Family planning and the role of the Genetic Counsellor

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Monash Genetics

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What is Genetic Counselling?

- Genetic diagnosis for affected individuals
- Information and education
  - Risk assessment
  - Genetic testing
- Counselling and support
- Informed decision making
- Management
- Provided by a team of healthcare professionals
  - Geneticists, Genetic Fellows, Genetic Counsellors
  - Social workers, Obstetricians, Haematologists, Paediatricians, Neurologists, Oncologists etc

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Genetics of Haemophilia

- Genetic (inherited) condition
- Major forms – Haemophilia A and Haemophilia B
- Haem A – factor VIII: approx. 1 in 4,000 – 1 in 5,000 males
- Haem B – factor IX: approx. 1 in 20,000 males
- More common in males than females
- Approx. 30% of female carriers mildly affected

- Genes affected *F8* and *F9*
- Both genes - instructions to make protein for coagulation factors VIII and IX
- Both located on X chromosome
- X-linked inheritance pattern

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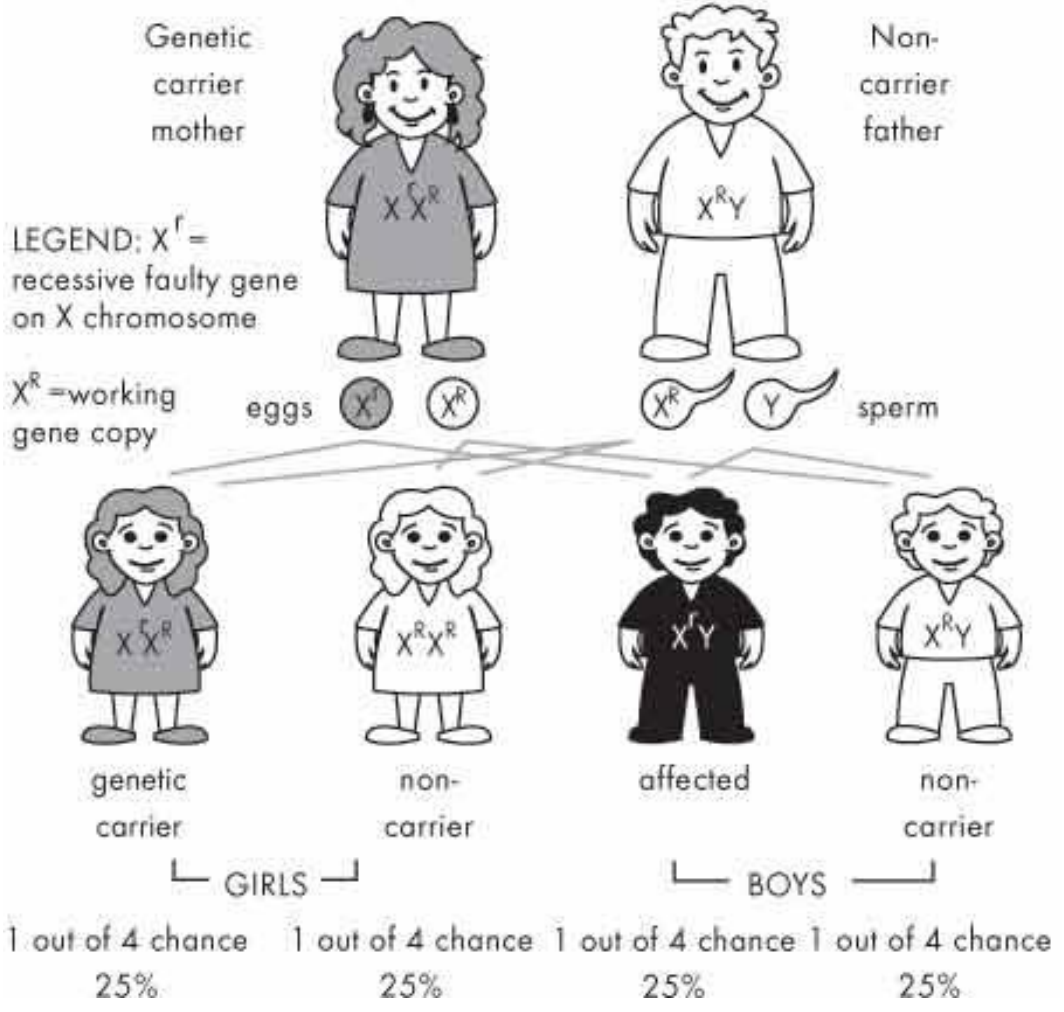
X-linked inheritance



