

Gathering information and drawing a genetic family tree

How can busy health professionals gather family history information in an efficient way? This factsheet outlines a set of questions, found useful in clinical practice, which if asked systematically will help to ensure that important information is recorded. A family tree will be built up on the following pages by using the questions.

Most people are happy to collect together the required information (often consulting other family members) if they are informed before attending a clinic. When asking for family information, please remember that some people may feel guilty or ashamed that they could have “passed on” an altered gene for a serious condition to another family member. Also, as people may have had recent bereavements, please enquire as sensitively as possible.

General questions to gain a quick overview of a genetic family history

Each specialty will have key questions about symptoms and signs which would alert the clinician to the possibility that a genetic condition might be present in a family, but the following are helpful in quickly gaining an overview.

“Do you have any concerns about diseases or conditions that seem to run on either yours or your partner’s side of the family?”

“Does anyone have a major medical, physical or mental problem? Has anyone ever needed treatment in hospital? Has anyone ever had any serious illnesses or operations? How old were they at diagnosis?” (Avoid just asking “Is everyone well” as past medical history may not be offered!)

“Have any adults, children or babies died? How old were they and what was the cause of death? Have there been any miscarriages or babies who were stillborn?”

The minimum information required for a targeted family history

- Draw the siblings, children and parents (first degree relatives) of your informant, and their names, dates/year of birth and any significant illnesses.
- It may not be necessary to record names of members of the extended family although it may be important to show how many unaffected people there are in the family.



Fig 1.

Drawing a family tree

Karen Hill has been diagnosed at the age of 35 as having breast cancer. As there are other cases of cancer in her family history, she is concerned about the chances of her children developing cancer.

The first step is to build up a family tree to appreciate the relationships of the people with cancer, which will now be illustrated, step by step. The parts of the pedigree newly drawn as a result of the next set of questions will be shown in red.

1. Start with the person giving the information (informant)

Use a pen (not a pencil) as you are writing a medical record. A pedigree drawing template (such as that devised by the NHS National Genetics Education and Development Centre) can be helpful to achieve uniform symbols.

Start at the bottom or top of the page according to whether you will be developing the tree “upwards” from a child, adding parents, aunts, uncles and grandparents, or “downwards” if the informant is a grandparent. The plan should be to move systematically through the family relationships, usually recording partner, children, brothers, sisters and parents moving across and then up/down the generations. *“Please may I ask if either of you come from large families with many brothers and sisters or aunts and uncles?”* If so, consider rotating the page and drawing in landscape format.

Draw the symbol appropriate for the gender of your informant

Karen Hill has been diagnosed as having breast cancer at the age of 35 years (Fig 2.).

- Mark your informant with an arrow.
- Write in her or his name and date of birth and medical information.
- Consider asking about occupations and environmental exposures if appropriate to the condition.

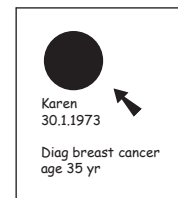


Fig 2.

Now ask about partners

Karen is married to Andrew (Fig 3.).

“Are you married; or have a partner?”

“Have you been married before or had any previous partners?”

Be sensitive when trying to determine if partners are related by blood (a consanguineous relationship).

Questions may include:

“Are you and your partner related (except than by marriage)?”

“Are there any surnames or maiden names in common in the family?”

“Did any couples in the family have the same surname before they were married to each other?”

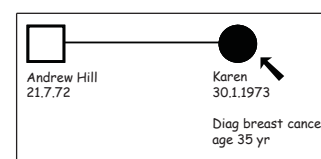


Fig 3.

2. Ask about the children of the person giving the family history

Karen and Andrew have a son and two daughters (Fig 4.).

“How many children have you had? Have you lost any children?”

“Are all your children with the same partner or were any with previous partners?”

“Please give me the names of your children, and their dates of birth in order of their ages, starting with the eldest first.”

If possible draw the firstborn on the left.

If there is a current pregnancy, record the date of the last period (LMP) or expected date of delivery (EDD).

For some conditions, it may be appropriate to ask about miscarriages, stillbirths or deaths in each partnership. Some people may find this upsetting. Being open often helps:

“Have you lost any children?”

“Did you lose any babies or have any other pregnancies?”

Record the number of weeks for pregnancy losses (*“I am sorry to hear of your loss”* if it occurred recently). If a pregnancy was terminated (rather than lost spontaneously), sensitively ask if there was a medical reason.

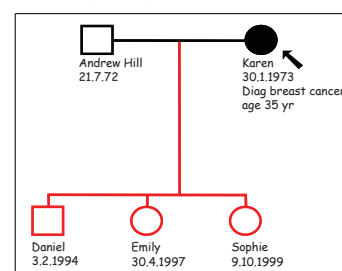


Fig 4.

