



## Newborn Metabolic Screening Programme (NMSP) Blood spot cards – the impact of correct sample taking

**Introduction** The NMSP screens for over 20 metabolic disorders. Metabolic disorders are rare, inherited disorders in which the pathways that produce certain proteins in a human malfunction. This can lead to an illness that is often irreversible – many of the chemicals that build up in a cell as a result of a metabolic condition can cause ill health, learning disabilities or death.

Metabolic disorders are screened for using a heel prick test. With the consent of parents/guardians, blood is collected from the baby's heel at 48 hours of age or as soon as possible thereafter (regardless of feeding status, antibiotic use or stool colour). The blood spot card is then sent to the National Testing Centre for analysis.

**Blood spot card detail** Each batch of paper meets stringent quality standards within the factory and then must meet the Centre for Disease Control (CDC) standards in Atlanta (independent testing) before it is used in blood spot cards. Each card has a paper lot number and a reference number which enables tracing back to the factory and production run if necessary.

**Sample taking** To achieve an accurate result, it is essential the sample is taken correctly. The collection paper has unique properties which lead to a quantitative result for each test. It is made to a very high standard of thickness and absorbitivity and will always contain the same amount of blood when the sample is collected accurately. Too much or too little blood will affect the screening result.

**Posting of blood spot card** When the blood spot card is dry (about 2 hours), the card should be posted to the National Testing Centre. The posting of biological materials requires triple containment. The first containment is the paper fibres, the second is the card wrap and the third is the mailing envelope.

**Sample testing** Once the blood spot card is received at the National Testing Centre, it is registered in the laboratory system and testing takes place. The testing process includes quality measures such as control and calibrator samples included in each sample batch. After testing, cards are held indefinitely in secure storage. Blood spots may be returned to parents/guardians/individuals at their request.

**Ordering of cards** Blood spot cards and parent pamphlets can be ordered from the National Testing Centre by phone: 09 3074949 ext. 6759, fax 09 307 4936 or email [ntc@adhb.govt.nz](mailto:ntc@adhb.govt.nz). For further information on the programme see [www.nsu.govt.nz](http://www.nsu.govt.nz).





**Metabolic disorders** are rare, inherited conditions in which the pathways that produce certain proteins in a human malfunction. These pathways are like assembly lines in a human cell and a blockage at any point along the assembly line can lead to a build-up of toxic chemicals in the cell or lack of an important protein or enzyme in the body.

**Screening** is when healthy children and adults are tested to see if they are likely to develop a condition. Screening tests don't generally confirm that a person has a disease - they are not diagnostic tests. Usually a person will not feel ill from these conditions in any way at the time when they're screened. Screening allows diseases to be identified early, before any signs of illness. This means people can be treated quickly with the aim of avoiding serious illness.

**Genes, proteins and enzymes** A gene is a section of DNA that gives a specific instruction to the cell. Most of the time, the instruction is a recipe for making a protein. Proteins are large molecules that are critical for most life processes. Some help form the structure of cells, while others (enzymes) assist with the many chemical reactions the body needs to perform every day. There are thousands of different proteins and each one has an important function. Proteins are made up of building blocks called amino acids.

**Autosomal recessive** When both parents are carriers of an autosomal recessive trait, there is a 25% chance of a child inheriting abnormal genes from both parents, and therefore of developing the disease. There is a 50% chance of each child inheriting one abnormal gene (being a carrier).

In other words, if it is assumed that 4 children are produced, and both parents are carriers (neither exhibits any disease), the STATISTICAL expectation is for:

- 1 child with 2 normal chromosomes (normal)
- 2 children with 1 normal and 1 abnormal chromosome (carriers, without disease)
- 1 child with 2 abnormal chromosomes (has the disease)

This does not mean that children WILL necessarily be affected. It does mean that EACH child has a one in four chance of inheriting the disorder and a 50:50 chance of being a carrier.

## **Contact information**

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