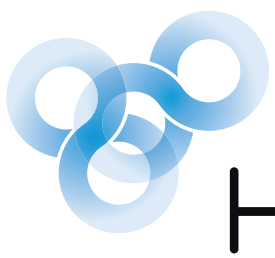
Supporting Family Testing: Talking to Your Family About HAE



HAEi

Supporting Family Testing: Talking to Your Family About HAE

Introduction and purpose

This booklet has been designed to support you in talking about Hereditary Angioedema (HAE) with your family and to help when making the decision to test undiagnosed family members for HAE. HAE is a genetic condition. If one parent has HAE there is a 50 percent likelihood that his or her child will have it too.[1]

If you are reading this leaflet, you are likely to have a confirmed diagnosis for HAE and may be considering how to talk to family members about it, as well as wondering if some of them may also have HAE and whether to seek confirmation.

The test that this leaflet talks about is to confirm a diagnosis of HAE. It involves a simple blood test that will check concentrations of the protein C1-esterase inhibitor (C1-esterase inhibitor (C1-INH) is a protein in the body) and the complement factor C4 as well as C1-INH activity.[2] For a more detailed explanation of this, please see the leaflet entitled 'The Test Process'.

There will be many different reasons that you and your family members may have concerns over the test and it is important that everyone comes to their own decision. This booklet is designed to help you think the test through together and consider the options in your own time.

Further information on the genetics

Unlike most hereditary diseases, HAE is an autosomal dominant disease, which means one copy of the altered gene in each cell is sufficient to cause the disorder.[1]

HAE is usually inherited from a parent and cannot be 'caught'. However, there are patients who have HAE but did not inherit it from their parents.[1] Around 25% of patients develop HAE as a result of a spontaneous (non-inherited) genetic mutation.[1]

Talking about it

Receiving a confirmed diagnosis of HAE has many implications and you are likely to have a lot of questions. At some stage you will find yourself wondering if other family members to whom you are related by blood may also have HAE, and considering if you should talk to them about your HAE, and whether they should seek confirmation of a diagnosis or not.[3] It can help to be honest about your feelings but it is also important to make sure you are well-informed and clear about the facts before you make decisions or embark on any difficult conversations.

If you have HAE and are concerned about other family members having it

If you have been diagnosed with HAE and are concerned that other family members may have it, you may wish to make an appointment to speak to your HAE doctor to talk through your concerns. This way you will have further background information and should feel more prepared before speaking to your family.

If you are considering talking to your children about it, you should make sure you have discussed your concerns with the other parent, if appropriate, and confirm that they are comfortable with you raising the matter with your child and are in agreement about the approach.

Your family members may or may not be aware of your symptoms or your condition. Therefore it is worth reflecting on how they are already behaving about it and around you, in the context of your HAE, as this may help you in how you approach talking to them.

Things you may want to consider are:

- Have they asked any questions in the past?
- Have they been interested or concerned about your symptoms in the past?
- How has the family member reacted to your swelling attacks or to your treatment?
- You may also want to take time to think about whether you remember if they have shown any signs of HAE or known about the condition and thought they might have it

It will be important to allow yourself time to think through all these different questions before you raise the matter and avoid putting pressure on anyone (including yourself) to make any instant decisions.

Once you have had the initial discussion, it is worth allowing everyone time to think things through, ask questions and then suggest regrouping another day to continue the discussion and perhaps reach a decision on whether to proceed with the test for HAE.

At this stage you may want to plan to see your HAE doctor as a family.

Making the decision to be tested

A diagnosis of HAE can be confirmed through a simple blood test.[2]

HAE is a condition that, with the right help, support and treatment can be successfully managed to enable people living with it to have fulfilling and active lives.[4] However, undiagnosed HAE and unmanaged swelling attacks can have serious consequences[5]. Around half of all HAE patients suffer from at least one attack of swelling in their throat or voicebox (also known as a laryngeal attack) during their lifetime.[1] This form of swelling attack can block the throat and is potentially life-threatening because of the risk of suffocation.[5] Having a positive diagnosis improves the likelihood of surviving a potentially fatal attack.[5]

How often attacks occur and how severe they are will be different for each patient.[6] The location and severity of each individual attack can also vary from patient to patient.[6] Frequency of attacks can also vary within a patients' own lifetime, for example puberty and pregnancy can be a time of particularly frequent attacks for some female patients.[7]

As explained above HAE swelling attacks may, for some patients, severely affect their lives and even be life-threatening[5], but with a proper understanding of the condition and the different treatment options, HAE patients and their families may be able to manage their symptoms and lead fulfilling and active lives.[4] While there is currently no cure for HAE, there are medications available to either prevent attacks or to treat an attack as it occurs.[2]

If an individual or the family around the individual is aware that they have HAE, there may be the opportunity to prepare for any swelling attacks that might happen in the future.[3] Being aware of the condition is the first step to taking control.[3]

A blood test can answer that question and set them on the path to receiving information and support and treatment, where appropriate and available.[2]

However, the decision to test for HAE may not always be so simple. Some patients find the decision is easy to make and others have concerns. Ultimately someone needs to feel comfortable with the decision they make and what the results may show.

I found not knowing more frightening than the truth – it was almost freeing to be rid of the question mark and able to move on with the knowledge I now had

I don't see the point of finding out when there is no cure

There was never a question about whether we would have the children tested – we needed to know so that we could give them the best care and treatment

I would rather not know

I had a responsibility to my partner to find out if I had it – only then could we make a decision about having children and how we would feel if we were to pass it on

I watched my mother live with HAE and struggling to manage it – I really can't face embarking on a similar experience

Our family life was so badly impacted by my father's HAE that I have a lot of resentment towards it and do not want to risk having to let it into our lives

Whatever the thoughts and feelings you and your family have, it is important to talk them through with each other. It may also be helpful to talk through with an HAE doctor or nurse or someone else that you know that has the condition, for example a member of an HAE patient support organization.[3] You may not be able to reach a conclusion immediately but it can help to air and talk through concerns, whatever kinds of thoughts and feelings you have.

Finally, do not make a rushed decision – you or your family member must be convinced that they are sure before proceeding with the test.

The prospect of a family member being tested for HAE, and a result confirming HAE can be daunting. It is important to remember that there is support available from other people who have HAE and specialist doctors.[3] If taking the test results in your family member/s being diagnosed with HAE, there are many treatment options to manage it effectively and reduce the impact on their daily life.[4] Please read the other booklet in this series 'The Test and Your Results' to find out more.

- HAE is a genetic condition. If one parent has HAE there is a 50% likelihood that his or her child will have it too[1]
- Testing for HAE involves a simple blood test[2]
- Whilst HAE is a condition that, with the right help, support and treatment, can be successfully managed to enable people living with it to have fulfilling and active lives, undiagnosed HAE and unmanaged swelling attacks can have serious consequences[5]
- If an individual or the family around the individual is aware that they have HAE, there is the opportunity to prepare for any swelling attacks that might happen in the future[3]
- If taking the test results in your family member/s being diagnosed with HAE, there are many treatment options to manage it effectively and reduce the impact on their daily life[2]

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