3. Next ask about the siblings of the informant, their spouses and their children

Karen has a brother Paul, whose daughter Jessica is fit and well. Karen’s mother had a son (John) with a previous partner.

John has a daughter (Fig 5.).

“How many brothers and sisters do you have; have any of your brothers or sisters died?”

“Do they all share the same mother and father?”

You may need to draw in their parents at this stage to make the relationships clear.

“How many children have each of your brothers/sisters had?”

“Did they lose any children during pregnancy or in childhood?”

Ask about medical conditions.

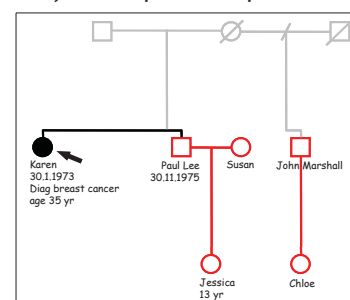


Fig 5.

4. Record details about one parent of your informant

Karen's mother Margaret Lee died at 62 yr with breast cancer; Karen wants to know if her daughters are at increased risk. Ernest, Margaret's first partner, died from lung cancer (Fig 6.).

Ask for name, date/year of birth and any medical conditions.

Do you need to collect details on the parent's brothers and sisters and their own parents?

Deciding how many generations to include usually depends on the reason for taking the pedigree.

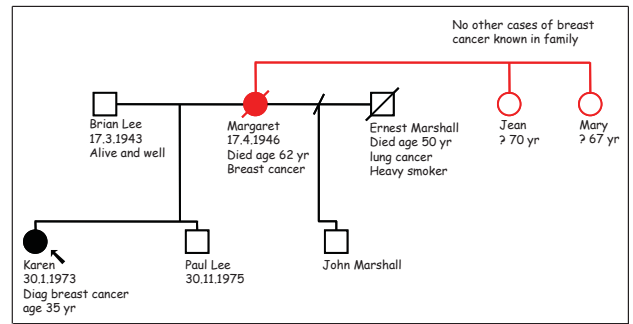


Fig 6.

5. Then ask about the other parent

Karen's father had three siblings, but Karen has no further details (Fig 7.).

Her side of the family is now complete.

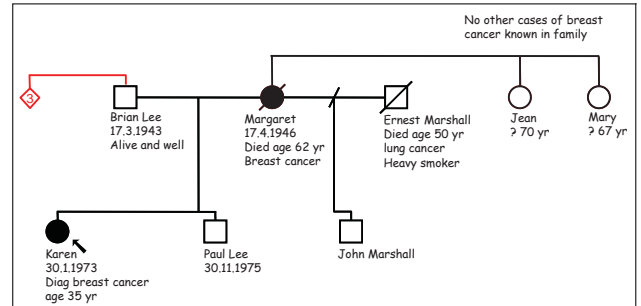


Fig 7.

6. Collect information about your informant's partner and his/her family

Return to Karen's partner, Andrew Hill, and collect information on his side of the family, working systematically through the generations using the same questions as before.

Andrew has a brother with haemophilia; their father has diabetes but their mother Patricia aged 52 is well. Her father died at the age of 24 years. He had had problems with excessive bleeding (Fig 8.).

To conclude

For some genetic conditions it is important to ask – "Originally, where did each of your grandparents come from?" This may help the laboratory target testing to specific gene alterations more common in certain populations.

As well as thanking the informant for the information, ask: "Is there anything else you think I should know that I haven't asked about?"

Most important last steps

- Date and write your name legibly on the pedigree together with an explanation of any abbreviations.
- Record the person giving the information (if you have not already placed an arrow next to their symbol).
- Always ensure that you have asked for permission to share family history information with colleagues and/or other family members and note this.

The completed family tree is shown on the next page, together with further tips about presenting information.

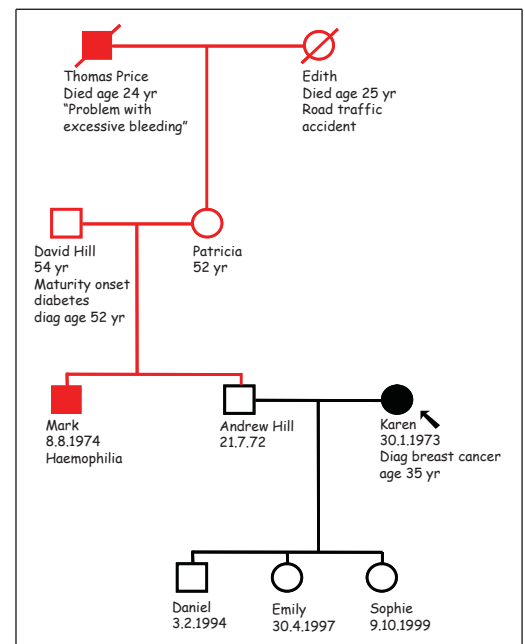


Fig 8.

