



# Making Sense of Abnormal Results



## What does an abnormal newborn blood spot screen result mean?

- If your baby has an abnormal newborn blood spot screen result they may have one of the treatable conditions and more testing is needed.
- If your baby has abnormal result, it doesn't mean that they have one of the treatable conditions. It means your baby has a higher chance of having one of the treatable conditions, and your baby needs more tests to find out for sure.
- Your doctor, midwife or a specialist will contact you to arrange more tests as soon as possible.

## What conditions does newborn blood spot screening look for?

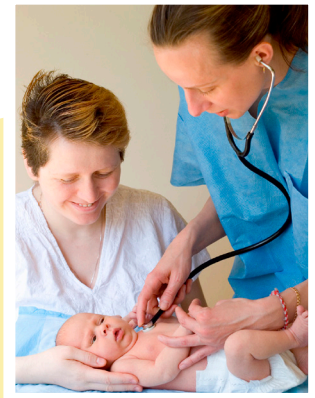
- Newborn blood spot screening looks for treatable conditions that include:
  - metabolic conditions cause problems with how the body uses food to grow and develop
  - endocrine conditions cause problems with how the body makes hormones
  - cystic fibrosis, a condition that affects the lungs and digestive system
  - sickle cell disease, a condition that affects the blood and causes damage to heart, lungs, and kidneys
  - severe combined immunodeficiency, a condition that has serious effects on the immune system

## What are metabolic conditions and how are they treated?

- In metabolic conditions, the body can't break down some substances in food like fats, proteins, or sugars. Your baby needs these things to be healthy, to grow, and to develop.
- The substances build up in the body and may cause serious health problems.
- The treatment for metabolic conditions is often special diets or nutritional supplements.

## Where can I find more information about metabolic conditions?

- Alberta's Newborn Metabolic Screening (NMS) Program looks for the following metabolic conditions:
  - visit: [www.ahs.ca/newbornscreening](http://www.ahs.ca/newbornscreening)
  - Citrullinemia (CIT)
  - Maple syrup urine disease (MSUD)
  - Phenylketonuria (PKU)
  - Glutaric acidemia type 1 (GA1)
  - 3-Hydroxy-3-methylglutaryl-CoA lyase (HMG) deficiency
  - Isovaleric acidemia (IVA)
  - Methylmalonic acidemia (MMA)
  - Propionic acidemia (PA)
  - Carnitine uptake defect (CUD)
  - Long chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency
  - Medium chain acyl-CoA dehydrogenase (MCAD) deficiency
  - Tri-functional protein (TFP) deficiency
  - Very long chain acyl-CoA dehydrogenase (VLCAD) deficiency
  - Biotinidase deficiency (BIOT)
  - Classic galactosemia (GALT)
  - Tyrosinemia type 1 (TYR1)



## Alberta's Newborn Metabolic Screening Program

Newborn blood spot screening helps your doctor or midwife find conditions that can be treated early to prevent health problems, improve your baby's health and maybe even save your baby's life.

### What is Alberta's Newborn Metabolic Screening Program?

- Alberta's Newborn Metabolic Screening (NMS) Program is about health care providers working together with parents or guardians to screen for certain treatable conditions.
- Timely screening helps find conditions that can be treated early, when the treatment can help your baby the most.
- Alberta's NMS Program makes sure all screened babies who may have one of the treatable conditions get more testing and early treatment.





## What are endocrine conditions and how are they treated?

- In endocrine conditions, the body can't make the right amount of hormones.
- Not having the right amount of hormones may cause problems with growing and developing.
- The treatment for endocrine conditions is giving hormones the body can't make itself.

## Where can I find more information about endocrine conditions?

- Alberta's NMS Program looks for the following endocrine conditions:
  - visit: [www.ahs.ca/newbornscreening](http://www.ahs.ca/newbornscreening)  
Congenital adrenal hyperplasia (CAH)  
Congenital hypothyroidism (CH)

## What is cystic fibrosis and how is it treated?

- Cystic fibrosis is a condition that causes thick, sticky mucous to build up in the lungs, digestive tract and other areas of the body.
- This causes problems with how your baby breathes and digests food.
- The treatment for cystic fibrosis is medicine and physical therapy.

## Where can I find more information about cystic fibrosis?

- Alberta's NMS Program looks for cystic fibrosis.
  - visit: [www.ahs.ca/newbornscreening](http://www.ahs.ca/newbornscreening)  
Cystic fibrosis (CF)

## What is sickle cell disease and how is it treated?

- Sickle cell disease is a condition that causes red blood cells to have a crescent moon or sickle shape.
- Red blood cells with the sickle shape get trapped in blood vessels. This causes pain, damages tissue, and causes red blood cells to break down too early.
- The treatment for sickle cell disease is antibiotics, immunizations, and care for fevers.
- Newborn blood spot screening will detect babies who carry the sickle cell trait but who don't have sickle cell disease.

## Where can I find more information about sickle cell disease?

- Alberta's NMS Program looks for sickle cell disease.
  - visit: [www.ahs.ca/newbornscreening](http://www.ahs.ca/newbornscreening)  
Sickle cell disease (SCD)

## What is severe combined immunodeficiency and how is it treated?

- Severe combined immunodeficiency causes serious problems with the immune system, so the body can't fight off germs.
- When the immune system doesn't work well, a person can get serious, life-threatening infections.
- The treatment for severe combined immunodeficiency is giving antibiotics before an infection happens and keeping the person away from any germs (isolation) to prevent infections.

## Where can I find more information about Severe combined immunodeficiency?

- Alberta's NMS Program looks for severe combined immunodeficiency.
  - visit: [www.ahs.ca/newbornscreening](http://www.ahs.ca/newbornscreening)  
Severe combined immunodeficiency (SCID)

## What happens if the final results show my baby has one of the treatable conditions?

- If your baby has one of the treatable conditions they'll get help from your doctor and probably a specialist.
- Your baby will get continued support to live a healthy life, usually with a change in diet or the use of special medicine.
- Your doctor or a specialist will talk with you and answer your questions about your baby's condition and treatment.

## What else can I do to take care of my baby?

- Screening only looks for certain treatable conditions. It doesn't check for any other health problems. So it's important that your baby sees a doctor, midwife, or public health nurse regularly.
- If you're worried about the way your baby is growing or if you think your baby isn't well, contact your doctor, midwife, public health nurse or Health Link.
- And always be sure to take good care of yourself, so you can care for your baby.

## What if I have more questions about newborn blood spot screening and Alberta's NMS Program?

- Talk to your public health nurse, doctor or midwife for more information.
- Visit: [www.ahs.ca/newbornscreening](http://www.ahs.ca/newbornscreening)
- If you need health advice or information call Health Link 24/7 by dialing 811

