

# GENETIC COUNSELLING FOR HEMOPHILIA

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Published by the World Federation of Hemophilia (WFH).

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Treatment of Hemophilia Monographs  
Series Editor  
Dr. Sam Schulman

# Table of contents

Introduction .....	1
About genetic counselling .....	1
Definition of counselling .....	1
Who should provide genetic counselling? .....	2
Levels of counselling .....	2
When is it best to raise the issue of inheritance? .....	2
Ethical considerations .....	2
Genetic testing of children .....	3
Factors affecting genetic counselling .....	3
Approaches to genetic counselling .....	6
Basic counselling principles .....	6
Aims of genetic counselling .....	6
Map for the session .....	7
Case illustration .....	11
Conclusion .....	13
Acknowledgements .....	13
About the author .....	13
References .....	14
Glossary .....	15



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# Genetic Counselling for Hemophilia

Riva Miller

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## Introduction

Genetic counselling remains an important part of haemophilia care whether in countries at the forefront of diagnosis and treatment or those where there are little to no facilities for diagnosis or treatment. People with haemophilia, carriers, and their families can, with some knowledge, be helped to make more informed choices about having children where there is a possibility that the child might have haemophilia.

Comprehensive genetic counselling includes addressing a wide range of issues and offering a spectrum of diagnostic and carrier tests. However, even very basic information can be a first step in helping people make choices that are right for them about having children. Providing information is very important where resources for diagnosis and treatment are limited, and in circumstances where there is an increased chance of inherited disease, such as when blood relatives marry(1).

Genetic counselling does not rely only on very expensive products. It does, however, require knowledge about haemophilia inheritance and an ability to talk sensitively to individuals whose differing religious, cultural, social, and personal situations impact profoundly on the decisions that they make about having children. Personal perceptions about haemophilia also influence these decisions. These differ between individuals, and undoubtedly are influenced by treatment availability and facilities for comprehensive haemophilia care in their country.

Genetic counselling is relatively easily integrated into routine medical care in all countries, whatever the level of facilities and services, once its importance is recognised alongside laboratory testing and clinical care. In some medical facilities with a high level of

technical resource, genetic counselling can all too easily be given a less important place in overall haemophilia care based on assumptions that people have explored and understood the issues related to inheritance that they might face. This is not always the case. In other facilities where some diagnostic capabilities exist, genetic counselling may be overlooked due to a lack of personnel able or willing to take on the task. In developing countries, where there are relatively few facilities for medical care, genetic counselling might be considered to be too complex a task amidst the many other medical and social problems. It is in these situations where the biggest contribution can be made in helping people make more informed decisions about having children.

This monograph, based on experience of working in facilities with range of skill and services (U.K., U.S.A., India, Pakistan, the Philippines, Iran, and Hong Kong), reviews genetic counselling, concentrating on some issues relevant to developing countries. It includes some guiding counselling principles and a framework for practice(2), which can be adapted appropriately in a range of different settings.

## About genetic counselling

### Definition of counselling

“Counselling” is a word that is often misunderstood. In many countries it is a foreign concept. Thus it is of importance to establish some common understanding of what counselling is and what it involves. There is a difference between having a discussion, giving information, and counselling. Discussion assumes that there is a dialogue, but does not imply that a method is used to ensure that the information being given by the health care provider is understood and accepted. Providing information is an integral part of all health care,

but does not necessarily allow for a dialogue or an assessment of the patient's understanding and beliefs. Counselling, on the other hand, is a way of addressing the implications of the information and of reaching a better understanding between the healthcare worker and patient about a range of issues.

Genetic counselling for haemophilia focuses on facts about the medical condition (what it is, how it is treated, and how it is passed from generation to generation; personal and relationship concerns related to this disease; beliefs and wishes of the person discussing haemophilia inheritance, as well as those of others who might be affected).

### **Who should provide genetic counselling?**

Doctors and nurses are best placed to raise issues about inheritance during routine care of patients. Their relationship with patients, as well as their specific health promotion and care roles, provides them with the most appropriate opportunities and access to families who might be affected by haemophilia. However, a range of other healthcare workers can contribute and offer some degree of genetic counselling through their specific roles with patients. In many countries, physiotherapists have important contact with patients with haemophilia, especially where treatment resources are limited or lacking. They can readily be trained to do some genetic counselling whilst treating patients simply by checking their patients' knowledge about haemophilia, giving some information, and providing links with appropriate experts. This also applies to social workers and psychologists. Laboratory personnel, in some circumstances, give results to patients and likewise can acquire skills to do some genetic counselling that uses the contact in an optimal way. Members of a haemophilia society, who have knowledge and interest, can increase their skills in counselling and giving information to their members. They are a key resource for gaining trust and offering support to members and their families. In many situations they may be the first point of contact. To use these opportunities to the maximum, good links with identified experts for genetic information and counselling, such as interested haematologists,

are necessary to provide a comprehensive service in this area of haemophilia care.