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Other bleeding disorders

- Different inheritance patterns:
- Eg. Von Willebrand disease – most common bleeding disorder (variable - can be very mild)
  - *VWF* gene mutation inhibits clots forming
  - most cases: dominant inheritance pattern: 50% risk to children
  - few cases autosomal recessive: 25% risk to children
- Eg. Factor VII deficiency (inherited form)
  - Autosomal recessive inheritance: 25% risk to children

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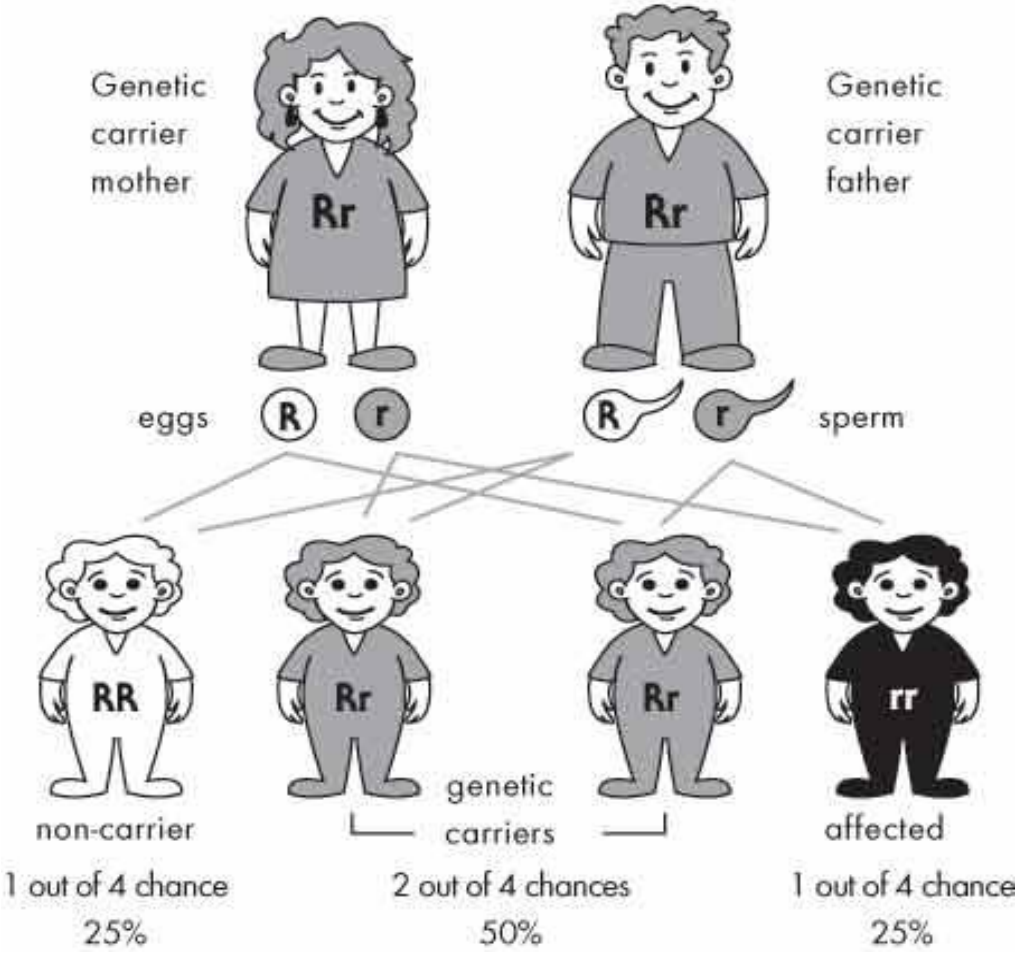
Autosomal recessive inheritance



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The genetic consultation

Genetic counselling for family planning

- Intake (information gathering prior to or at commencement of appt)
  - Reason for referral?
    - family history of haemophilia or other bleeding disorder, planning pregnancy
    - pregnant with family history – known carrier / carrier risk
  - Family history / proband?
  - Existing genetic file? (individual or family member)
  - Family mutation identified?
- Commencement of appt - patient agenda
  - Questions/issues/expectations
  - Prior knowledge
  - Any misconceptions
- Geneticist/Counsellor agenda
  - Any differences?

