Becoming a parent (for MACS adults with condition)

Becoming a parent is one of life's most exciting journeys and having a MACS condition shouldn't preclude you from this. Below is some information about genetic testing and things to consider, alongside a case study from one of our MACS adults who is now a parent.

- Considerations
    1. Genetic testing is a personal choice, and what's right for one family may not be right for another. It's important to have open conversations with family and medical professionals to determine what works best for you.
    2. If you decide not to pursue genetic testing, remember your children may want to explore it as they grow up. Respect their choices even if they differ from yours.
    3. Genetic testing can invoke a range of emotions, such as relief and guilt. Be prepared for these emotions and speak with others about your feelings—they are valid.
    4. Obtaining results can take time, ranging from months to years, and there's a chance that no genetic cause will be found. This process can be lengthy and difficult.

- Questions to ask

1. **What are the benefits and drawbacks of genetic testing in my situation?**
   The benefits of genetic testing in your situation include gaining a clearer understanding of the underlying cause of your condition, which can help inform healthcare decisions for you and your family. It can also provide valuable information about the likelihood of passing the condition on to your children, helping you make more informed choices. On the other hand, genetic testing can have emotional drawbacks, as the process may bring up difficult feelings, especially if unexpected results are found. Additionally, testing may not always provide clear answers or could reveal information you weren't prepared to handle.
2. **How will genetic testing impact my healthcare or my family's healthcare?**
   Genetic testing can significantly impact your healthcare by providing a clearer picture of your condition, which can guide treatment decisions and management. It may also help your

doctors better understand any associated risks or complications, allowing for more tailored healthcare plans. For your family, testing can identify whether other members are at risk or if they may need further monitoring, and it can help in making decisions about future pregnancies.

3. **Can genetic testing confirm an existing diagnosis or provide a diagnosis if none exists?**
   Yes, genetic testing can confirm an existing diagnosis by identifying the specific genetic mutation responsible for the condition. If no diagnosis has been made yet, genetic testing can potentially provide one by identifying an underlying genetic cause that explains the symptoms. This can be especially helpful if there is uncertainty about the condition or if other causes have not yet been ruled out.
4. **What emotions might arise from the testing process, and how can I manage them?**
   The testing process can bring up a wide range of emotions. You might experience feelings of relief if the results provide clarity, but you could also feel guilt, fear, or anxiety, especially if the results are unexpected or reveal difficult news. It's important to acknowledge these emotions and talk to someone you trust, whether it's a family member, friend, or a counsellor. Managing these emotions can also involve giving yourself time to process the results, seeking support from genetic counsellors, and taking care of your emotional well-being throughout the journey.
5. **How long will it take to receive results, and what happens if no genetic cause is found?**
   The time it takes to receive results can vary, ranging from a few months to a year, depending on the complexity of the testing and the workload of the laboratory. If no genetic cause is found, it can be disappointing, but it's important to remember that not all conditions have a clear genetic explanation. This may mean that the cause of your condition is still unknown, and further research or tests may be needed in the future. It's essential to stay in contact with your healthcare provider for guidance on next steps.

<u>Genetic testing</u>

Many regional genetics services have their own specific processes for

appointments, which can vary depending on the reason for your referral. It is often helpful for both parents to attend these appointments. If you have been referred, you don't have to go alone—feel free to bring a family member, carer, or friend for support. During the appointment, you may meet with a genetics doctor or genetic counsellor.

You can expect to discuss your personal medical information, which may include a physical examination or eye test. The clinician will also want to talk about your family history, and they may draw a family tree with you to better understand any inherited conditions. If appropriate, a genetic test may be offered. This test may be suggested for the individual with MACS, or potentially for their parents as well. You'll have the opportunity to decide if you want to proceed with testing or if you'd prefer to wait and make a decision later. Throughout the appointment, there will be plenty of time for you to ask questions and discuss any concerns with your clinician.