### **Levels of counselling**

It follows that different levels and depth of genetic counselling can be provided which address different issues. Basic requirements for providing counselling are knowledge of inheritance patterns and what tests are available, and personnel able to interpret test results. Doctors have to take major responsibility to oversee the area of genetic counselling and ensure that counselling is carried out by those who are well informed.

### **When is the best time to raise the issue of inheritance?**

Ideally, with women who have a family or bleeding history, the possibility or certainty of being a haemophilia carrier needs to be raised as early as possible, before their health or that of any offspring is compromised. With patients known to have haemophilia, opportunities should be taken to check knowledge about haemophilia and its inheritance whenever possible. Educating carriers about the possibility of inheriting or passing on haemophilia should take place when marriage is being contemplated or before any children are conceived. Inheritance needs to be discussed when a child or adult is newly diagnosed.

Reaching carrier women is often difficult and relies largely upon the willingness of the index patient (the person who comes for medical care, usually adults with haemophilia or the child with haemophilia and parents) to inform other family members. Finding out more about carriers in a family and encouraging them to come in for counselling can be incorporated into medical consultations with 'index' patients.

### **Ethical considerations**

Ethical considerations include human rights, issues surrounding consent, and those pertaining to confidentiality. These apply to genetic counselling in both national and international contexts and vary according to the legal and social customs in particular settings. Concerns

about disclosure of personal information (such as the results of genetic testing) to others (family members, insurance companies, etc.) and maintenance of confidentiality can raise difficulties for some individuals because genetic testing and screening may reveal unexpected or awkward information, for example about paternity.

Although individuals have a right to confidentiality regarding personal issues, this raises concerns about the rights of those who might be affected through inheriting genetic disease to such pertinent information. Uninformed family members might suffer in different ways. For example, someone who does not know he has haemophilia might lack appropriate medical care and treatment, or a carrier might have a low level of factor VIII herself. On the other hand, a woman might assume that she carries haemophilia when in fact she does not because of different paternity.

Disclosure of information by healthcare professionals to family members should uphold accepted standards of confidentiality by encouraging individuals to disclose haemophilia inheritance themselves to those who might benefit from the information. However, the best interests of the person with haemophilia, partner, sister, or child of a carrier can be in conflict. Genetic counselling can address and consider these very issues.

In the healthcare setting, part of responsible care is to obtain informed consent for whatever treatment, genetic testing, or screening is offered. Families faced with serious genetic disease need opportunities to obtain reliable information and to consider it before making decisions about their or their future children's lives. Key issues in haemophilia are diagnosis of symptomatic children and adults, and presymptomatic testing of those who may benefit from preventative treatment. Testing factor VIII and IX levels of potential carriers, the daughters of obligate carriers, and those with haemophilia is important for their own health.

## Genetic testing of children

Genetic testing to see if a child carries a gene for a disease raises issues about who should give consent, when it should be raised, and how to consider the child's best interests. In all contexts account must be taken of physical, emotional, social, cultural, and psychological aspects as they affect the child and family. In many Western countries the courts will consider the truth is best for the child. This may differ in other contexts and has to be appreciated when approaching health and inheritance concerns.

Carrier screening of children is a more complex issue because of obtaining informed consent. The right age to start talking about inheritance is a vital consideration in all countries and societies. The age when tests and screening can be implemented depends on the views of parents, which in turn are influenced by personal and family dynamics, cultural and religious beliefs, and the laws governing the rights of the child. Even when the young person understands the implications and clearly consents to testing, complex situations can arise from screening, such as paternity and future insurance cover.

Checking factor VIII levels of a child who might be a carrier can be an opportunity for discussion about inheritance. This includes potential and known carrier sisters, nieces, and nephews of the index patient who might be carriers or have haemophilia. It often takes time to bring together the wider family to talk. In some circumstances, geographical distance, lack of transport, and financial resources can make it impossible.

Despite these possible pitfalls, every opportunity should be taken to discuss carrier testing with parents and with people with haemophilia. This is especially true when medical follow-up and future contact are uncertain, and these young people may not be able to benefit from testing at a later date when having their own children.

## Factors affecting genetic counselling

A number of issues affect genetic counselling and should be considered when addressing this aspect of haemophilia care. Some issues are

universal, such as the influence that different perceptions of haemophilia have on the decisions people make about having children. Other issues are more specific to developing countries, where diagnostic testing and treatment may be limited and a number of divergent factors have to be considered.

#### *Perceptions of haemophilia*

Perceptions of haemophilia are complex and have a major impact on the choices people make about having a child who might be affected(4). Personal beliefs and religious and cultural traditions contribute to these perceptions and influence general attitudes to illness, disability, and medical treatment. Because of their profound impact, these beliefs should *always* be explored with patients. Alongside these influences, the prognosis, or the course of the disease, is affected by the availability of diagnostic tests and treatment(4). In developed countries, with adequate treatment availability, haemophilia may not seem such a difficult disease to cope with. The prognosis is different where treatment opportunities are limited and consequently children's joints are affected at an early age. In such situations the disease has a profound impact on the day-to-day personal, social, and economic life of the individual and family functioning. In addition, different family members have their own perspectives. The sisters of people with haemophilia are very often uninformed. They may be uncertain about their carrier status. They will be influenced by what they see in the family with their brother or other relative.

