

XX Why?

To be read in conjunction with reference sheets A, B and D

Jessica is currently 8 weeks pregnant to her partner, Luke. They come to see you - a genetic counsellor - at a genetics clinic because Jessica is concerned about her family history. Jessica's mother Judith had identical twin girls, Monique and Jessica, in 1975. After her first husband died, Judith married Stephen and together they had 3 children: Tamara, Adam and Andrew. Andrew has mild learning problems and a chromosomal analysis when he was 10 revealed that he has Klinefelter syndrome.

Klinefelter syndrome affects 1 in 600 males and occurs at random. It results when two X chromosomes and one Y chromosome are inherited, instead of the usual one X and one Y. Males who have Klinefelter syndrome therefore have a total of 47 chromosomes, not 46. The extra X chromosome may be inherited from either the mother or the father.

(Klinefelter syndrome refers specifically to males with an extra X. Females who inherit an extra X chromosome are said to have Triple X).

At birth a baby boy with Klinefelter will not look or behave any differently to other babies. In fact males with Klinefelter syndrome are often only diagnosed in adolescence or adulthood. The features of Klinefelter syndrome include being

taller than average with long limbs, intelligence within the average range although there may be mild intellectual disability or learning difficulties, testosterone levels are lower, testes smaller and the development of male secondary sexual characteristics may be poor. Males with Klinefelter syndrome are infertile.

Klinefelter syndrome is named after Dr Harry Klinefelter who first described the condition in 1942.

1. Draw the pedigree for this family.
2. A normal male karyotype is 46XY. What is Andrew's possible karyotype and why does Andrew look male even though he has two X chromosomes?
3. Do you think Jessica and Luke are at high risk of having a child with Klinefelter syndrome? Give a reason for your answer (Hint: is it hereditary?)
4. Jessica mentions that she is concerned about her brother, Andrew, not being able to have children of his own due to infertility. Can you suggest what other reproductive options might be available for Andrew?

Extension Activity

Judith, Jessica's mother, is a known genetic carrier for the autosomal recessive disorder, haemochromatosis (see worksheet 2).

If Jessica were to have testing how would her result compare to that of Monique? Explain your answer with reference to monozygous and dizygous twinning.

