UNIVERSITY OF CAPE TOWN

DIVISION OF HUMAN GENETICS

Level 3, Wernher & Beit North Building, Faculty of Health Sciences, Observatory 7925, Cape Town, South Africa
Fax: 021-406-6826, Telephone: 021-406-6297

UNIVERSITY OF CAPE TOWN PREDICTIVE TESTING PROTOCOL FOR HUNTINGTON DISEASE

The predictive test for Huntington disease is a process which allows an individual at risk for having inherited the gene for Huntington disease a chance of knowing whether he or she has the affected gene, before showing signs of the disease.

To take the test is a very serious decision as the knowledge of your test result will remain with you for life. Therefore, it is important that you are well informed and understand the programme and procedures you will have to go through before being given your result. A programme or protocol has been recommended by international genetic counselling teams based on their experiences in dealing with individuals and families at risk for the disease. The protocol outlined below is a guideline of the minimum required sessions. Additional sessions or a neurologist or psychiatrist referral may be arranged at the discretion of those involved in the process.

Predictive testing is not generally available to

- individuals under 18 years of age;
- pregnant couples; or
- individuals who have only recently become aware of Huntington disease being in their family as it is advisable to wait for a year before undergoing the predictive testing programme.

If, after careful consideration, you decide to undergo the test, you will be requested to come to the Groote Schuur Hospital Neurogenetic Clinic or the Division of Human Genetics on at least five occasions to see the different members of the genetic counselling team.

FIRST MEETING (GENETIC COUNSELLOR)

The genetic counsellor will ask you for your family history and explain the protocol. He/she will ask you for your reason for requesting a predictive test and will explain the inheritance pattern of the disease and the methods and limitations of the predictive test.

1. As you know, the affected gene for Huntington disease has been found and we are now able to offer a blood test that will predict whether or not you will get the disease.

   IMPORTANT: *It is very important at the beginning that you understand that there is approximately 1% possibility that the test may not show a clear result with absolute certainty. If this occurs in your case, then unfortunately your risk of carrying the gene remains exactly the same as before you entered the programme (i.e. 50%).* When the final result is given, it could be one of three:

   * *Positive* - you have the affected gene and will develop the symptoms at some stage in the future. We have no way of knowing when and how it will present.
   * *Negative* - based on our current knowledge of this gene, you will not develop Huntington disease.
   * *Uninterpretable* - no result. You are still at risk and have a 50% chance of carrying the affected gene.

2. We strongly recommend that you inform your family doctor of your decision to undertake the test. If you do not have a family doctor we recommend you find one. As part of the policy of this programme, we believe that the on-going medical care and support your doctor is able to give you is very important to you and your family. A letter will be sent to your doctor to inform him/her about the programme after the third visit.

3. The decision whether or not to undertake testing can be stressful and waiting for the result can be stressful. The results, even "good news" can take time for adjustment. Having support is essential and we therefore strongly advise that you choose a family member or a trusted friend to accompany you to all the meetings as they will also be able to provide support after the test result is given to you.

4. The final result will be given to you approximately six weeks after your blood samples have been taken and will be strictly confidential. No result will be given to you by telephone. Written confirmation of your result will be sent to the genetic centre that referred you. With your permission, your family doctor will be contacted and given the result.

5. If at any stage in the programme you decide you do not wish to continue, the decision is entirely yours. Your decision will in no way prejudice our relationship with you or your family. We will be happy to continue to offer you all the support and help you need.

6. In order to ensure confidentiality, family members who request testing together will attend individual counselling sessions with the genetic team members.

7. Because the process of being tested for Huntington disease and dealing with the results will be stressful and is often disruptive to a person's life it is best to choose a time to be tested when complicating factors from the outside are at a minimum.
    - For example, while in the middle of a divorce or break-up of a relationship, or at a stressful time at university or college or work is not a good time to be tested.
    - Testing at a time of celebration may not be optimal, for example right before or after marriage, or during important holidays.
    - Likewise soon after the birth or death of a family member testing should be delayed.
    - Predictive testing results may impact on health and life insurance, it is important to consider these implications before embarking on the testing protocol.
    - It is useful to make a decision about whether or not to be tested even if the decision is not a yes or no answer. For example, deciding not to be tested for a certain period of time (next year, or after I turn 30), can help you put this aspect of Huntington disease aside for a period of time until you are ready to readdress testing issues in future.

