

Prenatal scenarios

Scenario 1

Jill is pleased to be pregnant for the first time. She is meeting with her community midwife Paula as she has concerns about her family history. Several family members have children with congenital anomalies and learning difficulties. Following the drawing of the family tree (Fig. 1) it can be seen why she is concerned. There is also a family history of recurrent miscarriages.

Three family members are affected. On closer inspection, only two out of the three affected individuals are blood relatives. She knows that her niece has some sort of “chromosomal problem.” Her brother has had a chromosome test in the past but is unsure what it means.

After talking with Jill, Paula decides to refer her to the local genetics clinic. They would be able to gain consent to access the relevant medical notes and confirm if the family history had implications for the pregnancy.

(In some families, the presence of a chromosome rearrangement called a balanced translocation can result in a family history of miscarriages. Some of these families will also have a family history of children born with congenital anomalies and learning difficulties. Carriers of familial translocations are generally healthy. However, they, or their wives will be at increased risk of having miscarriages or children with congenital anomalies and developmental delay).

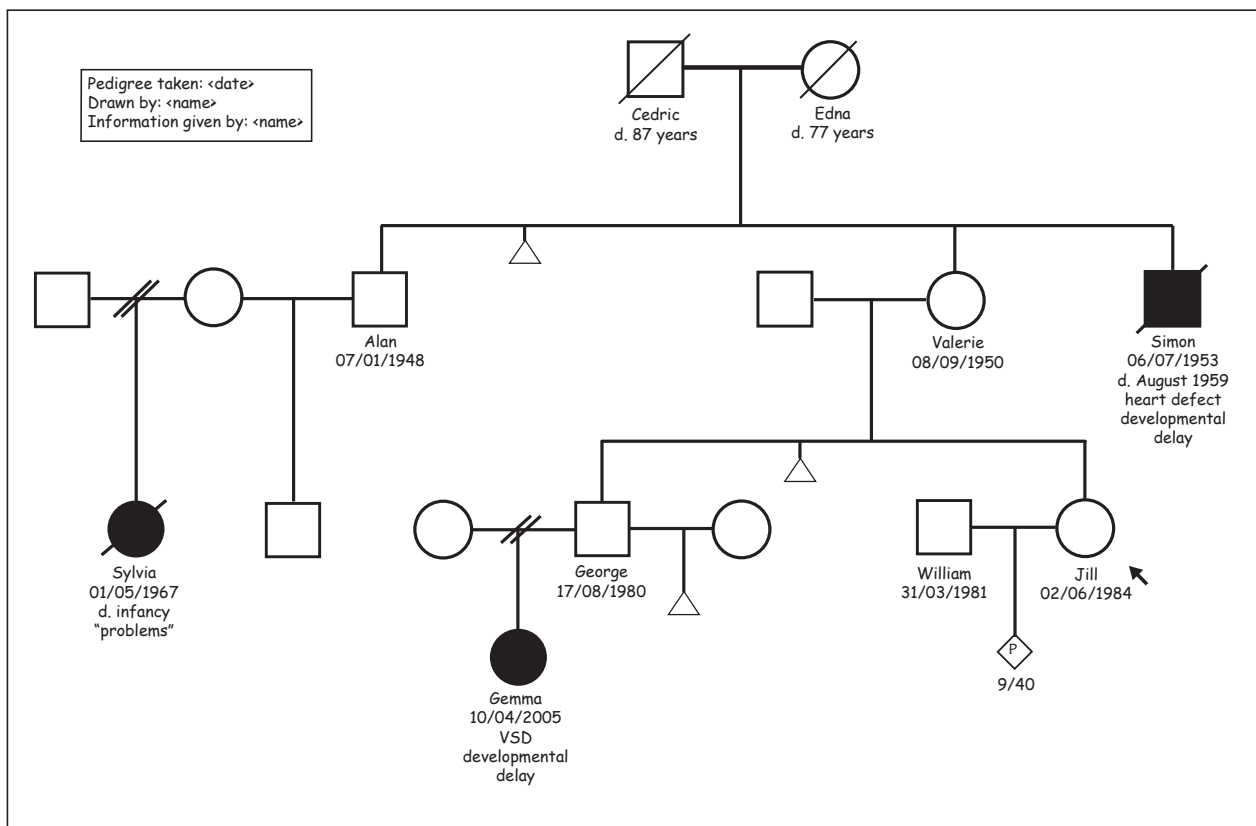


Fig. 1 Scenario 1 - Jill's completed family tree

Scenario 2

Helen and David are meeting with their midwife Carol. David's niece Lauren has been recently diagnosed with cystic fibrosis (CF). The couple have questions about his niece and whether or not they should be worried for their own baby. They appear to think that the family history "is just one of those 1 in a million things" and really wouldn't have implications for the pregnancy.