In many cultures it is not unusual for a couple's parents-in-laws to be very influential about the acceptance of disability, having children, and how haemophilia is managed. Often the in-laws are uninformed about the medical aspects of haemophilia.

#### *Disclosure factors*

Whether or not haemophilia is openly discussed by the person with it or amongst family members is closely linked to personal perceptions of haemophilia. Sometimes a man with haemophilia is unwilling and unable to accept his haemophilia and thus is reluctant to

tell his daughter that she is a carrier, fearing rejection by his partner and offspring. This may leave a daughter with unresolved issues and misconceptions about haemophilia. Or, the father with haemophilia may be willing to talk to his daughters but his wife resists due to the effect it may have on the daughters' chances of marriage. In other situations, family members may be ignorant of the possibility of inheritance. Parents may not tell female children about inheritance, or siblings with haemophilia may not pass on information to each other.

Some people with haemophilia may have misconceptions about having haemophilia. They may believe that it is no problem, or that it is devastating or fatal from seeing people with HIV and haemophilia.

Carrier daughters may have problems deciding whom to tell and when. They may be afraid it will affect their marriage chances. There is also the guilt of passing on haemophilia(5).

#### *Severity of haemophilia*

The severity of haemophilia A and B should always be kept in mind in genetic counselling as this too affects perceptions of haemophilia. Although people with less severe haemophilia who do not have spontaneous bleeds appear to have fewer problems on a daily basis, bleeds from trauma (an accident, activity, or invasive procedure, such as surgery) can lead to long-term severe joint damage. Undiagnosed haemophilia, even when it is mild to moderate, can be a risk to the health and life of the affected person(6).

#### *Clarity of diagnosis*

In some countries there can be confusion over haemophilia A and B and other bleeding disorders. This is especially the case where there is a high rate of rare bleeding disorders because of marriages between blood relatives. Families may have several affected members or a combination of bleeding disorders. This complicates genetic counselling.

#### *Healthcare priorities*

Priorities for health care generally and for haemophilia specifically may make even limited

care difficult to deliver because of the high cost of treatment. The proportion of people affected by haemophilia is small compared with more common diseases (such as tuberculosis, heart and infectious disease, diabetes, diarrhoea) particularly in developing countries(6). Many developing countries either lack or have very limited supplies of blood product treatment and no facilities for diagnosis of haemophilia and carrier testing. In delivering haemophilia care, the first priority is reliable diagnostic laboratories alongside a structure for clinical care of those diagnoses(7,8,9). Even where these facilities do not yet exist, whenever possible basic information about haemophilia can be distributed amongst a wide range of health carers to ensure that patients suspected of having bleeding disorders are not given improper treatment. In one developing country, for example, many hysterectomies were performed on women who were later found to have an inherited bleeding disorder. The task giving out information about inherited bleeding problems will inevitably fall upon a few self-selected, interested doctors. Qualified haematologists may have the job of distributing information amongst those specialties most likely to meet patients with bleeding disorders, such as general practitioners, dentists, gynaecologists, obstetricians, and paediatricians.

#### *Practical considerations*

Access to diagnosis and care in countries where there are few specialist facilities, vast distances to travel, and very limited finances severely limit opportunities for testing of many affected people. In many countries there is also a difference between care in rural and urban areas. In some countries language barriers can be a problem. In India, for example, there are 14 official languages and 500 different dialects, as well as different castes and sub-castes with many religions. This makes genetic counselling even more complex.

#### *Attitudes to disability*

Having a healthy boy is very important in male-dominated cultures especially. Couples may try several times to have a healthy son, resulting in some families having several children with haemophilia. Different religious beliefs strongly influence attitudes to disability.

#### *Religious and ethical beliefs*

Different religious, ethical, and cultural factors related to marriage, childbearing and childrearing, disability, and sexual relationships can impact on how individuals and families manage inherited disease and disability. A major challenge for healthcare professionals is to balance the realities of haemophilia inheritance with hope and actions that are consistent with a person's religious beliefs.

#### *Marriage*

Beliefs about marriage are strongly influenced by religion but also by cultural and family traditions. For example, in countries where Islam is dominant there is insistence on marriage for young people. Close relatives often marry, as did the saints in Islam which serve as models for some religious families. In India a man with haemophilia may find a partner, but a woman who has a child with a disability risks being rejected by her in-laws.

#### *Sexual taboos*

Traditions related to sexual experience also have relevance for genetic counselling. The age of marriage and sexual intercourse influences when it is appropriate to raise inheritance issues. In most developing countries adults do not talk openly about sex with their children. Children gain information from peers or, very seldomly, from school teachers. The subject is avoided by everyone.