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Mutation detection

- Family history – no mutation
- Genetic testing (proband)
- Some common mutations for haemophilia
- Many families have a “private mutation”
- Tiered testing process dependant on clinical diagnosis and severity
- Clinical severity tends to run true in families, but other genetic/environmental factors may moderate
- Consent includes permission to use for other family members

Mutation identified in ~98% of cases

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Carrier testing

- Females with family history
- In the past…
  - Factor VIII/IX levels
  - but some carriers have normal Factor VIII/IX levels (>50% of carriers)
    - ➡ not always helpful
- Today…
  - Genetic testing
  - Accurate carrier testing

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Reproductive options

For female carriers:

- Accept the 50/50 risk
  - have children
  - not have children (or adopt)
- Use technologies
  - chorionic villus sampling (CVS) or amniocentesis
  - IVF and pre-implantation genetic diagnosis (PGD)
  - donor egg

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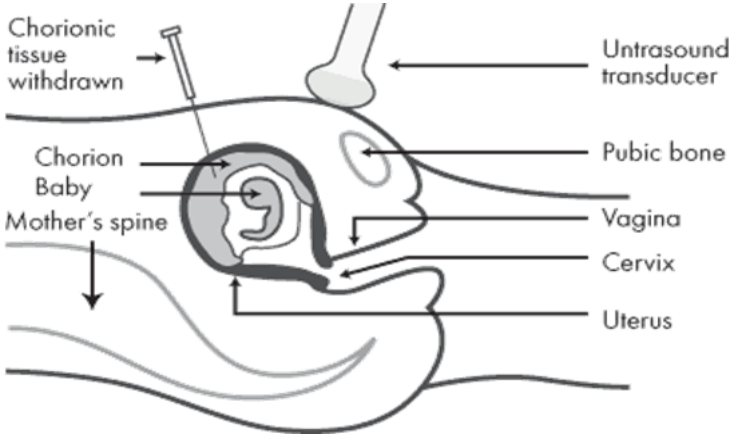
Chorionic Villus sampling

Mutation must be known

- Prenatal diagnosis
  - Routine, accurate
  - CVS (~ 12 weeks, <1% risk miscarriage)
  - Amniocentesis option

    (~ 16 weeks, <0.5% risk miscarriage)
  - FISH for fetal sex
  - Chromosome analysis
  - TOP an option (if male? if female?)
  - Difficult decision making



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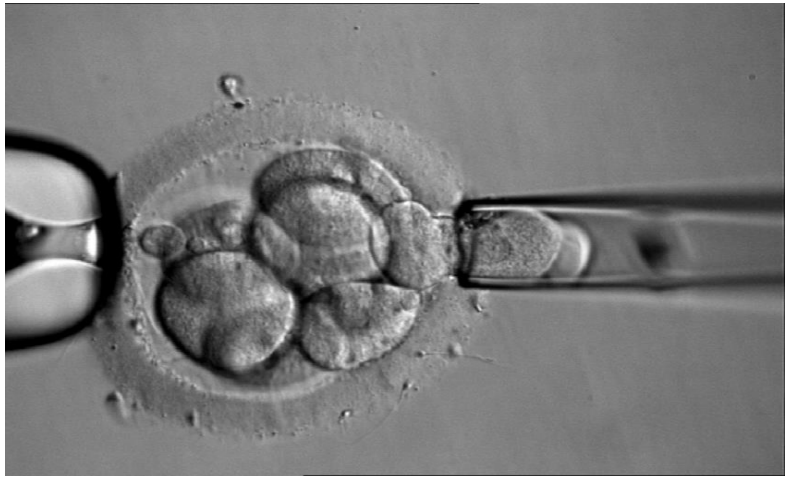
IVF & Pre-implantation Genetic Diagnosis

Mutation must be known

- Pre-implantation genetic diagnosis
  - utilises IVF technology
  - Highly accurate but not 100%
  - Confirmatory prenatal testing recommended
  - Can avoid TOP
  - Financial, emotional, physical drawbacks
  - No guarantees



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Informed decision making for pregnancy testing

Pros and cons of reproductive options

- Perception of risk
- Personal/family experiences of the condition
- Emotional state
- Perceived burden of condition
- Natural history & quality of life; treatment; physical, emotional & financial burden
- Conflict between beliefs/values
- Consideration of other family members

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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

- Advice for Obstetricians regarding:
  - Delivery
  - Pregnancy management
  - Possible risks, including maternal risks
  - Diagnostic testing of male infant

