GeneLinx

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| **Full Name:** | **Jane Doe** | | |
| Date of birth: | 01.01.1900 | Referred by: | Self-referral |
| Unique ID: | P000000 | Date seen: | 01.01.2024 |
| Booking ID: | GL-000000 | Date issued: | 02.01.2024 |

# SUMMARY REPORT
## Initial Consultation - Other

***Reason for Referral:*** Predictive testing for Spinocerebellar Ataxia type 7 (SCA7). Currently at 50% risk

Dear Jane,

It was a pleasure to meet you in our video consultation on 29/10/2024. As promised, I am writing to summarise the main points of our discussion.

You were referred to our clinic due to your family history of Spinocerebellar Ataxia type 7 (SCA7).

In terms of your family history, you informed me that your birth father has SCA7, and he is found to carry 37 CAG repeats in the *ATXN7* gene. You also mentioned one of your birth father's daughters is quite severely affected with the condition and the symptoms started at a younger age, in her 20s.

### *Genetics of SCA7*

Spinocerebellar Ataxia Type 7 (SCA7) is a rare, inherited condition that primarily affects the brain and vision. It belongs to a group of disorders known as Spinocerebellar Ataxias, which are all conditions that cause progressive problems with coordination and balance. With SCA7, genetic changes in *ATXN7* gene (CAG expansion) affect the cerebellum, the part of the brain responsible for coordinating movement, as well as the retina in the eye, which can affect vision over time.

CAG repeat size in the *ATXN7* gene and its impact on SCA7

- **Normal Range**: 7-27 CAG repeats. No disease symptoms; these repeats are stable across generations.
- **Mutable Normal**: 28-33 CAG repeats. Generally no disease symptoms, but the repeats can be unstable, which means they can expand to a pathogenic (disease causing) range from one generation to the next.
- **Pathogenic (Reduced Penetrance)**: 34-36 CAG repeats. May cause symptoms, typically later in life and milder, but the repeats can be unstable and expand further.
- **Pathogenic (Full Penetrance)**: 37-460 CAG repeats. This range is associated with definite symptoms, earlier onset, and severity that progresses overtime. Usually larger repeat size is associated with more severe disease.

This gene change is inherited in an “autosomal dominant” pattern, which means that a person only needs one copy of the altered gene to develop the condition. If a parent has SCA7, each child has a 50% chance of inheriting the altered gene and developing the condition at some point in their life. It’s important to remember that while SCA7 is inherited, its symptoms can vary widely even within the

same family. We also talked about sometimes the size of the CAG repeat can expand or in a very rare case contract when being passed from one generation to the next.

## *Symptoms of SCA7*

The symptoms of SCA7 often begin with problems in coordination and balance, leading to difficulties with activities like walking, writing, and other movements. Over time, SCA7 can also affect the eyes, causing gradual vision loss, often beginning with difficulty seeing in low light. Some people may experience symptoms earlier or more severely than others, and this variation can depend on several factors, including the CAG repeat size.

## *Genetic testing for SCA7*

You had a good understanding of the condition before our discussion. Hopefully our discussion provided you with sufficient information to make an informed decision regarding testing. Through our discussion clearly you are preparing thoughtfully for any outcome. Having a good support system in place is also a real strength.

While awaiting results, it may be helpful to consider both possibilities. If the test is positive, what measures you put in place to cope with the results, including speaking with your doctor for appropriate referrals. If it's negative, this will provide you with clarity and reassurance.

As it is a predictive testing for a known gene change in the family, you understand there are two types of results that can be obtained from this genetic testing:

- **Positive**: You carry ATXN7 gene with CAG repeats in the 'Pathogenic' range. This scenario you will likely develop symptoms of the condition at some point in your lifetime. Additionally, your future offspring will be at 50% risk of inheriting the gene change from you.
- **Negative**: You carry ATXN7 gene with CAG repeats in the 'Normal' range, discussed above. This will remove the possibility of you being affected with SCA7. Additionally, the risk of you passing SCA7 to your future offspring is low.

We can go through your results and what it means depending on the repeats found, once they are back.

## *Benefits and risks of genetic testing*

We discussed that genetic testing is a highly personal choice and that you should process to testing only if you feel ready and would like to know your genetic status. As this is an important decision with implications for the future, if at any time during this process you change your mind about testing, you can notify us not to receive the results.

You mentioned that you do not have symptoms of SCA7 but felt that testing can help you:

- understand your risks and plan for the future
- to guide your family planning and reproductive choices
- to help relieve the uncertainty
- access ongoing or upcoming gene-specific clinical trials and treatments, if appropriate

After considering the benefits and risks, you decided to proceed with genetic testing. You consented to the test and have made arrangements with the laboratory to have your blood drawn. A copy of this letter and the signed consent form will be shared with the laboratory to initiate the requested test.

When the results are ready, a link to book an appointment to discuss your results will be shared with you.

If you have any additional questions please do not hesitate to reach out us at appointments@gene-linx.com.

**Electronically signed:**
Ms Genetic Counsellor, MSc
AHCS Registered Genetic Counsellor #00000