#### *Pregnancy and having children*

In most developing countries having children is seen as "settling" the family and supporting the continuation of the family. Thus, in some families, there are several children with haemophilia. In Islamic countries, pressure to have children increases after marriage: divorce is not uncommon if couples cannot have children. Termination of pregnancy is often a sensitive issue. It is rarely an option for religious Catholics. In Judaism and Islam there is permission for termination of pregnancy in carriers when there is a genetic disorder. In addition, exceptions can be made to allow contraception if there is a serious medical problem. In some male-dominated societies men do not want a vasectomy. Prevention of pregnancy because of disability or for any other reason is

difficult. However, in some countries, carriers are permitted to terminate a pregnancy if there is a genetic disorder. In India, women with a genetic disorder carry a stigma, and therefore they often wish to terminate female fetuses.

#### *Carriers*

Carriers of haemophilia may have difficulty getting married and they often keep their carrier status a secret. Symptomatic carriers who have long menstrual periods may experience additional difficulties due to religious obligations of prayer and fasting. This can be very embarrassing in their family and may lead to feelings of guilt. In many circumstances they do not seek medical help. Religious women often avoid going to male doctors, but at the same time trust in female doctors is very low in some developing countries.

#### *Circumcision*

Circumcision is very important for the Islamic and Jewish religions. Some religious people would not allow their daughters to marry uncircumcised men with haemophilia. Under Islamic law exceptions can be made in some rare circumstances if there is a serious medical risk. This is an important issue when there is a severe bleeding risk and thus must be included in genetic counselling.

### **Approaches to genetic counselling**

Issues about childbirth are complex even without problems of genetic inheritance. Having information about inheritance allows individuals, whatever their religion or beliefs, and level of resources and education, to make more informed choices. There is a need to approach the subject with sensitivity in accordance with beliefs, religion, and family values, particularly in developing countries, where many people with haemophilia remain undiagnosed. It can be hard to know where to start when the level of knowledge about haemophilia and resources for treatment are very limited and may be hard to access.

Having a theoretical framework in which to think about these problems when doing genetic counselling can help cover the many issues(2). It

allows the counsellor to have a point of reference from which to guide practice, and to monitor and evaluate the outcome of counselling. The cornerstones of this framework are a) guiding counselling principles, b) clarity about the aims of genetic counselling and c) the “map” or structure for the session(10). The proposed framework can readily be adapted to different approaches and various settings.

### **Basic counselling principles**

The basic principles guiding practice include:

- Do not make *assumptions* about people’s wishes, beliefs, knowledge, or concerns, but ask questions;
- Set *small goals* for the session that are achievable;
- Remember that *everything said has an impact* so use words carefully and avoid jargon;
- *Share responsibility* with patients and do not try to reassure them about issues that cannot be resolved easily or where there is uncertainty.

### **Aims of genetic counselling**

Being clear about the aims of genetic counselling in each circumstance is of first importance both for the person conducting the session and for the person seeking help or being informed. Clarity about the aims of counselling helps to achieve the tasks effectively in a limited amount of time. The main aims of genetic counselling are to:

- Help people *make informed choices* about their own and their offspring’s well-being.
- Ensure *informed consent* for testing and procedures is obtained and that the discussion includes information about:
  - haemophilia, its prognosis, and the therapeutic options;
  - how haemophilia is transmitted;
  - tests for diagnosis of haemophilia and carrier status and their reliability;
  - procedures for carrier screening;
  - prenatal diagnosis, tests, and risks to mother and fetus(11);
  - procedure for giving information about test results;
  - views about termination of pregnancy;
  - implications for future and existing children and other family members;

- unexpected or awkward information that might be revealed by genetic testing (for example, about paternity).
- *Balance the realities* of haemophilia inheritance with a person's religious and personal beliefs and wishes.
- Enable patients with haemophilia to *disclose* their condition and alert other family members to the inheritance risks, and support them in the decisions that they make.
- Offer *opportunities* for individuals, couples, and families to be included in some genetic counselling sessions.

### Map for the session

The "map" for the session includes the steps which can guide the genetic counselling session and enables many complex issues to be covered in whatever time is available.

These steps are given as a guide to the discussion. They are not all-inclusive or prescriptive and do not need to be covered in the exact order. They can be adapted to suit each situation.

1. *Introduce* the discussion and start engaging with the patient by clarifying:

- your role (doctor, physiotherapist, social, or laboratory worker).  
For example: *"I am the physiotherapist, and whilst we are treating your knee I would like to find out how much you know about haemophilia and how it might pass from one person to another"*.
- how much time is available as this helps contain the session and to put the person more at ease.  
For example: *"We will be together today for half an hour, but can find time to meet again if necessary."*
- what the patient wants to gain from the meeting (expectations), as this guides how best to start and use the time.  
For example: *"What do you most want to gain from our meeting today?"*

- what you consider is necessary to impart.  
For example: *"I want to inform you about what haemophilia is and about what tests we can offer."*

2. Use questions to find out the *person's concerns, knowledge, beliefs, and wishes* before giving information as all these influence how it is received. Ensure, as far as possible, that it is understood that haemophilia is a life-long bleeding disorder that can lead to disability.