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CASE EXAMPLE

Peter

Severe Haemophilia A

Currently 45 years

Inhibitors, joint disease, HIV

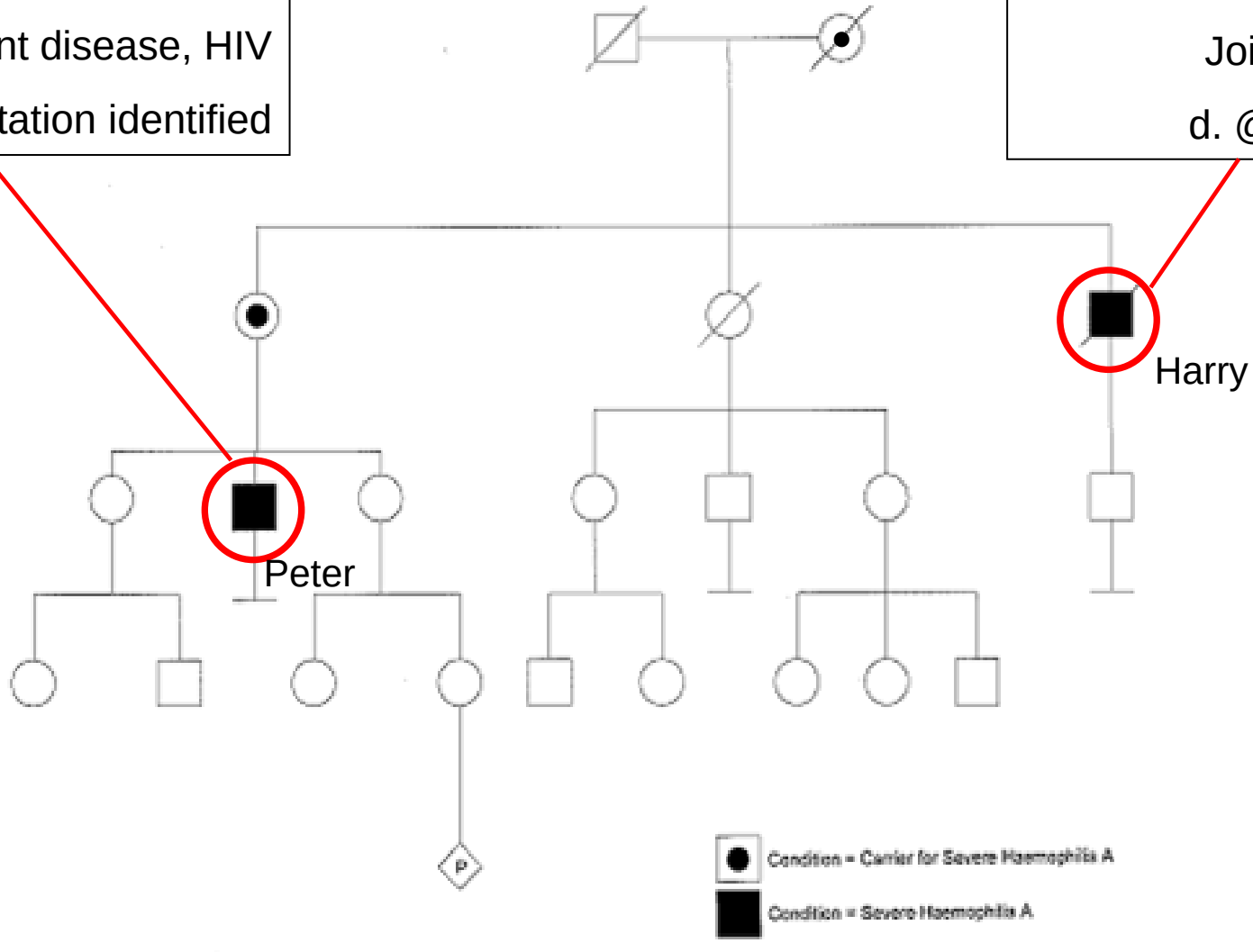
Mutation identified

Harry

Severe Haemophilia A

Joint disease

d. @ 29 years



