"Almost all babies are born healthy, and only a small number of babies per year, perhaps about 1 in 800, have a hereditary (genetic) health condition.

Mostly, the disorders are inherited from their two healthy carrier parents, each carrying a single copy of an abnormal gene, as well as a healthy copy."

Contact us

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TASMANIAN HEALTH SERVICE

Tasmanian Government

Screening tests for your new baby

"helping to ensure the health of your child"

Why is screening a good idea?

- So that a number of health conditions can be identified early and treatment started.
- Early treatment can prevent serious permanent mental and physical disabilities.
- Babies with these health conditions do not show signs or symptoms of illness at birth.
- By the time these conditions become obvious, a baby's development may already be harmed.
- Screening new babies is universal and done all over the developed world.

How is screening done?

You will be offered a free screening test for your new baby, to be performed at or near 48 hours and up to 72 hours after birth. A few drops of blood from a heel-prick are collected onto a blotting filter paper card which is sent to the SA Pathology Neonatal Screening Centre, located at Women's and Children's Hospital, Adelaide.

It helps to breast feed your baby (or offer some oral sweet syrup) during the small procedure, as this will minimise any discomfort felt by baby.

Consent

You will be asked to give verbal consent before the sample is collected. If you choose not to have your baby screened, the card will still be filled out, but marked "test refused by parents" and sent to the laboratory.

What conditions does the test detect?

The screening test covers over 30 different conditions, most of which are listed in the following table.

Disorder	Problems if untreated	Management
Congenital hypothyroidism	Unable to produce hormones from the thyroid gland causing growth and mental delay	Thyroid hormone supplements
Cystic fibrosis (CF)	Thick secretions which block the lungs and pancreas gland	Physiotherapy, medications, digestive medicine, diet supplements
Congenital adrenal hyperplasia (CAH)	Deficiency of certain adrenal hormones, leading to life threatening salt deficiency, low blood glucose and blood pressure if untreated	Adrenal hormone supplements
Galactosaemia	Liver failure, mental disability, serious infection	Milk-free diet
Amino-acid disorders (eg PKU)	Physical and mental developmental delay, seizures	Modified diet, vitamin supplements
Organic acid disorders	Seizures, vomiting, failure to thrive, life-threatening decompensation	Modified diet and supplements
Fatty acid disorders	Muscle weakness, inability to manage fasting	Avoid fasting; diet supplements
Spinal muscular atrophy (SMA)	Muscle weakness, delayed motor development, swallowing difficulties, breathing difficulties, shortened lifespan	Disease modifying drug therapy, supportive physiotherapy
Severe combined immune deficiency complex (SCID)	High susceptibility to recurrent, life-threatening infections	Bone marrow transplant, therapies to prevent infection

Should the screening test raise the possibility of one of these conditions, your baby will be referred to a specialist for more exact and specialised diagnostic testing.

Where are test results sent?

In more than 99% of babies the results are perfectly normal and the parents are not notified. However, results are available to the midwife or doctor after about 2 weeks. These results are not sent to "My Health Record".

Sometimes a repeat bloodspot is needed by the laboratory for a number of reasons, usually because of a sampling problem or due to a short-term elevation in a test result. You will be contacted by your caregiver if this is required and your midwife or doctor will arrange for a re-collection.

On rare occasions though, results raise concern about one of the disorders. In this case, a specialist doctor will contact you directly to discuss this and arrange for more in-depth confirmatory testing.

What happens to the bloodspot card?

The SA Neonatal Screening Centre is bound by state regulations relating to privacy and confidentiality that require us to keep your personal information, the test results and the bloodspot filter screening cards secure.

The cards are kept permanently, and are only available in the event of special circumstances such as but not limited to:

- requested by the family
- for ethics approved health research where all personal information has been removed
- by the SA Neonatal Screening Centre to standardise new tests and methods.

DNA testing

If a bloodspot sample gives an abnormal result, sometimes a DNA test is performed to confirm the result. This testing is restricted to only that section of DNA known to be linked to the disorder, and no other testing is performed.