For example:

*"Tell me what you understand about haemophilia."*

*"From whom, or how, did you gain this information?"*

*"What is your view about the possibility of having a child with haemophilia?"*

*"What are your beliefs about how you might manage and treat a son with haemophilia?"*

When talking to mothers of patients with haemophilia, or to fathers with haemophilia about their daughters or sisters, the following questions may help to draw out their views, and thus promote thought about complex issues.

For example:

*"At what age do you think you could talk to your daughter about haemophilia?"*

*"What do you wish to do about informing your daughter that she will carry the gene for haemophilia?"*

*"What do you want to do about telling your sister about the possibility of being a carrier?"*

*"What is your belief about having a child with haemophilia?"*

3. *Provide information in small steps* by using questions to find out how much is known, clarifying and correcting misconceptions, and then filling in the gaps. This is a way of moving at the patient's pace, and avoids making assumptions and ensures understanding.

For example:

*“You say that you know little about haemophilia – what is the little that you do know?”*

*“You are correct that it has something to do with the blood, but not that it can be cured.”*

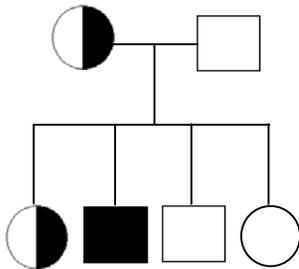
*“At present, in this country, we can do tests to see if it is haemophilia or some other bleeding disorder. If it is haemophilia we can offer some treatment if you have a child with haemophilia. This treatment will only help to stop a bleed, but will not cure the condition which is life-long”.*

This approach is especially important when discussing complex antenatal tests, their risks, and the steps involved (see step 15).

4. *Check understanding* throughout the discussion by asking the individual to review their understanding of what has been said.

For example: *“Can you tell me what you have understood so far about what haemophilia is and how it is passed on?”*

**Figure 1:**  
Inheritance of haemophilia  
Female carrier



**Legend:**

- ◐ Female carrier
- Female (not carrier)
- Male with haemophilia
- Male without haemophilia

This chart shows how haemophilia is inherited from a woman who is a carrier. Sons of a carrier have a 50 percent chance of having haemophilia. Daughters of a carrier have a 50 percent chance of being a carrier.

5. *Discuss haemophilia inheritance* by using questions and then fill in the gaps in information about how it is passed from one generation to the next.

For example:

*“What do you know about how haemophilia is passed on from one generation to another?”*

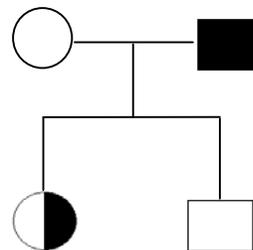
*“What do you know about your daughters’ chances of being a carrier of haemophilia?”*

The use of a pictorial chart of inheritance is helpful, whatever level of understanding. (See figures 1, 2, and 3.)

Clarify the genetic inheritance pattern of haemophilia using the chart, particularly that:

- Sons of men with haemophilia will not have the condition, nor pass it on to the next generation.
- All daughters of men with haemophilia will be carriers and will have a 50 percent chance of having daughters who are carriers and sons that have haemophilia.

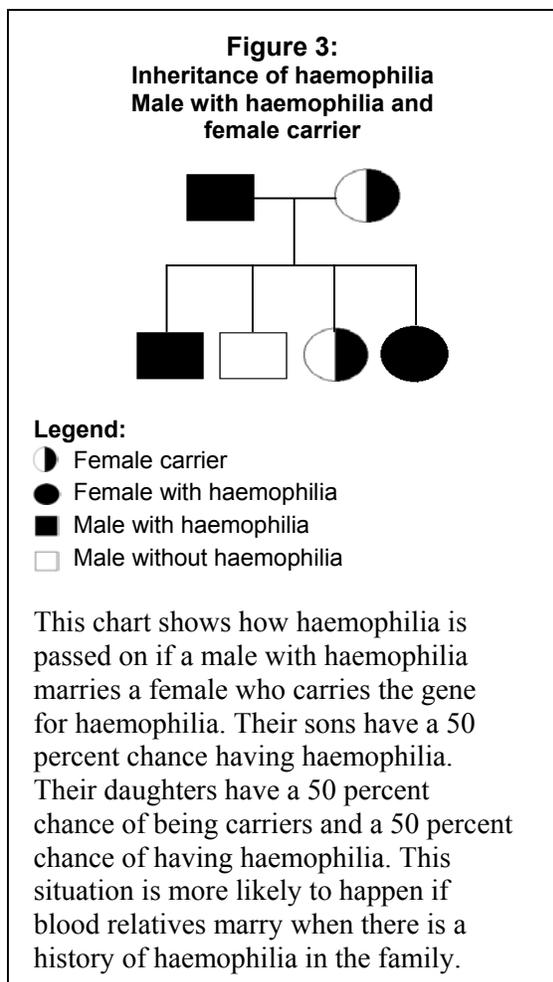
**Figure 2:**  
Inheritance of haemophilia  
Male with haemophilia



**Legend:**

- ◐ Female carrier
- Female (not carrier)
- Male with haemophilia
- Male without haemophilia

This chart shows how haemophilia is passed on from a man with haemophilia. All the daughters of a man with haemophilia will be carriers. All sons of a man with haemophilia will not have haemophilia.