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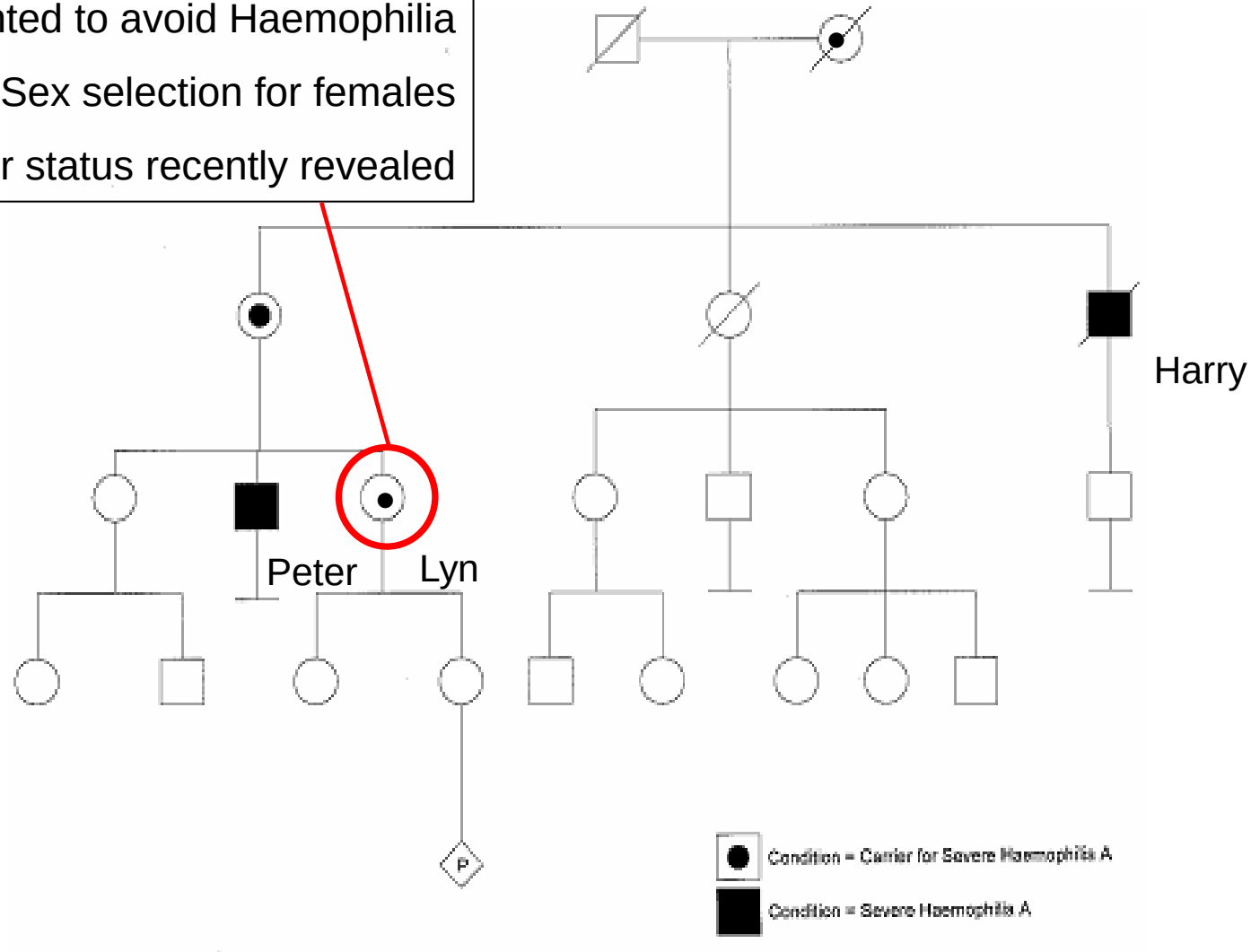
Lyn

Genetic counselling prior to family planning (many years ago)

Wanted to avoid Haemophilia

Sex selection for females

Carrier status recently revealed



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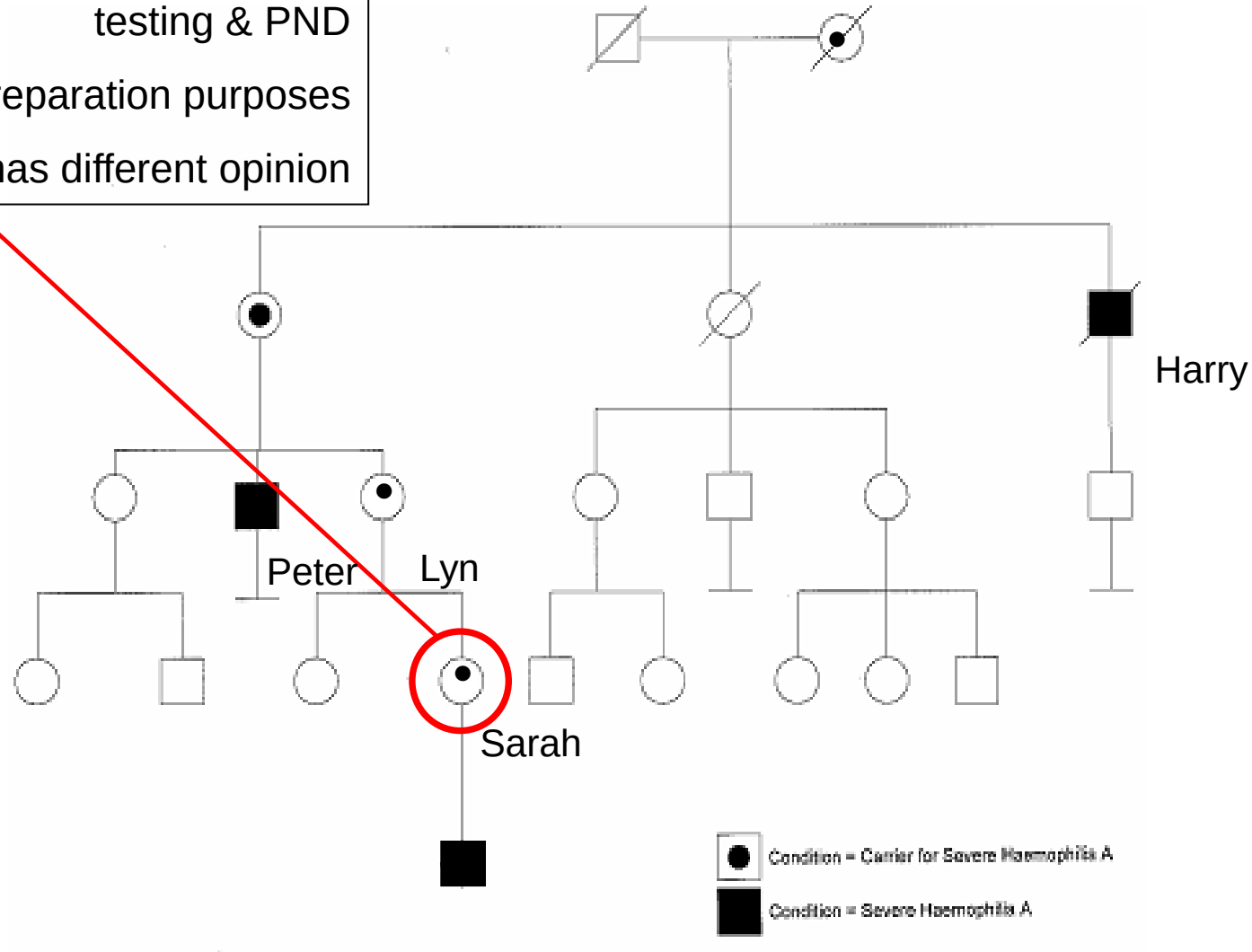
Sarah

