

# GENETIC COUNSELLING FOR HEMOPHILIA

Revised edition

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# WFH

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*Treatment of Hemophilia* Series Editor:  
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# GENETIC COUNSELLING FOR HEMOPHILIA

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## Introduction

Knowledge is power. There is no other aspect of our lives where this statement is truer than when it comes to our health. Increased awareness of diagnosis, treatment, and support for hemophilia worldwide has led to increased recognition for **genetic counselling** within this disease. Advances and improved availability of **genetic testing** ensure a firm place for genetics in the management of hemophilia, through genetic counselling, **carrier** detection, and reproductive management.

In countries where hemophilia management and care is fully established, genetic counselling forms an integral part of the comprehensive care team. It provides people with hemophilia, and their families, clinical and scientific information, supports individuals through the decision-making process regarding prenatal and carrier testing, and offers individualized psychosocial assessment and counselling before and after genetic testing. The process improves an individual's control of their situation and reduces the psychological distress of adapting to a genetic disease.

Access to trained genetic counsellors is limited in developing countries where resources are restricted or scarce. Therefore, it is the role of the hemophilia treatment centre (HTC), national member organization (NMO), and the comprehensive care team to provide genetic counselling and the necessary information to help patients make informed decisions.

Whatever the level of facilities and services, genetic counselling should take into account a patient's experience and perception, as well as the influence of social, cultural, and religious contexts. For example, decisions surrounding conception or an ongoing pregnancy depend heavily on the attitude towards and acceptance of hemophilia by the at-risk individual and family.

This publication provides genetic counsellors, medical social workers, medical and laboratory geneticists, and other health educators with a premier resource regarding

genetic counselling for hemophilia. It focuses on genetic counsellors as distinct professionals, opportunities for collaboration with other healthcare providers within the HTC/NMO, the genetic counselling process, and a framework for practice. Factors that influence genetic counselling and challenges that arise in a resource-poor setting are discussed. It also addresses topics stemming from the interface between rapidly advancing genetic technologies and the views of individuals who may have hemophilia or are carriers. The information is intended to prepare and support providers to fulfill the process of genetic counselling for hemophilia. While some details provided in this monograph are specific to hemophilia, many of the general concepts also apply to genetic counselling of patients and families with a history of other bleeding disorders.

## About genetic counselling

### Definition of genetic counselling

The term "genetic counselling" was first coined in 1947, with many definitions being proposed over time. A widely accepted definition states that (1):

"Genetic counselling is the process of helping people understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease. This process integrates:

- 1) Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.
- 2) Education about inheritance, testing, management, prevention, resources, and research.
- 3) Counselling to promote informed choices and adaptation to the risk or condition." (2)

The ultimate goal of genetic counselling is to help patients use key genetic information to make informed decisions about their health. It enables people to understand and adapt to the medical, psychological, and familial implications of the genetic aspect of their disease. Genetic

counselling involves the transfer of information and the assessment of the impact of that information on the patient and their family. Regardless of whether an individual has an inherited or sporadic genetic condition, genetic counselling can have a profound impact on them and their family.

In hemophilia, genetic counselling helps patients and families understand the diagnosis and its implications, ultimately helping affected individuals to adjust to living with the condition. This is particularly important for the reproductive choices of people who have hemophilia or are carriers. Genetic counselling provides information about natural, assisted, and non-reproductive opportunities available for family planning (3), and also helps to proactively prepare for the possibility of having a child with hemophilia.

### Who are genetic counsellors?

Genetic counsellors are specifically trained healthcare professionals who possess the expertise to discuss **risk assessment**, genetic testing, disease management and prevention, as well as available resources and research opportunities. In essence, they are advocates, communicators, and educators equipped to discuss the benefits and limitations of options, while adapting to the patient's understanding and beliefs. Together with the results of genetic testing, genetic counsellors can provide a higher quality assessment of the risks and the options facing the person with hemophilia.

Traditionally, genetic counsellors work in public and private hospitals and clinics, serving patients of all ages and in a variety of healthcare domains. For example, they may work with prenatal, pediatric, or cancer departments. Advances in genetic technology have increased the need for genetic counsellors in non-traditional settings, such as genetic testing laboratories, insurance companies, and specific multidisciplinary clinics, including HTC's.

### Training of genetic counsellors

Academic training comprises coursework, laboratory exposure, research experience, and extensive clinical training. Typical programs are two years in duration and offer medical- and psychosocial-driven courses in disciplines such as developmental anatomy, medical genetics, statistics, ethics, and cultural competency. Clinical training sites include prenatal, pediatric, cancer, multidisciplinary clinics, adult genetic counselling and laboratory exposures.

To become certified genetic counsellors at the end of their training program, genetic counsellors typically undergo an assessment by a credentialing organization. For example in the U.S.A. individuals must take an examination offered by the American Board of Genetic Counselors. In the U.K., Australia, and South Africa the process involves submitting a portfolio of evidence. This process is administered either through a credentialing body such as the Health Professions Council of South Africa or a professional body such as the UK Association for Genetic Nurses and Counsellors. This process helps to ensure that genetic counsellors have achieved the necessary standards for academic and clinical experience, and are committed to maintaining their knowledge throughout their careers.

While the majority of genetic counselling training programs are located in the U.S.A., many training programs have also been initiated internationally. Programs in Australia, Canada, Japan, South Africa, and the U.K. are fostering the training and development of genetic counsellors in geographic areas less familiar with the profession. These programs are similar in structure and aim to develop individuals who can independently and successfully provide genetic counselling services. The Transnational Alliance for Genetic Counseling was founded in 2006 to unify the efforts of training programs worldwide (1).

### Role of genetic counsellors in specialized care centres

Specialized care centres, which originated in the U.K., have become a mainstay in the care of patients with inherited bleeding disorders. In the U.S.A., specialized care centres for hemophilia were created in the late 1940s (National Hemophilia Foundation), and the comprehensive care model for hemophilia was formally introduced through a federally funded campaign in the early 1970s. This approach provides specialized care through a team of healthcare professionals, each with clinical expertise in a particular area. Multiple studies have since demonstrated the effectiveness of this approach (3). The WFH has long held that the wide-ranging needs of people with hemophilia and their families are best met through the coordinated delivery of comprehensive care by a multidisciplinary team of healthcare professionals with the experience and expertise to attend to their physical and psychosocial health (4).