- There can be sporadic cases detected through the birth of a child with haemophilia when there is no known family history of bleeding.
- The female will be a carrier if she has more than one son with haemophilia.
- If a male with haemophilia marries a female who carries the gene for haemophilia, their daughters have a 50 percent chance of being carriers and a 50 percent chance of having haemophilia. There is a 50 percent chance that the couple's sons will be affected by inheriting their mother's x chromosome. This situation is more likely to happen if blood relatives marry when there is a history of haemophilia in the family.

Ensure that the person does not have another disorder such as factor X or XI deficiency or von Willebrand disease.

6. Explore the personal *bleeding history* of the person attending the session including:

- bleeding or bruising at birth or when giving birth;
- bleeding associated with teeth coming in and dental extraction;
- circumcision;
- menstruation;
- easy bruising; and
- bleeding after trauma and surgery.

7. Continue with a *family history of bleeding* by constructing a pictorial representation or family tree (see page 12 for a sample family tree).

Careful discussion often reveals either past deaths or bleeding in the family. Pay particular attention to:

- birth trauma of infants or mothers;
- bleeding after surgery or trauma; and
- deaths due to unknown/uncertain cause.

Using a diagrammatic representation of the family tree with the individual or family makes it easier for the patient or couple to follow and to trace offspring, despite level of education and degree of information about haemophilia. The family tree is a helpful way of engaging the index patient (whether person with haemophilia or carrier) and raising their interest/concern sufficiently to bring in extended family(12).

As members of the family are mentioned they can be added to the chart using symbols that depict male and females and lines that connect the generations. Dates of birth and death can be included. This is a way of recording affected members and those possible carriers who might need testing. It is easier to look at this family tree than to read written notes about family connections.

8. Explore the *implications of haemophilia* for the patient and others in the family and their views about this.

For example:

*“What does your husband know about the possibility of having a child with haemophilia?”*

*“How might having a child with haemophilia affect your husband?”*

*“Who else knows about the haemophilia in your family?”*

*“What do your parents-in-law know about haemophilia? How might they react if they knew?”*

*“If your mother were here would she have a different view as she has had a son with haemophilia?”*

*“How do you think haemophilia might affect your son as a small child/adolescent/adult?”*

9. Clarify the different *levels of haemophilia* from mild to severe:

- The severity of the missing clotting factor usually affects whether bleeds are spontaneous or result only from trauma.
- Those with mild/moderate haemophilia need information in case of trauma or the need for invasive procedures.
- Members of the same family usually have the same factor level.

10. Discuss *haemophilia treatment* available to the person. However, because people have access to information via the Internet and other media, it is important not to avoid discussion about treatment available in other countries if it is raised(3). Issues to cover include:

- what the person knows about treatment;
- what is available in his or her situation;
- what is ideally available; and
- how best to access care.

For example: *“How does the information you have had so far influence your attitude to having a child with haemophilia?”*

11. Explore how the individual or couple might *cope with a child with haemophilia*, from a practical and social perspective (finance, housing, transport to medical care, schooling, and dealing with friends and neighbours). Consider who might be helpful and supportive and who might make it more difficult.

For example:

*“If your husband was here what might he say was his main concern?”*

*“If you had a child with haemophilia what might be your greatest concern?”*

*“If you had a child with haemophilia how might you manage?”*

*“Who or what might be of help to you? Is there anyone who might make it more difficult?”*

12. Consider *who might be involved in the discussion*. Whenever possible see people with partners and other family members who are involved. Usually the individual is seen first, then invited to bring the partner. Sometimes it is the members of the extended family who have concerns and misinformation which might place restrictions on the couple, such as a mother-in-law.

For example:

*“If your mother-in-law learnt about your concerns what might worry her most?”*

*“You say that it is the social stigma that made your parents stop you telling your husband before marriage. What was it that worried them most?”*

*“Do you think if they were here and heard about your wish for a child, but also your fears, they would understand why you haven’t had one yet?”*

13. *Find out and rank concerns* in order of priority. This helps to reduce anxiety to manageable proportions by dealing with one issue at a time. In genetic counselling this is particularly difficult as there is so much information to be given as well as the patient’s concerns to address.

