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# Newsletter to Health Professionals

Number 4

## Genetic counselling

**Recommendations from the experts in spinocerebellar disorders by the Medical and Paramedical Committee of AFAF, CSC and ASL**

## Edito

The purpose of the genetic counselling consultation is to inform an affected person, or his or her relatives, about the risk, or to inform parents about their risk of having a child affected by a genetic disease. The risk depends on the disease, and the clinical and molecular diagnosis must be accurate. Depending on the disease, genetic tests need to be performed to find out the cause. In this consultation, the geneticist takes the necessary time to explain the difficulties and the consequences of both the clinical diagnosis and the genetic test.

# Questions to be considered...

**To find out whether a genetic test would be useful or possible, a "genetic counselling" consultation with a geneticist or a genetic counsellor is essential.**

## a Who is it intended for?

Genetic counselling is for people confronted with a genetic disease where:

- The person is himself or herself affected
- The person has at least one family member affected (one child or more, father or mother, brother or sister, cousin, uncle, etc.)
- Couples, where one partner is affected by or at risk of a genetic disease, ask about the risk of passing the gene responsible for the disease on to their current or future children
- Members of the couple are related (for example, first cousins, etc.)

## b Why ?

The main objective of genetic counselling is to provide access to clear and understandable **information** for the person seeking advice (the consultee)

This information concerns:

- The method and risk of transmission of the disease
- The risk of being affected or of having a child affected by the disease
- The possibility of making a presymptomatic diagnosis for persons at risk
- The possibilities of making a prenatal diagnosis before birth where the estimated risk for the unborn child is high and the mutation has been identified (prenatal diagnosis, PND, or preimplantation diagnosis, PID) and of discussing other possibilities for the couple (recourse to sperm donation, egg donation, etc.)
- The treatment, where this exists, or methods of prevention, where these exist

## c The consultation procedure

During this consultation, the geneticist draws up a **family tree** with the help of the consultee. This tree allows the geneticist to carry out an "investigation" into the disease in the family and to deduce from this the method of transmission of the disease (dominant or recessive).

This is a strange moment for the person who has to gather together information about his or her family (or "tell the family story"), and recall each member and their links. This stage, which sets out the family history, may mean that the person has to confront sensitive situations, for example, by reliving difficult personal or family events or things that have been left unsaid.

Finally, this consultation, which needs to establish a climate of confidence between doctor and consultee, is private. However, joint time can be arranged if more than one family member wishes to attend together.

# What is the genetic test used for?

The genetic test determines whether or not a person is carrying a pathological gene.
It is carried out after genetic counselling, on the prescription of a geneticist. It consists of a blood test.
**The genetic test is carried out in two situations:**

## a To confirm the clinical diagnosis

A disease can have several different genetic causes, but a genetic abnormality can also lead to different clinical pictures or diseases. It is now possible to identify some genes responsible for cerebellar ataxias, including Friedreich's ataxia, and 7 of the 28 SCA genes, etc. At the present time, however, most of the genes for these diseases are not known or accessible for routine genetic testing, and many tests are carried out within the framework of research subject to different legal regulations. For this reason, explanations adapted to each person and to his or her condition must be provided in the consultation.

**Confirmation of the diagnosis can be given at any age and represents an important step for:**

- The patient, who can put a name to the disease and thereby give a meaning to what is happening to him or her. The act of naming is reassuring in itself
- The doctor, who can now propose appropriate care for the person according to the disease
- Relatives "at risk", who will need this information if they wish to undergo a (presymptomatic or prenatal) genetic test

## b To find out about genetic status through the performance of a presymptomatic

The presymptomatic test concerns any person **of legal age who** is "at risk" of developing a genetically determined disease and who wishes to find out about his or her genetic status before signs of the disease appear. In order for the genetic test to be possible, the geneticist needs to check that a genetic test has been carried out on another member of the family. The test can be performed only at the request of the person at risk. The consultee is free to abandon his or her request at any time, including the moment of the result.

# What are the stages of the presymptomatic test?

This test is prescribed by the geneticist and falls within a strict legal framework. It requires supervision by a multidisciplinary team (geneticist, psychologist, nurse, social worker, etc.) and is carried out in several stages:

- The **information phase** by the geneticist
- The **preparation and reflection** phase: the psychological interview. The aim is to allow the person to verbalise in a different way his or her questions and concerns about the genetic test and about him or herself, his or her family and future; the aim is also to help the person to anticipate the impact of the test result on his or her future life
- **Sample taking.** This consists of two blood tests and two molecular analyses that are carried out in the laboratory approved by the Ministry of Health and contacted by the geneticist for the consultee. In general, and where the test is carried out in a hospital setting, it is not charged to the patient/consultee
- **Announcement of the result.** This is never given out by telephone or letter. The result is announced by a geneticist during a consultation. A document containing the result is given out at the request of the person who has had the test

Whether favourable or unfavourable, the result always represents a psychological, family and social upheaval. As each person has his or her own particular history, however, the experience of the test procedure and the result will depend on each individual.

# And after the result?

- **For carriers of the gene:** even after an unfavourable result, carriers may initially feel a certain relief as there is no longer any doubt and the result allows the person to be informed about his or her genetic status. It is not unusual, however, for a person to be demoralised, sad and to feel like crying. This is a normal response to the shock that the result may have caused
- **For non-carriers of the gene:** they may feel relieved to learn that they will not become ill and will not pass the disease on to their descendants. But they may also be confronted with feelings that are disconcerting and distressing, such as guilt towards siblings that are affected ("why them and not me?"). When the person felt sure that he or she was a carrier of the gene and this turns out not to be true, the result may be difficult to accept because it calls into question past life choices ("I never wanted to have children for fear of passing on the disease").
- After the result, **psychological support** may prove to be necessary to allow the person gradually to "face up to" and to assimilate his or her new status

**Drawn up by the Medical and Paramedical Committee of AFAF, ASL and CSC.**
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**Responses and questions can be sent to the Medical and Paramedical Committee at conseilmedicalataxie@yahoo.fr, or 12 place Brisset – 02500 Hirson.**
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