

A family history of bowel cancer

End of life - Scenario 2

David Collins (date of birth: 13.12.1956) was diagnosed with bowel cancer at the age of 49. He is now terminally ill and is being cared for by his wife Tracey (date of birth: 25.02.1962) at home. On one of your visits David mentions that he always expected to die of cancer because “it’s just what happens in my family”. Both David and Tracey seem to accept this and do not express any concerns about their three children, Kyle, Amy and Josh.

You ask David about his family history and he gives you the following information:

- David Collins was diagnosed with bowel cancer at the age of 49 years.
- He is married to Tracey (DOB 25.02.1962).
- David and Tracey have three children; Kyle (DOB 12.02.1992), Amy (DOB 6.04.1995) and Josh (DOB 07.06.1998).
- David has three sisters; Karen Roberts (born 1949, diagnosed with endometrial cancer at aged 49 years and died aged 50 years), Catherine Breene (born 1950, diagnosed with bowel cancer aged 42 years and died aged 48 years), and Christine Adams (born 1953, alive and well).
- Karen Roberts has two children; Liz (born 1983) and Simon (born 1985).
- Catherine Breene has one son; Nathan (born 1982).
- Christine Adams has two sons; Chris (born 1984) and Jamie (born 1988).
- David’s parents are George and Freda. George (born 1926) died from bowel cancer at the age of 49, Freda (born 1929) is alive and well.

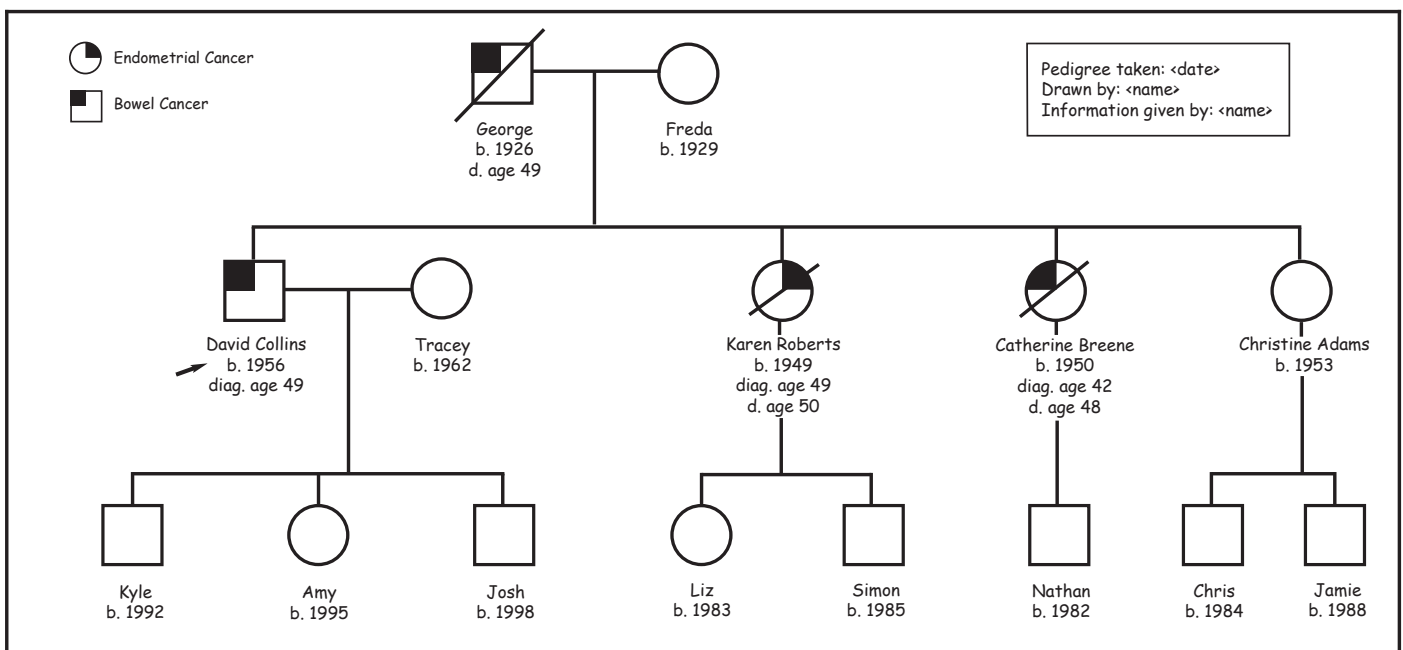


Fig. 1 David Collins’s completed family tree

Practice points

With four people on the same side of the family who have had bowel or endometrial cancer spanning two generations and involving first degree relatives, this family history is suggestive of an inherited predisposition to cancer known as HNPCC (hereditary non polyposis colorectal cancer) or Lynch syndrome. This family is eligible for referral to a specialist genetic service. Family members who are at risk of inheriting this predisposition are Christine and her children and the children of David, Karen and Catherine.

Genetic testing to identify the specific gene alteration that is causing a predisposition to developing cancer should begin with an individual who is affected with cancer. If an alteration is identified then predictive testing may be offered to other family members at risk and surveillance recommendations modified.

Following discussion with David and his family, blood was taken from David so that DNA could be stored for use at a later time should the other family members wish to pursue genetic testing.

The children in the family would not be offered predictive testing until they have reached an age to consent for themselves. This is because HNPCC is an adult onset condition and surveillance is offered from aged 20 years upwards. As there is no direct benefit to the child of having an early predictive test there is no justification for removing the child's autonomy.