For example:

*“Of all your concerns about having to tell your in-laws, dealing with your husband’s worries about a child with a disability, the pressure to have a circumcision, and your reluctance to have antenatal diagnosis – which worries you most right now?”*

14. *Address concerns and help manage whatever decisions* are made.

For example: *“You say your main worry is dealing with your in-laws. How might you start to do that? Who could help most?”*

15. *Discuss the options* from the perspective of the individual and the couple:

- what ideally can be done;
- what can be offered in their situation;
- what are the chances of passing on haemophilia;
- what are the options for antenatal diagnosis (when can it be done, opportunities and risks, fetal sexing)(11); and
- what are their views about termination of pregnancy.

16. Allow *time for questions* towards the end of the session. These questions highlight what has not been covered or understood and reveal individual concerns.

For example: *“Is there anything you would like to ask or say at this point that you haven’t understood, you would like to discuss again, or has not already been raised?”*

17. *Assess* the situation from what has been heard and seen before discussing decisions with the individual or couple. Important aspects to note are:

- physical situation (health of person, distance from centre of diagnosis and care);
- psychological state (past and present); and
- stage of consultation (pre-pregnancy, pregnant, further children).

18. Decision-making for the patient and/or partner and the doctor/counsellor includes *obtaining consent* for testing. Too much discussion can make decision-making more difficult so this should be kept in mind and a balance be achieved through giving information, finding out their views, and guiding the interview.

For example: *“From our discussion do you agree to go ahead with the tests?”*

If a decision has been made to go ahead with pregnancy without full information about carrier status, review and explore:

- possibilities of having a child with haemophilia; and
- concerns and how the individual or couple might manage these with respect to the person

with haemophilia, family, other children, marital relationship; whom to tell/not tell.

For example:

*“As you have decided to go ahead and take your chances what is your main thought at this time? Have you any concerns?”*

*“What helped you come to this decision?”*

19. End by:

- *summarizing* the discussion including the areas of certainty, those that remain unclear, and the patient’s strengths and difficulties.
- arranging follow-up or clarify when and who can be contacted in case of need.

20. *Make notes* on the conversation including:

- topics discussed;
- areas of difficulty or uncertainty;
- decisions made; and
- actions to be taken.

It is helpful to put some of the summary points in writing for the patient if this is possible.

### **Case illustration**

A case example is used to further illustrate the theory and use of the “map” for the session in the practice of genetic counselling, taking into account some of the specific issues mentioned about religious and social pressures.

Mr and Mrs Aziz came to see the haematologist, having been referred by a gynaecologist because of her heavy periods which were of great concern to Mrs Aziz. She knew that she might be a carrier of haemophilia, but it took her some time to tell her husband. There was great pressure on her and her husband to have a child, starting after about one year of their two years of marriage. They were living with her husband’s parents and during Ramadan Mrs Aziz was feeling very unwell and upset as she tried to hide her bleeding problem. This crisis forced her to reveal that her brother had severe haemophilia and that prior to her marriage her parents had hidden this from her husband’s family. It only came to light when she was taken to hospital.

Many of Mr and Mrs Aziz's concerns came out in the first session with the haematologist, including the fear of the stigma of a disability; concerns about how they would cope if a child had haemophilia; worries about the need for circumcision if it was a boy; apprehension about antenatal diagnosis, which the couple wanted, but would be against the Islamic religion. The husband hid his feelings about only now hearing that his wife might be a carrier and was resigned to help them both come to a sensible decision at this point. At the time of this first meeting Mr Aziz's fear of his parents' reaction was greater than his need to make an informed decision with his wife about having a child who might have haemophilia. The following excerpt from the conversation covers some points made in the session.

**Haematologist:** You say that neither of you have yet told your husband's parents about the possibility of having a child with haemophilia. What has stopped you?

**Mr Aziz:** My mother would be afraid of the community hearing.

**Haematologist:** What would her fear be about that?

**Mr Aziz:** That it might affect my sisters' chances of marriage, as they will think I carry bad genes.

**Haematologist:** Mrs Aziz, what are your parents' views about you telling others?

**Mrs Aziz:** They arranged my marriage and would not let me tell my husband until I was married that my brother has haemophilia.

**Haematologist:** Mr Aziz, what effect did that have on you when you heard?

**Mr Aziz:** I don't really know much about haemophilia and I am upset but understand why my wife did it and I want a child now.