Genetic counsellors possess a broad skillset easily integrated into specialized care centres. In the HTC/NMO setting, genetic counsellors must understand and explain the natural history and inheritance of hemophilia, educate people about how hemophilia can be passed through the family, and identify at-risk family members (3). While many healthcare professionals are able to collect a family history, genetic counsellors have been specifically trained to assess the family history and provide a risk assessment, which benefits the whole HTC/NMO team.

Genetic counsellors obtain a family history and draw a family tree, called a pedigree, to document individual family members and their relationship to the **index patient**, or person who initially came to medical attention. Approximately 50 per cent of cases have no known family history of hemophilia; in the remainder of cases, hemophilia is inherited through several generations of a family. By documenting affected, symptomatic, and unaffected individuals in a pedigree, genetic counsellors, physicians, and patients have access to a visual summary of the condition's inheritance pattern. In this way at-risk individuals may be identified and noted in the pedigree such that recommendations can also be made for family members. Additional roles of a genetic counsellor collaborating with an HTC/NMO may include facilitating familial communication about the diagnosis, outreach to relatives who may be at risk of having or being a carrier of hemophilia, and collaborating on research studies relating to genetics.

Genetic counsellors use their training in **gene** structure, testing strategies, testing methodologies, and application of findings to integrate genetic testing routinely into the care of patients and families with hemophilia. Genetic testing availability varies greatly between countries and laboratories, and comprehensive testing may not always be available. Sequencing of the *factor 8 (F8)* and *factor 9 (F9)* genes, for hemophilia A and B respectively, is available in numerous countries. Systematic analysis of these genes can identify variations from the expected sequence or structure, which may be the cause of hemophilia. While the majority of DNA variations found in *F8* and *F9* genes are likely disease causing, it cannot be assumed that every identified variation has a significant impact on gene function. Interpretation and classification of genetic variations, once identified, are critical to the clinical usefulness of test results. This is also critically

important when **potential carriers** in a family are interested in pursuing carrier testing. While there are many benefits to performing genetic testing, the cost of testing is often a limiting factor. Therefore, genetic counsellors should discuss the genetic test(s) that are available and appropriate, as well as the potential use of the findings of those tests. By listening to the patient's feelings and needs, genetic counsellors can support the patient to make an informed decision.

The psychosocial impact of chronic disease on a family is well documented (5, 6). Individuals and families with hemophilia are often concerned about job security, finances, childcare, schooling, medical insurance, and fear of the unknown. Genetic counsellors in the HTC/NMO setting are able to address many of these issues and relay relevant psychosocial concerns to other HTC/NMO providers or refer to outside resources.

### Value of genetic counsellors

Perhaps the greatest value that genetic counsellors provide to any clinical setting is their ability to adapt to the needs of the patient, provider, and clinic. They are trained to handle the complexity of genetic testing and integrate test results into clinical care. Genetic counsellors often act as a liaison between the healthcare field and patients, helping to identify and provide resources and support. As skilled communicators, they strive to help people understand key information, often breaking down complex medical information into lay terms that even young children can understand. It is very important that individuals who have hemophilia or are carriers have a good understanding of their diagnosis so that it can be managed appropriately. Genetic counsellors can help patients and their families to integrate medical and scientific concepts and apply them to their lives.

### Who can provide counselling in the absence of a genetic counsellor?

Despite the success of the comprehensive care model, an estimated 80 per cent of the worldwide hemophilia population do not have access to comprehensive care (WFH 1997). This tends to be the case in developing countries, where access to health care and factor treatment are limited, but also in developed countries where smaller care centres are located far from the main centres or in remote areas. Given that only a minority of patients are treated within the comprehensive care model, the majority have

little or no access to genetic counsellors. Even within HTC, the availability of a genetic counsellor varies.

Potential means to fill this gap include:

- Hiring a genetic counsellor.
- Contracting a genetic counsellor from a local genetics clinic to work with the hemophilia team as needed.
- Establishing a relationship with a local genetic counsellor to coordinate referrals.

In the absence of a formal relationship between an HTC/NMO and a genetic counsellor, the responsibility of delivering important genetic information is often met by other team members (3). Physicians, nurses, or psychosocial professionals within the clinic typically assume the role. However, these individuals have other clinical responsibilities, which can result in no one on the team viewing genetic counselling as his or her priority.

In this case, the HTC/NMO can assess the skills of existing staff and designate one individual to provide genetic counselling for patients and families. Allocating the responsibility of genetic counselling to one individual within the hemophilia team may maximize the consistency and standard of genetic counselling. This individual should become familiar with the genetic counselling process and critical educational topics, such as inheritance, genetic testing, and reproductive choices. Knowledge gaps may be addressed by identifying and obtaining appropriate learning resources or referring the patient to another professional. Clinics without direct access to genetics professionals are encouraged to organize periodic and collaborative educational sessions with such professionals, to increase team understanding and awareness (3).

If none of these options are feasible within the resources of a clinic, patients may receive fragmented and incomplete genetic counselling. The overall quality of the patient experience may also be diminished. If several healthcare providers are involved in the genetic counselling process, then it is essential to clearly define roles among team members.

#### EXAMPLE:

Ms. Smith spoke to the psychosocial professional at her clinic during a routine visit to the HTC. Recently married, she is considering having children, and reported feelings of anxiety and stress around the idea of having a child with hemophilia, since her brother and uncle have severe hemophilia A. The psychosocial professional counselled her about the risks of being a carrier of hemophilia and elucidated that the patient's anxiety stemmed from the possible financial burden she and her husband could incur. The psychosocial professional discussed financial strategies to better prepare for the long-term care of a child with hemophilia. The psychosocial professional offered follow-up discussions with Ms. Smith and her partner and ensured that she knows where to go for help, including learning more about the genetic testing options available.

#### Role of psychosocial professionals

Psychosocial professionals focus primarily on psychosocial needs assessments, emotional coping skills, and personal and family dynamics in order to address and support a wide variety of concerns or issues for patients with a diagnosis of hemophilia. Genetic counsellors are aware of these same aspects, but are primarily concerned with how they impact a patient's ability to process and integrate genetic information. Further, genetic counsellors have a primary role to educate about the natural history of hemophilia, perform risk assessments related to the inheritance of hemophilia, facilitate genetic testing, and discuss relevant reproductive options. The individual roles of the genetic counsellor and psychosocial professional are quite different; however, providing these services collaboratively can enhance the overall patient care experience.

