

INFORMATION FOR PARENTS/GUARDIANS:

My Baby Has a Positive Newborn Screening Result for Severe Combined Immune Deficiency (SCID) and related diseases

What is newborn screening?

These are routine tests done soon after birth on every baby born in Ontario. A small sample of blood is taken from your baby and is tested for rare, treatable diseases, including a disease called Severe Combined Immune Deficiency (SCID).

What does it mean if my baby has a positive SCID newborn screening result?

This result does **not** mean that your baby has SCID. It means that more testing is needed because your baby **might** have SCID. Babies with SCID can be healthier if treatment begins early, so it is important to have follow-up testing done to find out if your baby truly has SCID.

You may feel worried about your baby's screen positive result. Many parents in this situation feel this way. Remember, we do not know for sure that your baby has SCID until follow up testing has been done.

What is Severe Combined Immune Deficiency (SCID)?

Severe Combined Immune Deficiency is a disease that causes the immune system to not work well. The role of the immune system is to fight off infections. Children with SCID are at risk of getting serious infections.

How do I find out if my baby has SCID?

Blood tests are done to determine if a baby who is screen positive actually has SCID.

When can my baby have these tests?

Your baby's doctor or a health care professional at a Newborn Screening Treatment Centre will call you to talk about the results of your baby's positive newborn screen and arrange follow up testing as soon as possible.

Why screen for SCID?

There are usually no clues at birth that a baby has SCID but, without treatment, babies with SCID are prone to serious infections that can be life-threatening. Screening for SCID means that babies with SCID can be identified and treated early. Treatments help provide them with a working immune system to protect them from serious infections.

How is SCID treated?

Babies with SCID are followed by immune system specialists. They are given medications to help treat and prevent infections. Different treatments may be considered depending on the form of SCID a baby has. Sometimes, babies with SCID require a bone marrow transplant to provide them with an immune system that works properly to fight off infections.



How does a baby get SCID?

SCID is an inherited (genetic) disease. Changes (mutations) in any of the SCID genes cause the disease. Depending on which gene is changed, SCID can run in families in different ways.

Because your baby's newborn screen was positive for SCID, additional tests were done on his/her newborn screening blood sample to look for the following diseases, which cause or are associated with SCID.

- **Adenosine Deaminase (ADA) Deficiency:** ADA deficiency is a disease that harms the immune system and causes SCID.
- **Purine Nucleoside Phosphorylase (PNP) Deficiency:** PNP deficiency is a disease that harms the immune system and causes SCID. Some people with PNP deficiency can also have developmental delay and problems with their muscles and balance.
- **22q11.2 deletion syndrome (also known as velocardiofacial or Di George syndrome):** 22q11.2 deletion syndrome is a disease caused by a missing piece of chromosome 22. Children with 22q11.2 deletion syndrome can have birth defects and problems with learning and development. They can also have problems with their immune system, that can cause a positive newborn screen for SCID, but they do not have SCID.

Where can I get more information?

For more information on newborn screening, please visit the Parents section of the Newborn Screening Ontario (NSO) website at <http://www.newbornscreening.on.ca> or talk to your local health care provider.

For more information on SCID, please visit the Immunodeficiency Canada website at <http://cisociety.com/blog/> or call 416-964-3434.

NOTE TO PARENTS/GUARDIANS: This information is only for parents whose baby has had a positive newborn screening result for Severe Combined Immune Deficiency (SCID) and related diseases. Please remember that this fact sheet was written for information purposes only. The fact sheet should not replace professional medical advice, diagnosis or treatment.