**Haematologist:** What do you know about haemophilia?

**Mr Aziz:** Not much.

**Haematologist:** What is the little that you do know?

**Mr Aziz:** It has to do with bleeding.

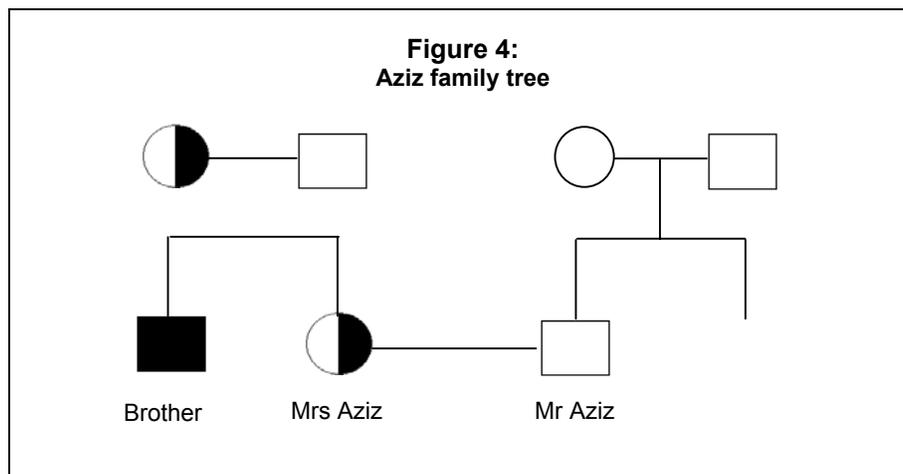
**Haematologist:** Mrs Aziz can you tell your husband what you understand? (Information is fed in about the condition and the counsellor moves to discuss inheritance and then returns to the impact on the family).

**Haematologist:** So I understand, if I am correct, that your main concern right now is telling your husband's parents because you live with them. What might we do about that? (The counsellor introduces the idea that he is willing to help).

**Mr Aziz:** Maybe you could tell them about it.

**Haematologist:** How would I do that? (It is important to help the couple to do the telling and not to take over).

**Mrs Aziz:** Maybe we could ask them to come in with us.



**Haematologist:** *Mr Aziz what do you think about that? Could you find a time to begin to talk to them and persuade them to come? (The counsellor again helps Mr Aziz do the task himself).*

**Mr Aziz:** *I could, but it is not usual for us to talk like that. In addition, they will say that it is God's will. In our culture we are pushed to have children as soon as we are married and we have been married two years now.*

**Haematologist:** *So from all the issues we have discussed what is the main thought you might take away?*

**Mr Aziz:** *That I have to find a way to break the news to my parents, and that it might be a way of relieving us of some stress. Even if they are angry at first I think they will finally accept it.*

**Haematologist:** *And you Mrs Aziz, what thought might you have?*

**Mrs Aziz:** *Only that my husband does not reject me.*

Very few of the important issues related to haemophilia or antenatal diagnosis and carrier status were covered in this first session. Nevertheless, the beliefs and fears of importance to the couple were uncovered and some first steps were made towards helping the couple to deal with them firstly themselves and then with the help of the haematologist.

## Conclusion

Childbirth and disability are sensitive issues which are influenced by family, practical, social, religious, and personal beliefs. Genetic counselling is complex because it covers both factual and non-factual issues(13).

In developing countries, genetic counselling is one relatively inexpensive way to help people deal with haemophilia whilst conditions for diagnosis and treatment improve. In some situations it may only be possible to give people information about inheritance without being able to offer carrier tests and treatment for those who have a child with haemophilia. This might seem hard to do, but

offering people time to explore the issues is a first step towards helping them cope in their particular situation. It is important to achieve a balance between giving information and discussing the implications so that people do not leave confused and unclear about the main issues for them.

Genetic counselling, including education, is an aspect of haemophilia care that should accompany the setting up of laboratories for diagnosis and monitoring tests. The laboratories offer technical knowledge and expertise whilst genetic counselling can translate this knowledge into practice to help those affected make more informed decisions. Having one without the other is an inefficient way forward and this monograph suggests one approach to closing this gap. Having some clear aims and guidelines to help cover the important issues enables a range of healthcare professionals and haemophilia society members to carry out this task. It is, however, the doctors who should oversee this task to avoid misinformation and to ensure that information is updated and appropriate to the settings where care is offered.

## Acknowledgements

Some information about different religions and cultural beliefs was provided by the Chaplaincy Department of the Royal Free NHS Hospital Trust and I wish to thank Rev. Robert Mitchell and his team for this contribution.

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## Glossary

**Carrier:** A woman who carries the affected gene for haemophilia and passes it on to her offspring. Carriers can be mothers, daughters, or siblings of men with haemophilia, or other more distant female relatives.

**Carrier screening:** Testing to identify if the female siblings of boys with haemophilia or daughters of known or unknown carrier mothers have the gene for haemophilia.

**Gene:** A unit of heredity that is transferred from parent to child and determines some characteristic of the child.

**Genetic counselling:** A process to help people affected by a genetic disorder who are planning to have children to understand their condition, how it is passed on from generation to generation, and other issues that may have an impact on their decision to have a family.

**Genetic testing:** Testing to identify if a person has a defective gene capable of causing a hereditary condition.

**Index patient:** The first person in a family to be identified with haemophilia. It is usually the person who comes in for medical care, either an adult with haemophilia or a child with haemophilia and his parents.

**Obligate carrier:** Daughters of a man with haemophilia are called obligate carriers because all of them will inherit the haemophilia gene and be carriers.