Psychosocial professionals and genetic counsellors have similar goals regarding patient care. Both educate patients about their diagnosis, helping them to better understand their treatment, management, and long-term prognosis. They validate patient concerns, fears, and anxieties related to chronic illness. Through supportive counselling, validation of concerns often heard in the HTC/NMO setting (e.g. financial burdens, impact on the family, risk to offspring), and planning for potential outcomes, these healthcare professionals help patients develop coping strategies that minimize feelings of burden. Identifying and providing resources is a key component for genetic

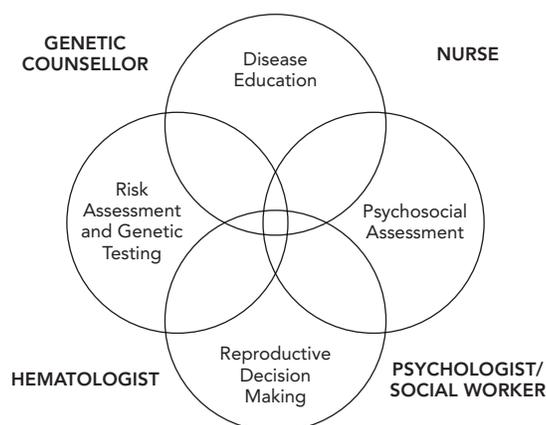
counsellors and psychosocial professionals, and urges patients to take ownership of managing their condition.

### Delivering collaborative genetic counselling services

Care within a comprehensive HTC/NMO typically involves coordination of services and close working relationships between hematologists, nurses, psychosocial professionals, physical therapists, genetic counsellors, and many other disciplines (Figure 1). While offering specific expertise for patient management, collaboration between providers ensures maximum patient care.

#### EXAMPLE:

A comprehensive HTC created an educational program for patients and their families. During the event, HTC staff arranged for the children of attendees to visit the local children's museum. Simultaneously, educational seminars were conducted for the adults. Team members presented topics in their area of expertise, including recognizing bleeds, working with the school nurse, financial planning, obtaining insurance, inheritance counselling and genetic testing, appropriate dental care, and safe exercises and physical activities. This successful collaborative approach with discipline-specific input increased knowledge within the hemophilia community and the patients benefited greatly.



**FIGURE 1.** Collaboration of HTC/NMO providers to deliver genetic counselling services

This figure depicts some of the potential overlap of HTC/NMO providers in addressing the various components of genetic counselling. Each multidisciplinary team should address the skills and qualifications of the staff in order to determine which provider(s) are covering the various components of genetic counselling within the HTC.

## Considerations for developing countries

Resources and technological advancements in developed countries have helped to integrate genetic counsellors and genetic testing into hemophilia care. While these resources may not be readily available or affordable in developing countries, it is still important to recognize the potential value of genetic counselling. Even in the absence of ideal resources, the process of genetic counselling is still important, as it may improve knowledge, reduce psychological distress, and improve informed decision making for a patient or family.

### Importance of family history

Genetic testing in an individual with hemophilia can often provide a definitive genetic explanation for disease. In this instance, genetic testing of female relatives can also definitively distinguish between carriers and non-carriers of hemophilia. This information maximizes risk assessment, enables patients to make informed decisions, and can provide the HTC team with important information to assist with the management of carriers. In the absence of genetic testing, the individual providing genetic counselling has the task of assisting the patient to make decisions and adjust to the potential diagnosis, despite the uncertainty. In such cases, the family history can be a very powerful tool for hemophilia risk assessment.

### Family and community outreach

Genetic counselling sessions may identify other family members who could benefit from knowing their genetic status. It helps them make informed decisions regarding their own health care and reproductive choices. Some patients readily share this information, while others need support and guidance from individuals providing genetic counselling to communicate with their family. Outreach that targets individuals with hemophilia or previously undiagnosed hemophilia carriers may benefit people who need help communicating. Educational or primary outreach programs could have greatest influence on females of reproductive age who are potential carriers.

The potential psychosocial impact of hemophilia carrier status is critical in certain societies and communities. A thorough assessment of sociocultural issues is essential and should form the basis of large-scale awareness programs aimed at improving comprehension of hemophilia among young adults, patients, family members, and

medical professionals. Such outreach efforts may be even more effective if there is involvement from the community's religious and social leaders, social organizations, and patient support groups. This can maximize the potential for risk assessment and case finding within given societies and cultures.

### **Identifying resources to maximize genetic counselling**

In developing countries (e.g. Botswana, Brazil, Ghana, India, Pakistan), genetic counselling is most often provided by physicians, psychosocial workers, and nurses. Development of a comprehensive care team is important for the provision of genetic counselling in such countries. To maximize impact, individuals providing genetic counselling should seek local, regional, national, or international resources for themselves, as well as for the patients and families whom they serve. For example, in South Africa, genetic counsellors can sometimes be found in private centres or in large, academic centres. These latter may attend hemophilia clinics.

HTCs/NMOs constitute a vital resource for obtaining information and discussing options available to the hemophilia community. Non genetic health professionals working with an HTC/NMO benefit from participating in conferences or training workshops on genetics and genetic counselling. These educational events are a good source of information to disseminate among colleagues, and ultimately to patients and families. As mentioned previously, establishing a working relationship with an expert in genetic counselling for hemophilia can provide an invaluable resource for questions or issues that may arise.

Hemophilia societies and national organizations may provide patient and family-oriented discussions, workshops, and training camps to disseminate information and increase awareness of various aspects of the disorder, including genetics and genetic counselling. They may have access to some educational and communication resources that explain the essentials of the genetic counselling process for individuals, couples, and families. Peer support groups for women who are carriers may also provide information and education about hemophilia and genetic counselling.

## **Factors affecting genetic counselling**

Understanding the information shared during a genetic counselling session is often the first step in furthering any decision-making process. Several factors influence the way patients and their families perceive the genetic counselling process; some issues are universal, whereas others are specific to certain cultures, religions, or individual situations. It is the role of the genetic counsellor or supporting professional to identify and find ways for patients to overcome factors that may affect genetic counselling (7).

