

Activity: Construction of a family pedigree

Instructions: Use the information provided below to draw a family pedigree for a specific form of inherited colon cancer, HNPCC. Remember to use all the rules of pedigree construction as you complete this activity.

You are a genetic counsellor based in Melbourne, working with patients to assess their risk for inherited conditions and to explore appropriate genetic testing and management options.

Your patient is **Ben T**, a 46-year-old man in good health. His wife, **Joanna**, is 43 and also in good health. Ben and Joanna have two children: a **20-year-old daughter named Chloe** and an **18-year-old son named Daniel**.

Ben explains that his **older sister, Sharon (now 51)**, was diagnosed with **bowel cancer at age 43**. His **brother, Martin (48)**, was diagnosed with **bowel cancer last year at age 47**. Ben also has a **younger sister, Sophie (39)**, who is currently well.

Ben's parents are both deceased. His **father, Richard**, died in a car accident at age 36, and his **mother, Elaine**, died in a workplace incident at age 41. Elaine had a **brother named Malcolm**, now 71, who was diagnosed with **bowel cancer at age 51**. She also had three sisters and two brothers, all still living and reportedly healthy. Ben's **maternal grandmother** died at age 43 from an unspecified **gynaecological cancer**, while his **maternal grandfather** lived to 86.

Ben's **paternal grandparents** both passed away in their 60s from unknown causes. His father Richard had **three brothers** who are all deceased, and **three sisters** who are still living.

Joanna reports she has **two brothers** - one with **two sons**, and the other with **one daughter** - and a **sister** who has **three daughters**. Joanna's parents are both living. Her **mother, Robyn**, is 68, and her **father, Harold**, is 71. Robyn has three living sisters, two living brothers, and one brother who died of a **heart attack in his 70s**. Robyn's parents (Joanna's maternal grandparents) are deceased - her **grandfather died at 66 of a heart attack** and her **grandmother died at 76 from breast cancer**.

Harold has **two sisters who are deceased**, and a **79-year-old brother named Wayne**, who was diagnosed with **bowel cancer four years ago**. Harold's parents are both deceased; his **father died at 81**, and his **mother at 77**, with no known history of cancer.

Both Ben and Joanna identify as **Aboriginal Australians**. There are **no other known cases of cancer on either side of the family**.

Discussion Questions:

1. After constructing the pedigree, describe **Ben's risk** for bowel (colon) cancer. Describe **Joanna's risk**.
2. As Ben's genetic counsellor, what **key issues** would you discuss with him regarding family history and genetic testing?
3. Is there anyone on either side of the family you'd like to **investigate further** to support your view of a possible **inherited cancer syndrome**?
4. What is the relationship between **cell cycle regulation and cancer**? What distinguishes a **cancerous cell** from a **normal cell**?