8. It is important that, before undertaking the predictive test, you plan about who you will tell about your decision and when. It is also important to plan about who you will tell your result and when. Will you tell them on the same day that you are given your result? Exactly how and when do you plan to tell them? Will you tell them by phone or letter or at a family get together? What if you change your mind and do not want them to know at the moment or at all? Do you plan to tell the people at work about your decision to be tested or your test result?

9. If you have attended the support service of a therapist, psychologist, religious professional or psychiatrist during a difficult time in the past, it may be useful to discuss your decision about testing with this person. This is particularly important if you have had prior problems with depression, anxiety or stress.

DISCRETIONARY MEETING (NEUROLOGIST AND ANOTHER MEMBER OF THE GENETIC TEAM) (APPROXIMATELY FOUR WEEKS AFTER FIRST MEETING)

The neurologist will perform a neurological examination to exclude early symptoms.

SECOND MEETING (PSYCHOLOGIST) (APPROXIMATELY FOUR WEEKS AFTER PREVIOUS MEETING)

1. You must be accompanied by your support partner to this meeting.

2. A psychologist will assess your past and current levels of psychological functioning.

3. During this meeting the psychologist will discuss your reasons for requesting predictive testing as well as the possible effects that a positive or negative result may have on you, your partner and your family. Counselling may vary widely. For example, people who have the gene may suffer a sense of shock and grief however well they may have been prepared beforehand. People whose risk is negative may feel relief but at the same time suffer guilt and anxiety. For those with no result their feelings could be anger and frustration.

4. You will be asked to make an appointment to see the genetic counsellor approximately one month after this meeting.

5. It may be necessary to include another interview in the pre-test period if either you or we may need further information or discussion.

THIRD MEETING (GENETIC COUNSELLOR AND GENETIC NURSE IN ATTENDANCE) (APPROXIMATELY FOUR WEEKS AFTER PREVIOUS MEETING)

1. You must be accompanied by your support partner.

2. At this meeting the genetic team will need your written consent to take blood samples from you. Again there will be careful explanation of the test and the manner in which the final result will be given: about six weeks after the blood sample has been taken you will be contacted to make an appointment to see the genetic counsellor and psychologist when you will be given the final result.

3. Your GP will be contacted once your blood has been taken and a letter will be sent to him/her about the test. No result will be given to you over the telephone, whatever the result may be.

4. Due to the complexity of the test it may take longer than six weeks.

FOURTH MEETING (PSYCHOLOGIST, GENETIC COUNSELLOR AND/OR GENETIC NURSE IN ATTENDANCE) (APPROXIMATELY SIX WEEKS AFTER PREVIOUS MEETING)

IMPORTANT: It is important that you plan what you will do after you have received your result at this session. Will you go directly home and who will be there? Will you take some time off work or from family responsibilities?

1. You will be contacted and an appointment made to finally receive your result. No result will be given over the telephone, whatever it might be. ***Please note that the genetic nurse does <u>not</u> know the result when she calls you***.

2. The genetic team in attendance, and again only with your consent, will give you the final result. You must be accompanied by your support partner at this meeting. You will be given a short recess alone with your partner after the test result has been given and this will be followed by a discussion to answer any questions you may have.

3. Once the result has been discussed with you and again, with your consent, your GP will be informed telephonically.

4. The genetic nurse will make an appointment for one month later for a follow-up visit.

FIFTH MEETING (GENETIC COUNSELLOR AND GENETIC NURSE IN ATTENDANCE) (APPROXIMATELY FOUR WEEKS AFTER PREVIOUS MEETING)

1. You may bring your support partner to this session if you wish, but may also attend on your own if you would prefer to do so.

2. During this visit the genetic counsellor will revise your test result with you and the effects this has had on you, your partner and your family. Counselling may vary widely, but will generally focus on how you are coping with this knowledge and to who you have disclosed your test result and their reactions to this information.

3. The genetic counsellor will make a further follow-up visit for one month later if you would like it.

Contact persons: **Sister Diane Sklar** **Tel: (021) 404 6235 or 406 6304**
or
Sister Trish Legg **E-mail: Diane.Sklar@uct.ac.za/Patricia.Legg@uct.ac.za**

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