### **Severity of hemophilia**

The severity of hemophilia, and an individual's perception of this severity, influences the importance of information discussed during genetic counselling sessions. The need for education correlates with increasing severity of hemophilia and its increasing impact on the quality of life of patients and their families. Regardless of the severity, genetic counselling has the potential to empower individuals and minimize avoidable complications in the future.

### **Perception of genetic counselling**

The process of genetic counselling initially involves some degree of uncertainty for most people. Unfamiliarity may create unwanted anxiety, leaving some patients less receptive to genetic counselling or to hearing the information discussed. Patients and healthcare professionals may incorrectly perceive genetic counsellors as being directive when presenting reproductive choices to patients when in reality a fundamental characteristic of the profession is to be nondirective. Other misperceptions include equating genetic counselling with psychotherapy or expecting that genetic counselling consists solely of performing genetic testing.

### **Healthcare considerations**

Hemophilia affects a small proportion of a given population and is not generally considered to be the highest priority for healthcare services. This is particularly true in developing countries where major public health problems persist (e.g. tuberculosis, malaria) and poverty, tobacco and alcohol use, psychiatric disorders, and injury contribute additional burden.

### Practical considerations

Accessibility and affordability of genetic counselling are limiting factors in certain countries. Many HTC and NMO are unable to offer genetic counselling, and people are often required to travel vast distances to seek services. In some countries illiteracy is a problem. Literacy equips people to understand and deal with their diagnosis more effectively; literate people tend to take an interest in available literature, and seek services with an open mind. Illiterate people often form unrealistic expectations and resort to alternative methods of coping, leading to poor outcomes.

### Marriage and consanguinity

Marriages are arranged in some cultures, often with the head of the family deciding the marriage of his daughters. There may be concerns around disclosing whether a daughter is a carrier or potential carrier of hemophilia for fear that no suitable alliance will be forthcoming, or that having a child with a disability could lead her to be rejected by her in-laws. In contrast, a male with hemophilia may find a partner without much difficulty.

#### EXAMPLE:

The wife of a male with hemophilia was concerned because their soon to be married daughter was having difficulty understanding and accepting her **obligate carrier** status. The mother and father brought their daughter to her father's next clinic appointment at the HTC/NMO. The physician asked if the daughter would be willing to speak to the genetic counsellor, to which she agreed. During their discussion, the daughter disclosed that she was most concerned with how and when she should disclose this information to her future husband. The genetic counsellor was able to talk through this concern with the daughter, and helped her to develop a plan with which she was comfortable. After this, the daughter was more open to accepting her carrier status and pursuing further evaluation for herself by a hematologist.

The importance of genetic counselling has been recognized in developing countries where consanguineous marriage is an integral part of social and cultural life (8). Consanguinity in a family with a history of genetic disease, such as hemophilia, can have important effects. Using risk assessment, genetic counselling can minimize

complications and present options for reducing the impact on offspring, while also addressing the psychosocial needs, concerns, and fears of the individuals impacted.

### Sexual taboos

Reproductive choices, attitudes towards sexual orientation, societal traditions, and the age at which marriage is considered acceptable, vary widely between cultures and influence when it is appropriate to educate about inheritance. Some cultures speak more openly about risks to pregnancies and may more easily accept discussion of these topics.

### Religious and cultural beliefs

Different religious, societal, and cultural factors influence decisions regarding marriage, childbearing, treatment options, and interventions related to hemophilia. These can have significant implications in a genetic counselling session. Society and culture in many developing countries do not allow a woman to make decisions, even those related to her own body; decisions about abortion or childbearing are made by the family or the husband. This gender difference can influence the genetic counselling process, as this is contrary to typical gender roles whereby women are primarily responsible for the timing of marriage and childbearing, as well as aspirations regarding family size and sex composition.

#### EXAMPLE:

In certain cultures, the male head of house is responsible for making important decisions. This can leave a female carrier of hemophilia vulnerable and at risk for stigmatization and isolation based on the inheritance of hemophilia.

It is important to recognize that even today, in Indian society for example, the desire for a male child subjects a woman to immense pressure from the family and leaves her vulnerable. For a carrier of hemophilia, implications of that status on future offspring can lead to complications in married life. While knowledge about carrier status and inheritance may reduce complications for children with hemophilia or prevent the birth of more children with hemophilia, women giving birth to a child with hemophilia may be threatened by the husband and his family, and in some cases, even mistreated and ostracized. Given the potential for adverse consequences for the woman and her child, carrier status information must be dealt with very sensitively.

**EXAMPLE:**

In a women's group discussion, a woman voiced that it is difficult to terminate a pregnancy because, culturally, this is simply not done. However, women are considered to bring good fortune to the family when they bear a healthy boy as their first born. These conflicting views on childbearing can lead to distress for women.

**Disclosure factors**

Historically, the hemophilia community has been stigmatized by co-infection with HIV, hepatitis, and other comorbidities. Some individuals fear that simply discussing specific health implications will lead to unwanted outcomes, and therefore prefer to avoid these discussions entirely.

Acquiring and maintaining health insurance continues to be a primary focus in the HTC/NMO setting, particularly in developing countries where factor supply is dependent on governments and insurance agencies. Many people fear that family history and genetic testing information, particularly for women who are carriers, may lead to genetic discrimination or difficulty obtaining health and life insurance.

**The genetic counselling process**

There are several values innate to the profession of genetic counselling that must be upheld. Genetic counsellors are trained to emphasize patient education and nondirective counselling. They are also trained to demonstrate respect for patient autonomy, confidentiality, and privacy, disclose complete information, and recognize psychosocial implications throughout the genetic counselling process (9). Regardless of training or background, it is important that the individual providing genetic counselling is cognizant of and grounded in these values.

**Components of a genetic counselling session**

A genetic counselling session comprises six routine components: 1) contracting, 2) information gathering, 3) risk assessment, 4) education, 5) psychosocial assessment and counselling, and 6) case management. By incorporating these components, the individual providing genetic counselling will accomplish the overall goals for a session, while upholding the values of the profession.

*Contracting*

Contracting is the initial step and a critical component of a genetic counselling session. It involves sharing expectations and mutually agreeing upon an agenda.

