NAVIGATING ALS GENETICS

UNDERSTANDING YOUR OPTIONS



ALS SLA CANADA

Genetics and Amyotrophic Lateral Sclerosis (ALS)

Most of the time, ALS doesn't have a known genetic cause. However:

At least 15%[1] of people with ALS have a gene change linked to ALS. This is called genetic ALS.

ALS is more likely to be genetic **if other family members had ALS[2]** (also called familial ALS).

Someone can still have genetic ALS **even if no one else in their family is known to have had ALS** (also called sporadic ALS).

1 Based on current research and understanding of ALS genetics. This will continue to evolve.
2 Or related conditions, such as frontotemporal dementia (FTD). Some genes are unique to ALS, some are unique to FTD, and some genes overlap between both conditions.

What is a genetic test?



Our DNA, or genetic code, provides the instructions for our body to grow, develop, and work properly.



A gene is a small section of our genetic code. We have two copies of almost every gene — one inherited from each biological parent.

A genetic test looks for a change in a person's genetic code, also known as a variant (or mutation).



Some variants can affect how a gene works. A variant in a gene linked to ALS may increase someone's chance of having ALS.

Which best describes you?

"I don't have any symptoms of ALS, and...

"I have ALS."
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...I have a biological relative with genetic ALS and am interested in learning what this could mean for me."
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...I have one biological relative who had ALS. I don't know if it was genetic."
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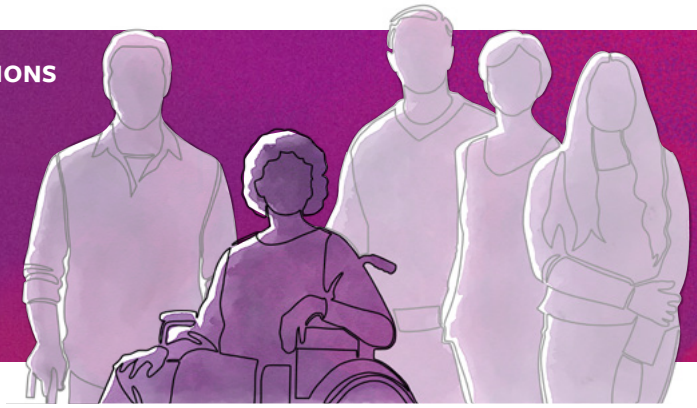
...I have multiple (2 or more) biological relatives with ALS. I don't know if it's genetic."
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Biological relative
A family member who is related to you by blood (e.g., parent, sibling, cousin).

*If none of these scenarios apply to your situation, but you still have questions, please email **genetics@als.ca**.*



"I have ALS."

Is genetic testing an option for me?

YES. Current guidelines recommend that everyone with ALS is offered genetic testing.

- It is your decision whether you would like to have genetic testing or not.
- Your ALS doctor can discuss the necessary information about genetic testing with you, so that you can make the decision that feels right for you.
- Your ALS doctor can also refer you to a genetic counsellor to have this discussion.

Why consider genetic testing?

GENETIC TESTING MIGHT:

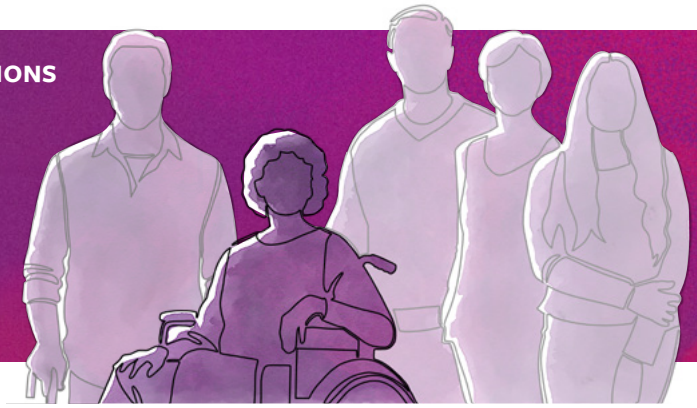
- Give you more information about what to expect for your disease over time.
- Help you better understand why you may have developed ALS.
- Allow you to take part in research or clinical trials for genetic ALS.
- Help you access treatment targeted to a specific genetic form of ALS (e.g., Qalsody [tofersen]).
- Help your family members understand their chance of developing ALS, and consider different family planning options.

Questions to ask my healthcare provider

Here are some questions you may want to consider asking your healthcare provider before deciding about genetic testing:

- "What are the **possible results** of the genetic test?"
- "What is the **likelihood** that a genetic contribution will be identified?"
- "Why should I consider genetic testing even if **no one else in my family has had ALS**?"
- "What are the potential implications of the result **for myself**?"
- "What are the potential implications of the result **for my relatives**?"

If you have questions about genetic testing, speaking with a genetic counsellor may also be helpful.



"I have ALS."

What are my next steps?

I'm interested in genetic testing

Speak to your ALS doctor about genetic testing.