Attended with mother, Lyn

Pregnant, requesting carrier testing & PND

For preparation purposes

Lyn has different opinion



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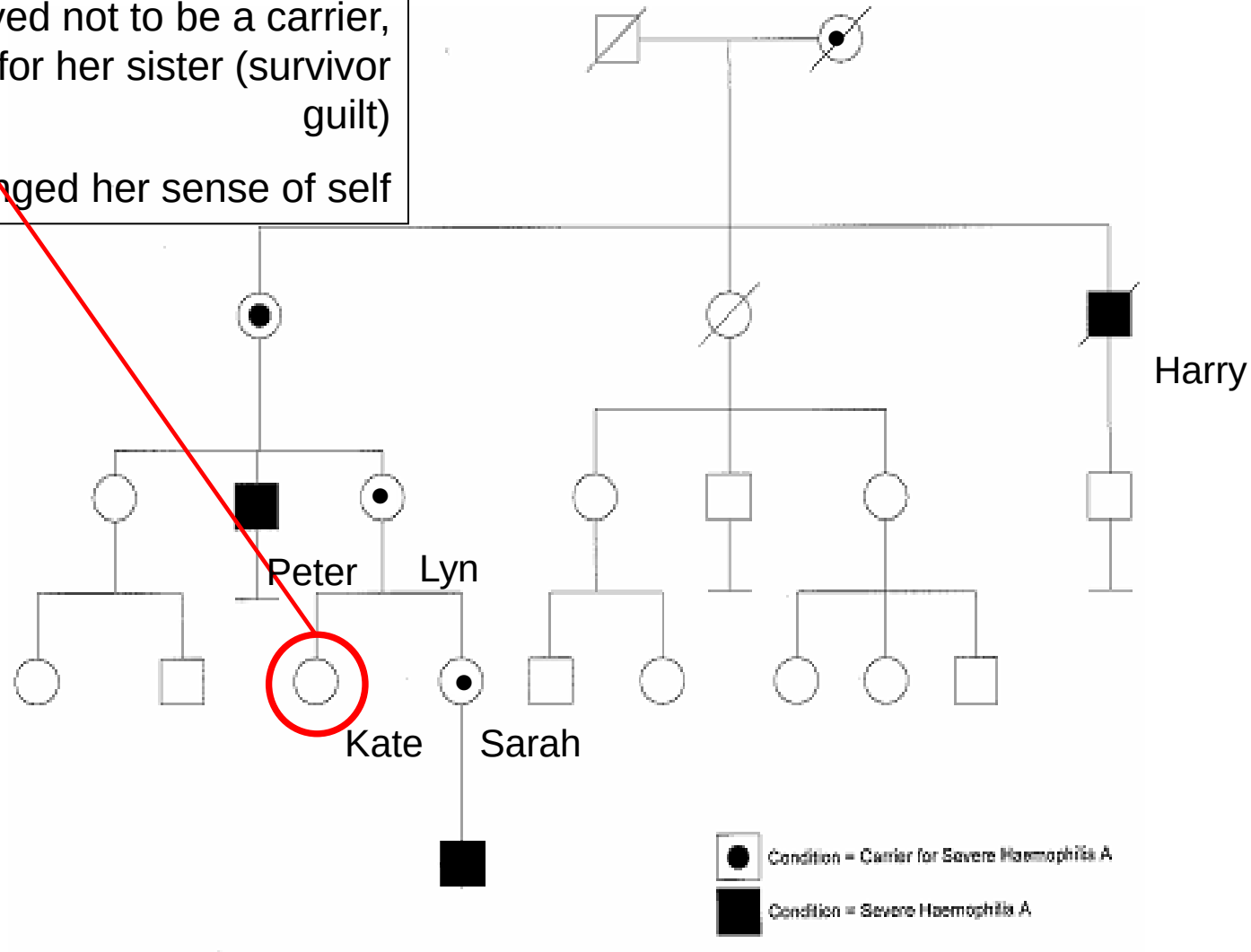
Kate