*Information gathering*

The provider gathers clinical and nonclinical information that may impact risk assessment or education. The timing and method of information gathering may vary. It is not always an isolated event, but can occur before, throughout, and after a session.

*Risk assessment*

Once information is gathered, the individual providing genetic counselling uses critical thinking and knowledge to synthesize the information and perform risk assessment for the patient and family. The overall risk assessment should be conveyed to the patient and will greatly influence discussions during the remainder of the session.

*Education*

A substantial portion of the genetic counselling session consists of patient (and family) education. It is essential that the patient be made aware of the current natural history of hemophilia. This is especially important for individuals receiving genetic counselling because of a family history of hemophilia, as the patient's knowledge may be skewed by their relative's experience. The genetic counsellor should provide education at an appropriate level for the patient and assess their level of understanding throughout the session.

*Psychosocial assessment and counselling*

Psychosocial assessment and support are critical throughout the genetic counselling process, particularly when test results lead to disclosure of a new diagnosis. Simply gauging a patient's understanding of factual information is not sufficient in genetic counselling. The information discussed may elicit emotions or feelings that are not directly expressed. Genetic counsellors have specialized training to disclose sensitive implications and to help patients recognize the psychosocial impact of such news. Ultimately, the patient should feel supported and free to express feelings, discuss concerns, and reach their own informed decisions. Individuals providing genetic counselling should be aware of their own limitations, and familiar with external resources to offer more in-depth psychosocial counselling if necessary.

### Case management

Providing genetic counselling involves much more than simply providing information. The patient-provider relationship develops during and extends beyond the genetic counselling session, which means it is often appropriate for the individual providing genetic counselling to take on a case management role. Incorporating case management into the genetic counselling process maximizes the efficiency and effectiveness of the entire process, improves patient comfort, and preserves continuity of care.

### Genetic counselling session map

This “map” is intended to guide a healthcare professional through a genetic counselling session with a patient. The composition of any session may deviate from this map due to clinical, demographic, and cultural factors. The individual providing genetic counselling should determine what to include in any specific session, and in which order.

### Contracting

The individual providing genetic counselling should synthesize the following, often brief, conversation (making adjustments where necessary), and use it to develop a tentative plan.

#### Introductions

Introduce yourself to establish your role and start building rapport with the patient.

#### EXAMPLE:

*“I am the social worker in this comprehensive care team. (One of) my role(s) is to address the familial implications of a diagnosis of hemophilia/family history of hemophilia.”*

#### Time management

Provide guidance of how much time is available.

#### EXAMPLE:

*“We have 30 minutes to meet today. If we need to continue this conversation, we can set up another session.”*

### Assess understanding

Elicit the patient’s understanding of the reason to involve genetics. This will help to gain insight into his or her perception and address any inaccuracies.

#### EXAMPLES:

*“What is your understanding of why we include genetic counselling as part of our comprehensive care?”*

*“What is your understanding of why you were referred for genetic counselling?”*

### Propose topics for discussion

Provide a summary of topics that you feel are important to cover.

#### EXAMPLE:

*“Today we can talk about your family history of hemophilia. I want to draw out your family tree, discuss the inheritance of hemophilia, and review genetic testing.”*

### Prioritize discussions

Identify important topics to cover, and the order of priority, by learning what the patient wants to gain from the session.

#### EXAMPLE:

*“What are you hoping to gain from our session today?”*

### Information gathering

Obtaining clinical and nonclinical information about a patient is critical to an effective genetic counselling session.

#### Clinical information

A personal medical history targeted to uncover information pertinent to hemophilia includes:

- Known diagnosis of hemophilia, including severity and inhibitor status.
- Previous testing, to confirm a diagnosis or to assess if further testing is available.

#### EXAMPLE:

*“You mentioned that you underwent **carrier screening**. Can you tell me what you remember about this test, such as when it was performed, who ordered it, and what the results were?”*

- Bleeding symptoms, such as easy bruising, prolonged bleeding with minor injury, menorrhagia, postoperative hemorrhaging, and postpartum hemorrhaging.
- Treatment history that may suggest a bleeding disorder, such as requiring blood transfusion.
- Complications during labor or delivery.

Family history can aid in risk assessment, help to identify relatives with a chance of having hemophilia or being a carrier of hemophilia, and determine the best testing strategy and interpretation of results. This is achieved by:

- Documenting family history in standard pedigree form, which is often clearer and more concise than describing it in words.
- Including self-reported or appropriately obtained medical information of family members.
- Including hemophilia-related symptoms or complications for any relatives (see personal medical history details, above). It can be helpful to elicit individuals previously tested for hemophilia or hemophilia carrier status, or who died from an unknown cause.

The process of taking a detailed family history can also uncover social dynamics of the family, which can be helpful when addressing psychosocial issues and responses.

### *Nonclinical information*

This includes knowledge the patient has regarding the clinical and familial implications of hemophilia. It is important to assess if the patient and family have an accurate understanding of the current clinical picture of hemophilia care and management, as this may impact views and perceptions.

#### EXAMPLES:

*"Tell me how you would describe hemophilia today."*

*"With no family history of hemophilia, where did you learn the information you know about this diagnosis?"*

*"You know a lot about hemophilia. How did you learn this information?"*

*"How do you think having a child with hemophilia would impact your day-to-day activities?"*

*"Have you thought about how or when you might tell your daughter that she is a carrier?"*

*"Now that we have discussed the benefits of knowing carrier status and identified who in your family may be a potential carrier, how do you feel about sharing this information with them?"*

*"How does the possibility of having a child with hemophilia make you feel?"*

Several factors also influence genetic counselling. Some will be inferred by other information shared by the patient or recognized by the provider (see Factors affecting genetic counselling, above). Awareness of such variables can ensure a patient-focused session.

### **Risk assessment**

Personal medical and family histories are extremely crucial for risk assessment. The individual providing genetic counselling should synthesize the information gathered to assess the following:

- Usefulness of previous test results in the context of the clinical setting.

#### EXAMPLE:

A normal factor level does not preclude a female from being a carrier.

- Probability that the person may have hemophilia or be a carrier of hemophilia.
- Probability that other relatives, including future offspring, may have hemophilia or be a carrier of hemophilia.

#### EXAMPLE:

An individual may have a 50 per cent chance of being affected with hemophilia based on family history alone. Additionally, a female with a personal history of bleeding symptoms or complications may have an increased likelihood of being a carrier.