- You may want to bring our clinician handout ***Navigating ALS Genetics: A Clinician's Guide to Genetic Testing in ALS*** to your doctor. **Download it here** or scan the QR code on the right to access the handout.
- They may refer you to speak to a genetic counsellor.
- Please contact **genetics@als.ca** with any questions, or if you are having difficulties accessing genetic testing.

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I have a genetic test result

Ask your ALS doctor for a genetic counselling referral if you would like more information about your genetic test result.

- They may refer you to your local genetics clinic or the ALS National Genetic Counsellor.
- You can download the referral form for the ALS National Genetic Counsellor here: **als.ca/geneticreferral** or by scanning the QR code on the right.
- Please contact **genetics@als.ca** with any questions.

als.ca/geneticreferral

$ Is genetic testing covered in my province?

While the genetic test available in every province may differ, the provincial healthcare system currently covers genetic testing for ALS in every province.

"I have a biological relative with genetic ALS and am interested in learning what this could mean for me."



Is genetic testing an option for me?

Predictive genetic testing may be an option for you.

- Predictive genetic testing is for someone who **does not have symptoms of ALS.**
- This testing may show whether you have an increased risk of developing ALS, but it **cannot tell if, when, or how symptoms will appear.**

PREDICTIVE GENETIC COUNSELLING

A predictive genetic counselling appointment is an opportunity to talk more about genetic testing and receive support as you make your decision. **You do not need to have decided about genetic testing to schedule an appointment.** Some people may not be interested in predictive genetic testing at this time, or ever.

CONSIDERATIONS

Inheriting a genetic variant with a higher risk of ALS does not guarantee a person will have ALS.

This type of testing is generally not available if you are under 18 years old.

What are my next steps?

If you are interested in having a predictive genetic counselling appointment, you can ask your family doctor (or a doctor at a walk-in clinic, if you do not have a family doctor) to provide a referral to your local genetics clinic.

- You may want to bring our clinician handout ***Navigating ALS Genetics: A Clinician's Guide to Genetic Testing in ALS*** to your doctor. **Download it here** or scan the QR code on the right to access the handout.
- There are usually wait times to get an appointment in genetics.
- If possible, having a copy of your relative's genetic test results can be helpful. You may consider contacting the ALS clinic where your relative was seen to ask whether they can share that information with you.
- Please contact **genetics@als.ca** with any questions.

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"I have one biological relative who had ALS. I don't know if it was genetic."

Is genetic testing an option for me?

Genetic testing may be an option for you, though it may not be offered in all cases.

- If only one person in a family has ALS, a genetic cause is less likely.
- Genetic testing for someone who does not have symptoms is usually more straightforward if a gene change has already been found in a family member.
- Speaking with a genetic counsellor may help provide a better understanding of your specific situation.

What are my next steps?

1 If possible, try to gather information about whether genetic testing has been done in your family, and what the results were.

- If possible, the best next step is for a living family member with ALS to have genetic testing. This would provide the most useful information for other family members.
  - They may want to bring our clinician handout ***Navigating ALS Genetics: A Clinician's Guide to Genetic Testing in ALS*** to their doctor. **Download it here** or scan the QR code on the right to access the handout.
- It can also be helpful to gather information about your family. For example, have any of your relatives had other neurological conditions (e.g., dementia, Alzheimer's disease, Parkinson's disease)?

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2 Speak with your family doctor (or a doctor at a walk-in clinic, if you do not have a family doctor).

- You can ask them to provide a referral to your local genetics clinic for a genetic counselling appointment. However, the referral may not be accepted. This will vary across health systems.
- It can be helpful to include information about relatives who may have had other neurological conditions (e.g., dementia, Alzheimer's disease, Parkinson's disease) on the referral.
- **Please contact genetics@als.ca with any questions, or if your referral to the genetics clinic was not accepted.**



"I have multiple (2 or more) biological relatives with ALS. I don't know if it's genetic."

Is genetic testing an option for me?

Genetic testing may be an option for you.

- If multiple people in a family have ALS, a genetic cause is more likely. However, up to 30% of the time, a genetic cause in the family is not identifiable at this time.
- Genetic testing for someone who does not have symptoms is usually more straightforward if a gene change has already been found in a family member.
- Genetic counselling is recommended to review your specific situation, help interpret your results, and explain the limitations of testing.

What are my next steps?

If possible, try to gather information about whether genetic testing has been done in your family, and what the results were.

- If possible, the best next step is for a living family member with ALS to have genetic testing. This would provide the most useful information for other family members.
  - They may want to bring our clinician handout ***Navigating ALS Genetics: A Clinician's Guide to Genetic Testing in ALS*** to your doctor. **Download it here** or scan the QR code on the right to access the handout.
- Try to gather as much information as you can about ALS (or frontotemporal dementia) in your family.
- You can ask your family doctor (or a doctor at a walk-in clinic, if you do not have a family doctor) to provide a referral to your local genetics clinic for a genetic counselling appointment.
- Please contact **genetics@als.ca** with any questions.

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