Always believed she was a carrier

Very relieved not to be a carrier, but felt bad for her sister (survivor guilt)

Changed her sense of self



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Susan

Increased responsibility

Mixed emotions

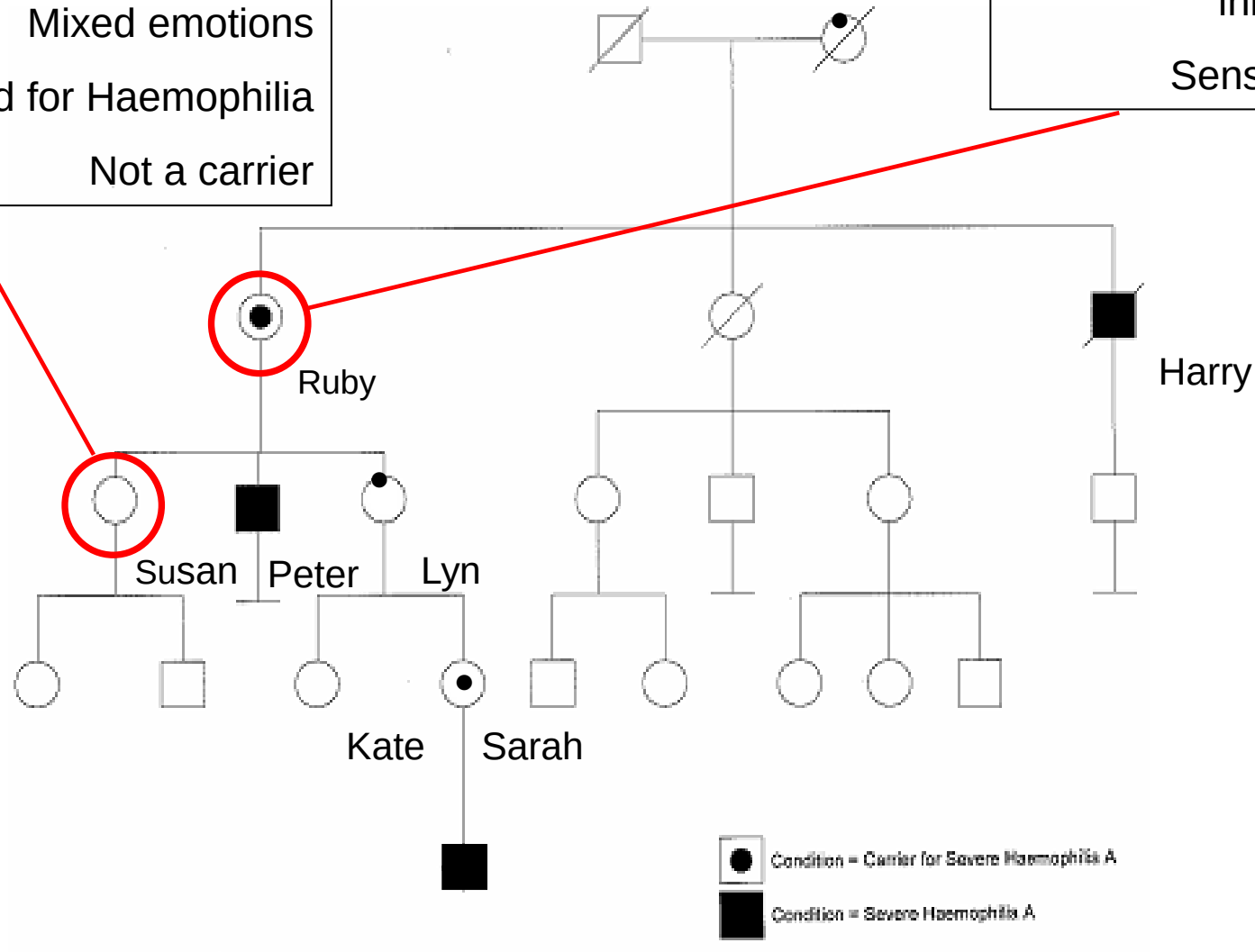
Never tested for Haemophilia

Not a carrier

Ruby

Never understood inheritance

Sense of guilt



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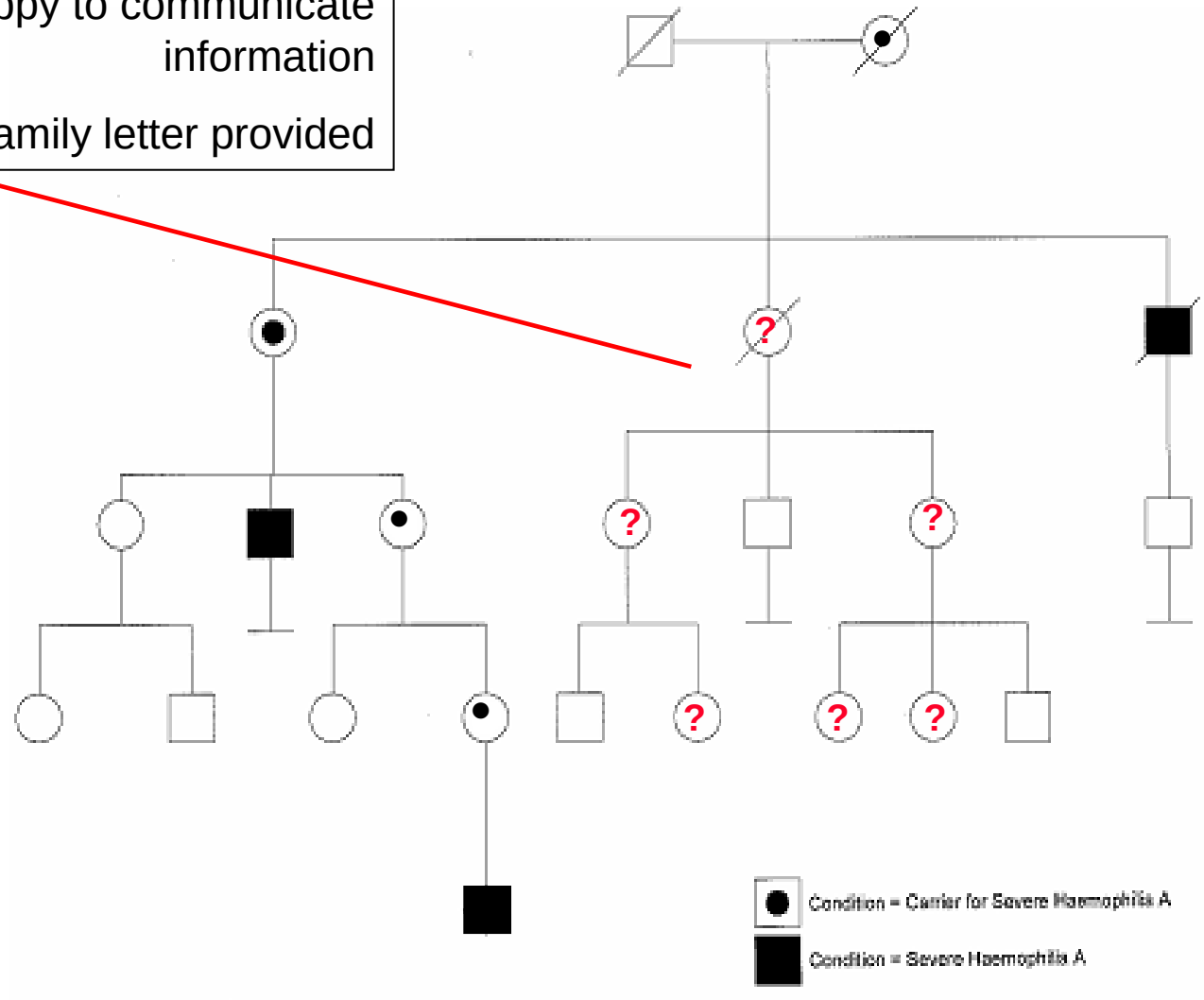
Implications for other family members

Happy to communicate information

Family letter provided



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Counselling issues

- Individual/family experience
- Issues for pregnant women
- Differing opinions of family members
- Beliefs about carrier status
- Multiple emotions
- Devaluing a life
- Family communication of information

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Monash Health Genetic Services

General Genetics

GP/specialist referral

Monash Genetics,
Special Medicine Centre,
Monash Medical Centre,
246 Clayton Road,
Clayton 3168
ph: 9594 2026, fx: 9594 6022

Clinical geneticists

- Dr Matt Hunter (Head of Unit)
- Dr Emma Krzesinski
- Dr Alison Yeung
- Clayton, Berwick Clinics

Ward consults available

Genetic Counsellors (appts Mon-Fri)

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