If no family history of hemophilia is known for an individual with a diagnosis of hemophilia, it is possible that his mother is a carrier or that he was the first person in his family to be born with the

mutation causing his hemophilia. The chance that a mother in this case is a carrier is estimated to be 80 per cent, unless the affected individual is known to have the intron 22 inversion mutation, in which case the mother's risk to be a carrier is estimated to be 98 per cent.

- Ideal testing strategy for the patient or family.

#### EXAMPLES:

If a female is an obligate hemophilia B carrier, it may be appropriate to forgo genetic testing and only obtain a baseline factor IX measurement to inform management.

If a patient has severe hemophilia A and the familial mutation is unknown, it would be most appropriate to start genetic testing by looking for intron 22 and intron 1 inversions in the F8 gene, which collectively account for approximately 50 per cent of severe hemophilia A.

If a potential carrier is interested in carrier testing but the familial mutation is not known, it would be ideal for a male relative with hemophilia to pursue testing first and then release his results to his family member to facilitate their testing.

## Education

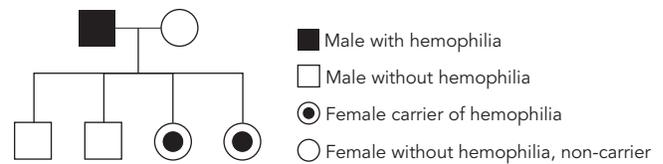
Patients and families should be educated on topics that will assist them through genetic counselling and the decision-making process related to a diagnosis of hemophilia or hemophilia carrier.

### *Natural history of hemophilia*

Perception and knowledge of hemophilia can impact the patient's feelings and reactions to their own risk assessment, or that of their offspring. The patient or family should be educated about the etiology, symptoms, management, treatment, and complications of hemophilia. Checking a patient's knowledge of hemophilia will ensure their understanding is accurate, and it will identify areas for further education.

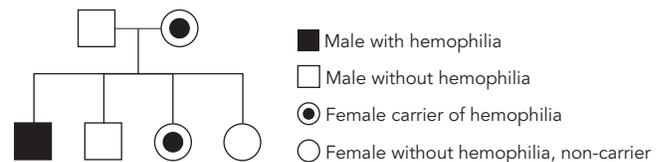
### *Inheritance of hemophilia*

Knowledge about an X-linked inheritance pattern can help a patient understand their risk assessment, as well as that of their offspring and other family members. (Figures 2, 3, and 4.)



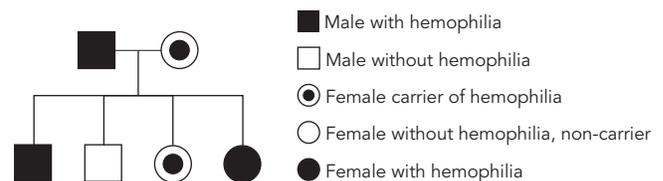
**FIGURE 2. Inheritance pattern for a male with hemophilia**

This figure depicts the chance that offspring of a male with hemophilia and a non-carrier female will inherit the gene causing hemophilia. In this scenario, all biological daughters will inherit the gene, making them obligate carriers; however, all biological sons will not have hemophilia.



**FIGURE 3. Inheritance pattern for a female carrier of hemophilia**

This figure depicts the chance that offspring of a female carrier of hemophilia and a male without hemophilia will inherit the gene causing hemophilia. In this scenario, each biological son has a 50 per cent chance to inherit the gene causing hemophilia and a 50 per cent chance to inherit the normal gene. Each daughter has a 50 per cent chance to inherit the gene causing hemophilia (making her a carrier) and a 50 per cent chance to inherit the normal, non-hemophilia gene (making her a non-carrier). The likelihood of inheriting the gene causing hemophilia is not altered by the status of previous offspring, since each pregnancy is an independent event.



**FIGURE 4. Inheritance pattern for a male with hemophilia and female carrier of hemophilia**

This figure depicts the chance that offspring of a male with hemophilia and a female carrier of hemophilia will inherit the gene causing hemophilia. The **gene mutation** causing hemophilia may be different on each side of the family, which could have implications for **genotype-phenotype correlations**. In this scenario, biological sons have a 50 per cent chance to inherit the gene from the mother. No biological sons will inherit the gene from the father. Biological daughters have a 50 per cent chance to inherit the gene from the mother and will always inherit the gene from the father. Therefore, daughters have a 50 per cent chance to be a carrier and a 50 per cent chance to have mutations causing hemophilia in both gene copies, making her affected. The likelihood of inheriting the gene causing hemophilia is not altered by the status of previous offspring, since each pregnancy is an independent event.

Hemophilia A and hemophilia B are inherited in an X-linked manner because the gene that causes each of these disorders is found on the X chromosome. Hemophilia A is caused by a change in the *F8* gene and hemophilia B is caused by a change in the *F9* gene. Males typically have one copy of every gene on the X chromosome, while females typically have two copies. This leads to gender differences in risk assessment and clinical presentation. Even if an individual is thought not to be at-risk based on the reported family history, it is important to remember that any individual can be born with a de novo mutation, which can cause hemophilia.

Historically, it was thought that only males could be diagnosed with hemophilia, and females were carriers of hemophilia. It is important to understand that female carriers may be symptomatic.

### *Testing options for hemophilia*

Two commonly used tests are sufficient to make a diagnosis of hemophilia or hemophilia carrier. An individual's risk assessment will help to determine which test(s) may be helpful. It is important to educate the patient or family on the differences between the two, as well as the testing process, benefits, limitations, costs, and expected turn-around times.

The two commonly used tests for hemophilia are:

1. Factor level testing (blood test) that measures factor VIII or factor IX protein circulating in an individual's blood. It is usually diagnostic for hemophilia in males. However, it cannot definitively determine carrier status, because the result can be normal in an individual who is a hemophilia carrier.
2. Genetic testing that assesses an individual's *F8* or *F9* gene, for hemophilia A or hemophilia B respectively, to identify if there is deviation from the normal, functional version of the gene. It is often performed on a blood sample, but can be performed on other specimen types.

