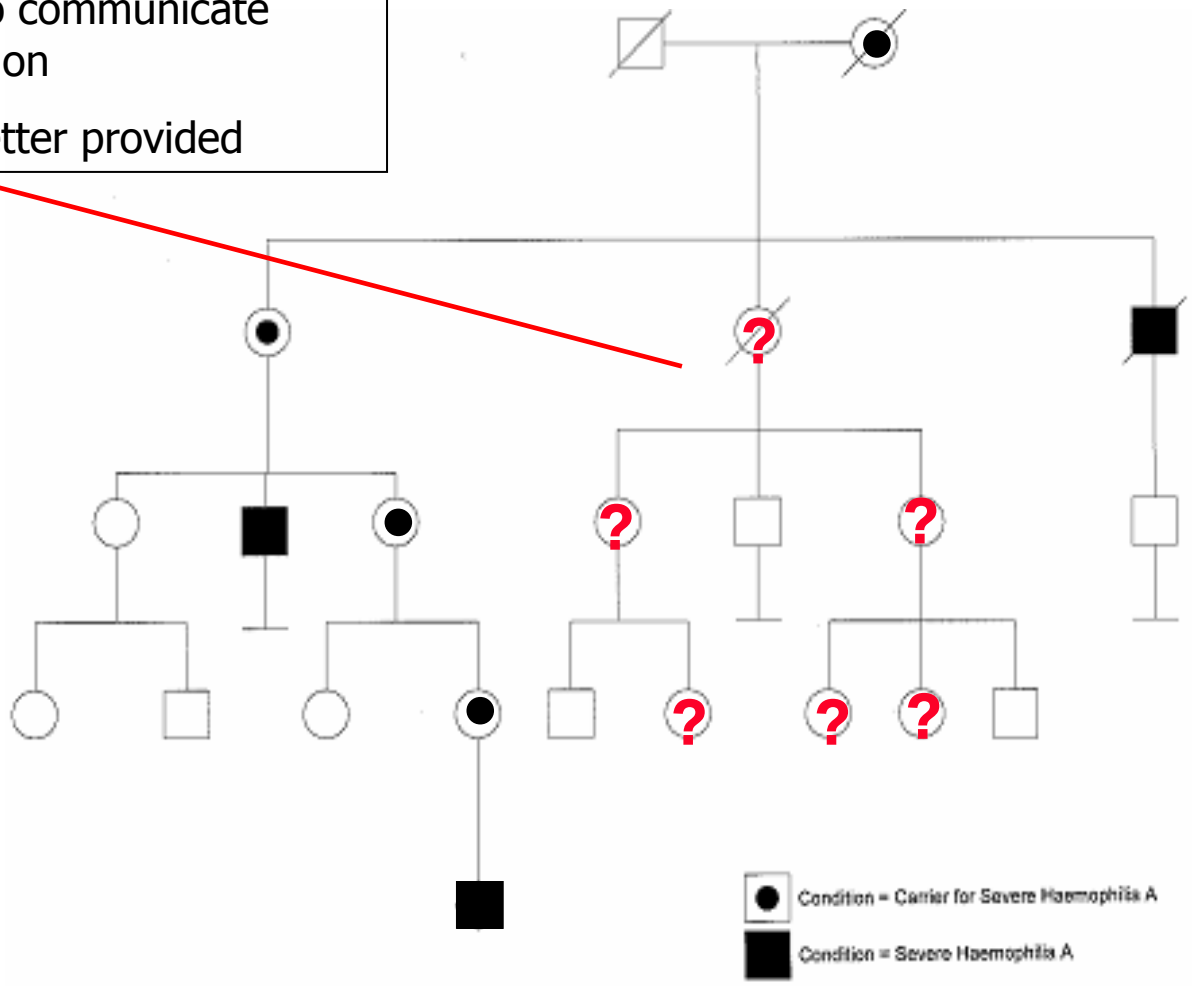
Sense of guilt



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