# What does the testing look for?

The newborn testing looks for over 32 rare disorders that can cause health problems in babies and children.

### **Disorders screened:**

- Organic Acid Disorders
- Fatty Acid Oxidation Defects
- Amino Acid Disorders

When the body is not able to break down (metabolize) certain substances in food like fats, proteins or sugars, they can collect in the body and cause serious health problems such as those listed above.

## Other disorders that are screened for include:

- Congenital hypothyroidism
- Cystic fibrosis
- Spinal Muscular Atrophy (SMA)
- Congenital Cytomegalovirus (cCMV)

#### Want to learn more?

For more information about newborn screening in Saskatchewan, speak to your healthcare provider, or visit <a href="https://www.saskhealthauthority.ca/facilities-locations/roy-romanow-provincial-laboratory-rrpl">https://www.saskhealthauthority.ca/facilities-locations/roy-romanow-provincial-laboratory-rrpl</a>

Please note that you have the right to refuse newborn screening. If this is your wish, ask your healthcare provider for a refusal form.



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saskhealthauthority.ca





### **Newborn Screening**

A healthy start leads to a healthier life



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Healthy People, Healthy Saskatchewan

This brochure provides information about newborn screening in Saskatchewan and answers questions that you may have about the newborn screening test.

#### **Getting the best start**

To ensure that your baby gets the best start in life and stays healthy, your newborn (and every newborn in Saskatchewan) will be screened for at least 32 plus rare disorders. Although most babies with these disorders look healthy at birth, if their disorder is not detected and treated, they may be at risk of having serious health problems including: developmental disabilities, recurrent sickness and even death. Early detection is the key to effective treatment.

## Early detection leads to early treatment

Individually, these disorders are very rare. As a group, they will affect about 1 in 750 babies born each year in Saskatchewan. By testing newborns within the first days of their lives, many disorders can be treated early, reducing the chance of serious health problems later in life.

## A small test producing big benefits

The screening test is completed by pricking your baby's heel to take a small sample of blood. The blood is collected on a special paper card and sent to Roy Romanow Provincial Laboratory (RRPL) for testing. Blood samples can be taken any time between one day (24 hours) to seven days after your baby is born. The best time to collect the blood sample is when your baby is between two days (48 hours) and three days (72 hours) old. If your baby is tested before one day (24 hours) of age, your baby's healthcare provider should **repeat** the test within five days, at the baby's first check-up.

### Screening results: high risk and low risk

A screening test only shows if there is a high or low risk that your baby has a disorder.

It is important to understand that the test does not make a diagnosis of a disorder, but only identifies babies who need further testing.

Once the lab has received and analyzed your baby's blood sample, one of the following will occur:

### 1. Your baby screens negative for all the disorders

Over 99 per cent of babies tested receive a "screen negative" result.

RRPL sends a report to your hospital and/or healthcare provider which is filed in your baby's medical records.

#### 2. The lab may need another blood sample

You are contacted to arrange another blood sample to be taken as soon as possible if:

- the first sample was not taken properly
- there was not enough blood to complete the testing, or
- there was some other problem with the sample.

### 3. Your baby screens positive for one of the conditions

Less than one per cent of babies tested will receive a "screen positive" result.

A screen positive **does not mean** that your baby has a disorder, but only that further testing is needed. Your baby's healthcare provider contacts you to make arrangements for follow-up at a hospital where specialists can do further testing. If a diagnosis of a disorder is made, the specialist provides your baby with treatment and your family with counselling and advice.

RRPL sends a report to your hospital and/or health care provider which is filed in your baby's medical records.