Genetic testing for hemophilia should be considered for all patients with hemophilia, since it has several potential benefits, including:

- Provides molecular confirmation of a diagnosis
- Confirms a known familial mutation in potential carriers (the most accurate method of carrier testing)

- Identifies patients with increased risk of developing an inhibitor, and other genotype-phenotype correlations
- Enables prenatal diagnosis and future reproductive planning
- Determines eligibility for research studies

The ideal testing strategy is determined by risk assessment, which considers an individual's personal history, family history, and any previous testing in the family.

#### EXAMPLE:

If an individual has a known diagnosis of hemophilia by factor level, genetic testing may not be necessary to make a diagnosis. However, it may provide information about genotype-phenotype correlation or better testing options for relatives.

If a causative family mutation is known, family members with a chance of having hemophilia or being a carrier can undergo site-specific testing for the known mutation. This makes testing less costly and is the most accurate carrier testing option.

### *Genotype-phenotype correlations*

Various types of mutations and unique mutations have been identified within the *F8* and *F9* genes. Some are associated with a certain clinical presentation, such as disease severity or inhibitor risk. This is known as genotype-phenotype correlation. If a family mutation has been identified, any known genotype-phenotype correlation should be shared, as this can provide further insight into the clinical presentation of hemophilia specific to the mutation.

#### EXAMPLE:

*"The results of your brother's testing that you brought with you today indicate that he has the intron 22 inversion mutation. This mutation is associated with severe hemophilia A."*

### *Family planning options*

For individuals with a hereditary diagnosis, such as hemophilia, it is important they are aware of options available to support family planning wishes. A diagnosis of hemophilia or hemophilia carrier may influence an individual's thoughts about having children. Some people are certain of the path they want to take, while others may wish to explore alternatives. Options such as natural biological

children, non-invasive prenatal screening, prenatal diagnosis, pre-implantation genetic diagnosis, and adoption should be presented to patients and families. Importantly, individuals providing genetic counselling should become familiar with local resources to support any family planning decisions.

It is important to check understanding throughout the session. This can be achieved by:

- Using analogies to help simplify complex information.

EXAMPLE:

*"You can think of a gene like a sentence and genetic testing like a spellchecker."*

- Providing information in multiple ways to ensure the patient comprehends what is being communicated.

EXAMPLE:

*"Each son of yours has a 50 per cent chance of having hemophilia. This is the same as a one in two chance."*

- A critical part of education is assessing what the patient has learned. This assessment may take place in several ways. One method is to ask the patient to paraphrase the information covered during the session.

EXAMPLE:

*"Can you tell me what your understanding is of how hemophilia is inherited?"*

### **Psychosocial assessment and counselling**

Ongoing psychosocial assessment and counselling is important during (and sometimes after) a genetic counselling session. This can be accomplished using various methods and approaches. Attending and listening are crucial counselling skills.

#### *Determining existing support networks*

Identifying support networks allows the individual providing genetic counselling to anticipate what is available to the patient. They may have support from family, friends, or community groups. It is helpful to explore whether the patient plans to call upon the individual(s) or group(s) in this situation, and if so, how they think that support will respond.

EXAMPLES:

*"How might your husband respond to having a child with hemophilia?"*

*"You seem to be very close with your brother who has hemophilia. How might he react to your decision not to have biological children due to your carrier status?"*

*"What does your husband's family know about hemophilia?"*

*"Your family has been a big support to you through previous tough decisions. Have you discussed any of your thoughts or feelings with them yet? How do you anticipate they will react to this decision that you have made?"*

Understanding how a patient has coped in previous situations can shed some light on how they may cope in the current scenario. After a session, checking in with a patient can help assess if they are using healthy coping strategies.

#### *Exploring topics evoking a psychosocial response*

This can prompt the patient to think beyond the factual information provided, to a point where he or she reflects on how the information may impact feelings or actions.

EXAMPLES:

*"How do you think you having a son with hemophilia would impact your five-year-old daughter?"*

*"What is your biggest fear or concern about having a son with hemophilia?"*

*"Have you thought about how you might respond or feel if you find out that you are a carrier of hemophilia? What about if you find out that you are not?"*

#### *Evaluating verbal responses and nonverbal cues*

The individual providing genetic counselling can determine and rank the patient's feelings and concerns by assessing cues. For verbal responses, it may help to pay attention to trends, such as phrases that are repeated multiple times or phrases that are accompanied by nonverbal cues. The responses may prompt further investigation with the patient. Paying attention to nonverbal cues can uncover

topics that are important to the patient or emotions that the patient does not consciously recognize. Nonverbal cues may include crying, changing position, avoiding eye contact, easily being distracted, and engaging in anxious movements and fidgeting.

### *Addressing the patient's feelings and concerns*

This can help to create a safe environment to explore the implications of the information discussed, and build trust between the patient and individual providing genetic counselling. Some ways to actively address feelings and concerns include:

- Validating appropriate feelings. Patients express a wide range of feelings in response to information about hemophilia, including fear, anxiety, guilt, or a sense of loss. For patients openly expressing feelings, acknowledging reactions that are within normal limits can provide the patient with some reassurance that their response is valid. This may free the patient to continue expressing and work through their feelings.

#### EXAMPLE:

*"You appear to feel some guilt because your son inherited hemophilia from you. This is a normal reaction to learning this information and has been commonly expressed by mothers in a similar situation."*

For patients who do not openly express feelings, sharing with them some expected feelings may give them a sense of permission to express their internal reaction.

- Supporting decisions. It is important to provide genetic counselling in a nondirective way and set aside personal views and biases. Once a patient reaches a decision, it is important to ensure he or she feels supported in that decision. This may be accomplished by helping the patient secure next steps related to the decision and discussing ways to share this decision with whomever the patient chooses.
- Providing additional support or resources. After a new diagnosis of hemophilia or hemophilia carrier is made, the patient may feel overwhelmed or isolated, even if they have a solid support system. Patients may feel that others do not understand their

views or concerns about the diagnosis, or may want someone to provide guidance of what to expect.

Introducing them to an individual who has been in a similar situation can provide peer support that the patient may not find elsewhere. Existing support groups and other resources, such as brochures or videos, may also be beneficial.

### **Case management**

Case management is essential before, during, and after a genetic counselling session. Investing effort into case management issues can make the overall genetic counselling experience more efficient and effective for the patient. Some case management tasks are determined through institutional policies or practices, and individuals providing genetic counselling should be aware of those within their own institution. Examples of case management activities are detailed below.