It's important to note that genetics appointments are a chance to talk openly about your thoughts, feelings, and concerns regarding the condition. After your appointment, you and your clinician will make a plan for any necessary follow-up appointments. If you have any questions in the meantime, you should feel comfortable reaching out to your clinician again.

Genetic testing may be offered during these appointments. Your genetics doctor or counsellor will explain in more detail how genetic testing could be relevant to your or your family's situation and whether it's an appropriate next step. Genetic testing can confirm an existing diagnosis, or, if a diagnosis has not yet been made, it could provide one. This can be incredibly helpful in understanding how to manage the healthcare of the individual with the condition and identifying how the condition has been inherited within the family.

It's crucial to remember that the decision to undergo genetic testing is entirely up to the individual with the condition. Your clinician will support you in making this decision, and you should never feel pressured into testing if it doesn't feel right for you or your family.

Please note that regional genetics services may differ in their approach to patients, so it is worth looking into the specific services available to you. For more detailed information, visit the MACS website and search for "your genetic journey."

- What to expect

    1. Regional genetics services may have their own process, but typically you can expect to discuss your personal medical information, including a physical examination and eye test.
    2. A family history may be drawn up, and a genetic test may be offered, either to the person with MACS or to their parents.
    3. You'll have the opportunity to decide whether to proceed with genetic testing and to ask questions about the process with your clinician.
    4. The appointment may include discussions about your thoughts, feelings, and concerns, as well as a plan for follow-up appointments if needed.

- Case study

I'm an adult with MACS and was born with bilateral microphthalmia and coloboma. I also consider myself a MACS sibling, as my brother has anophthalmia and other disabilities related to our genetic condition.

Before I share our journey with genetic testing, I want to emphasise that testing is a personal choice. What works for one family might not be right for another. It's important to have open discussions with your family and medical professionals to decide what's best for you.

About 20 years ago, my family and I took part in a genetics research project funded by MACS. This included blood tests, eye exams, and questions about our medical history.

Even as a teenager, it felt important for me to understand the cause of my MACS condition. I wanted to know if I could pass it on to any children I might have. And, to be honest, I was simply curious about what caused my eyes to develop the way they did.

After about two years, we received the results. In 2007, we learned that the cause of my brother's and my MACS conditions was a genetic issue called Branchio-Oculo-Facial Syndrome. This syndrome is caused by a mutation in the TFAP2A gene, which can lead to MAC conditions, as well as other physical and developmental challenges.

While discovering this was a huge positive for me, I understand it can be difficult for parents. Knowing that a genetic mutation may have been passed down can bring up complex emotions. But I've never resented or blamed my parents. They've done an incredible job despite many challenges, and our diagnosis is not their fault. So if you choose to explore genetic testing, remember: no one is to blame.

Now, I'm a mother of three boys. Thanks to knowing my diagnosis, I was able to have pre-birth genetic testing to see if they had inherited my gene mutation. Waiting for the results was an emotional experience, but I wanted to be prepared for any outcome. This was the right choice for my husband and me, but again, it's a personal decision and the path that worked best for us.

Having been involved with genetic testing throughout my teenage and adult years, here are the key points I would share with families considering testing:

- It's your choice. Talk openly with your family, ask questions, and do what feels right for you.
- If you choose not to pursue testing, remember that your children may want to explore it as they grow. Respect their choices, even if they differ from yours.
- Genetic testing can bring up a range of emotions—from relief to guilt. Be prepared for this, and talk to someone you trust about your feelings. Whatever you feel is valid.
- Results can take time—sometimes months or even years. And there's a chance that no genetic cause will be found. This can be difficult, and the process can be long.

For me, knowing my diagnosis has been one of the most positive things MACS has given me. It helped me plan when to have children and gave me a better understanding of the potential issues related to our genetic mutation. Without this knowledge, I would have struggled much more during my pregnancies. While genetic testing can be an emotional journey, for me, the positives far outweigh the negatives, and I'm deeply grateful for the insight it has given me.