Carol draws out the family tree (Fig. 2) and discusses with them the pattern of inheritance that is associated with CF. It is passed on through the family in what is called an autosomal recessive way. This means that the condition can affect both boys and girls. Also, the parents of an individual with CF are generally fit and well and do not know that they are CF carriers. She explains that many parents who have a child with CF do not have a family history of the condition.

Carol tells the couple that as there is a family history of CF, that there could be implications for the pregnancy and if they wanted to discuss this further then she could refer them to the genetics clinic. The couple are very shocked. Carol refers them to a genetic counsellor colleague who will be able to see the couple quickly to discuss how they want to proceed.

(Carol is aware that the CF test does not detect all CF mutations. She is aware that she would not be in a position to confirm the diagnosis in the family and gain consent to access the relevant medical notes and test results. She is also aware that the couple need some time to discuss the information that she has given them).

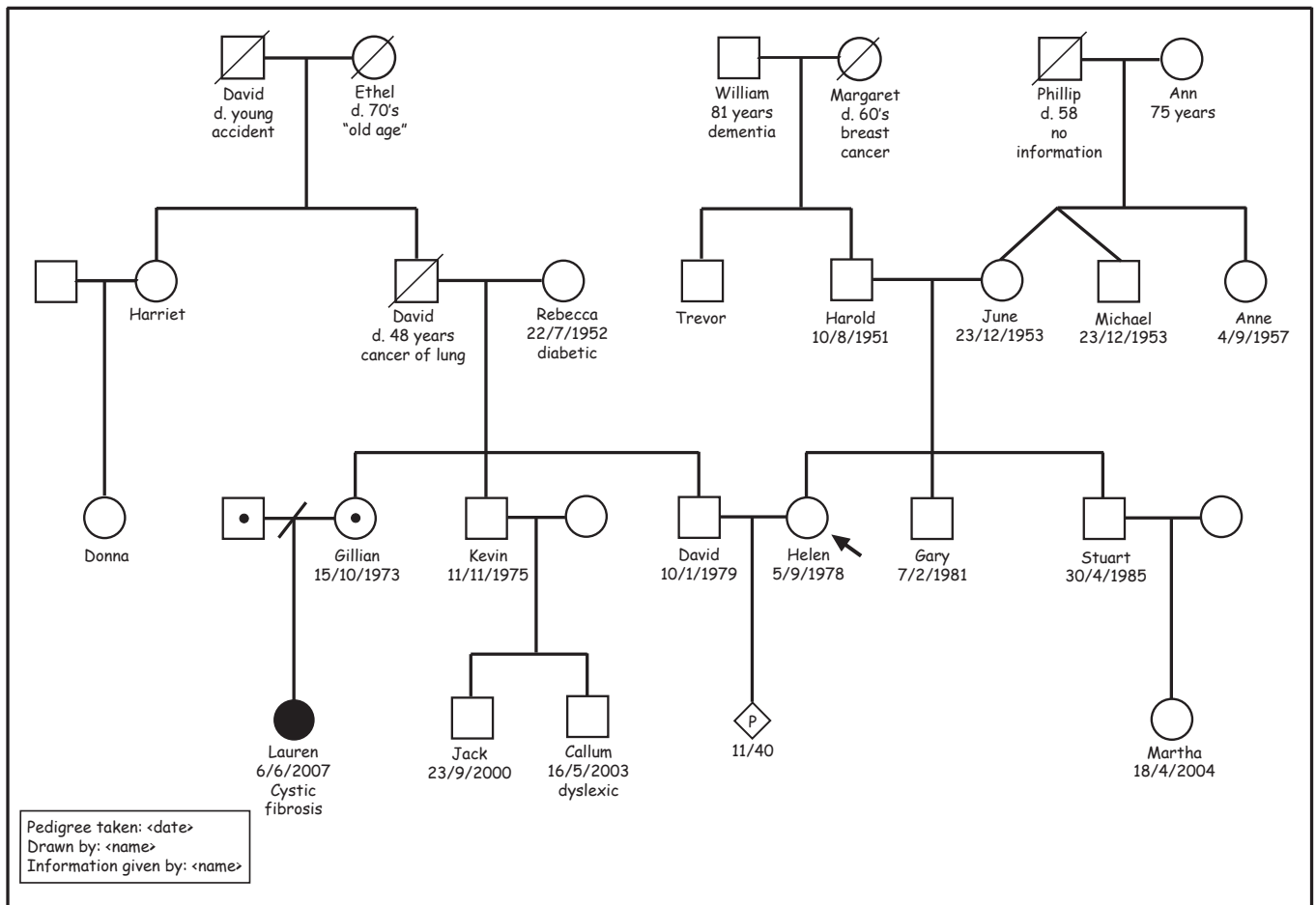


Fig. 2 Scenario 2 - Helen's completed family tree