#### **PRIOR TO THE SESSION**

##### *Gather standard information*

Collect information that will be useful for the session, such as reason for referral and family history. This can be achieved by developing a standard intake form or family history collection tool that could be sent to the patient in advance or given to them when they check in for their appointment. This reduces the time required for information gathering during the session and allows the patient to investigate and reflect on the questions being asked. The information gathered provides a starting point for discussion during the genetic counselling session.

Common reasons for a genetic counselling referral include a known diagnosis, family history, and diagnostic tests suggestive of hemophilia or hemophilia carrier status, as well as an interest in pursuing genetic testing for hemophilia.

##### *Obtain and review relevant medical records*

This is particularly important for performing initial patient risk assessment. Reviewing this information before the session also clarifies the reason for genetic counselling and saves time during the session.

##### *Review the current clinical picture*

This will help when assessing the individual's knowledge of hemophilia and hemophilia carrier status. It is also important for individuals providing genetic counselling

to research the natural history of hemophilia prior to the visit if they have not dealt with hemophilia recently. This can be accomplished by reading publications or discussing the condition with a provider currently caring for patients who have hemophilia or who are carriers of hemophilia.

#### DURING THE SESSION

##### *Consider the time available*

This ensures that all critical topics are addressed and sufficient time is available for questions, consents, blood draws, or other potential outcomes.

##### EXAMPLE:

During contracting, a patient expressed that her main interest for this session was to pursue genetic carrier testing. Nearly half the time has passed and this topic has not been discussed. It is appropriate to make the patient aware of the time remaining and discuss how she would like to proceed. Her options are either continue the current conversation and schedule a follow-up visit, or to shift focus to discuss genetic carrier testing.

##### *Facilitate informed consent for tests or procedures*

Having established a rapport with the patient and possessing first-hand knowledge of what took place during the session, the individual providing genetic counselling may be the best suited and most efficient person to obtain informed consent for testing or procedures the patient is interested in pursuing.

##### *Discuss next steps*

Based on the dialogue and any decisions reached by the end of the session, identify with the patient the next steps and establish a plan for moving forward.

##### EXAMPLE:

*"You were unsure of the severity of hemophilia in your family. As we discussed, reviewing records of a relative with hemophilia may clarify this. If you are able to obtain records, you plan to share them with me to include in your chart."*

*"As we discussed, your brother would be the best person to initiate genetic testing in your family. You are planning to update me after you discuss this with him."*

*"Although you do not want to have blood drawn today, it sounds as though you would like to pursue genetic testing for hemophilia. Your options would be to schedule a separate appointment for this blood draw, or to coordinate this testing with your next blood draw at our clinic. Which would you prefer?"*

##### *Provide anticipatory guidance about future events*

Walking the patient through upcoming situations can help to reduce patient anxiety and offer them time to prepare and reflect. For the individual providing genetic counselling, this discussion can give insight into the patient's feelings and concerns for future events.

##### EXAMPLES:

*"Your test results should take approximately two weeks to come back. Regardless of the result, I typically call patients with their results as soon as they are available. Are you comfortable with this plan?"*

*"As a carrier, becoming pregnant can have implications for yourself and your offspring. If you find that you are pregnant, please let us know so that we can proactively discuss and recommend medical management options for you and your offspring."*

##### *Summarize the session*

Recap what was discussed during the session, as well as any decisions and plans that were made. The genetic counsellor may ask the patient to summarize this information, in order to gain insight into his or her perception of the session. The counsellor must ensure that this exercise does not turn into a quiz that the patient may feel they could fail. If needed, a subsequent discussion can resolve any discrepancies.

#### AFTER THE SESSION

##### *Document details of the session in the patient's medical record*

This will serve as an official reference of the session for future providers, as well as for legal or billing purposes.

##### *Coordinate any necessary follow-up*

This involves arranging tests and referrals, and communicating test results. It is often helpful to estimate

turn-around time and establish a plan for disclosure of test results at the time of testing. This can help to eliminate unwarranted anxiety or concern experienced by the patient at the time of results disclosure. For instance, determine if you will call the patient with the results or if the patient should return to the clinic (depending on geographic proximity).

## Conclusion

Genetic counselling is a complex and essential part of comprehensive care for patients and families with a diagnosis of hemophilia. It focuses on educating patients and families about the clinical presentation, underlying genetics, and familial implications of hemophilia, while being aware of and adapting to factors that may influence patient perception. Genetic counsellors provide a supportive environment for patients and families to explore the information, make informed decisions, and work through evoked feelings and emotions. Although genetic counsellors have specialized training to facilitate this process,

### *Make yourself available*

Genetic counselling often covers new concepts that can elicit varied responses over time. Patients should be provided with information about the best way to contact you if questions or concerns arise.

other healthcare providers may assume the role of genetic counselling within an HTC/NMO.

Genetic counselling will become more critical as further information materializes about the clinical use of genetic testing and genotype-phenotype correlations in hemophilia. Even in the absence of genetic testing, genetic counselling remains integral to comprehensive hemophilia care and empowers individuals to understand the implications of a family history of hemophilia. 🌐

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## Glossary

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

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

**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 to understand their condition, how it is passed from generation to generation, and other issues that may have an impact on family planning.

**Genetic testing:** Testing of an individual's DNA to establish if he or she has a defective gene capable of causing a hereditary condition. It typically identifies a single causative mutation, which can then be used to test at-risk family members.

**Gene mutation/gene change:** Individual change in the DNA sequence. While most mutations in the *factor 8* and *factor 9* genes are disease causing, some DNA variations may have limited or unknown impacts on overall gene function.

**Genotype-phenotype correlation:** Occurs when a specific genetic alteration in DNA is known to be associated with a specific disease characteristic. For example, a specific mutation in the *factor 8* gene is known to be highly associated with severe hemophilia A.

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

**Obligate carrier(s):** Females who are definitively carriers based on family history, including all daughters of a man with hemophilia and mothers of at least two sons with hemophilia.

**Potential carriers:** Females who may or may not be carriers based on family history, including female relatives who are not obligate carriers

**Risk assessment:** Involves analysis of an individual's risk of inheriting a disease based on personal and family history, as well as results of genetic tests.

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