

Newborn Screening



*Screen Today
for a Safer Tomorrow*

What is "Newborn Screening"?

With Newborn Screening we aim to identify babies who are at risk to develop certain rare but serious medical conditions. Newborn Screening may identify harmful or potentially fatal conditions that are not otherwise apparent at birth. The early detection and treatment of these conditions could improve health and prevent severe disability or even death.

Screening is not the same as diagnosis.

Screening is only the first step to identify babies with the highest risk of having these harmful conditions. These babies will require further tests before a diagnosis can be confirmed.

What are these disorders?

Many of the conditions screened for are metabolic disorders – also called "inborn errors of metabolism" – which interfere with the body's normal use of nutrients to maintain healthy tissues and produce energy. Other disorders that may be detected through screening include genetic and hormonal disorders.

More than 20 specific disorders are screened for – including disorders of the Amino acid-, Organic acid-, Fat- and Urea cycle metabolism pathways; as well as Cystic Fibrosis, Galactosaemia, Phenylketonuria, Congenital Adrenal Hyperplasia and Congenital Hypothyroidism.

Why does my baby need Newborn Screening tests?

Screening is designed to identify babies with the highest risk of certain disorders.

Screening aims to identify babies with these disorders before they develop symptoms to ensure that treatment commences as soon as possible.

Early treatment can improve the health of babies with these rare disorders, e.g. for some, severe mental disability can be prevented.

While screening can be an apprehensive process for families, it prevents the later anxiety associated with uncertainty over severe symptoms before a clinical diagnosis is made and treatment commences.

How will my baby be tested?

Within 48 to 72 hours after birth, a specially trained laboratory phlebotomist will take a few drops of blood from your baby's heel – the so called "heel-prick test".

How will I get the results of the test?

The results of the Newborn Screening test will be delivered to your Paediatrician / General Practitioner who will notify you of the results. If the Newborn Screening test is positive, further testing for confirmation will be required in consultation with our expert panel of Specialists.

Why do some babies need to be re-tested?

A second blood test is sometimes needed, and it is usually done because the first test gave an equivocal or positive result. This second analysis will be free of charge.

If you are asked for a second test, please get it done immediately.

Important note: The Newborn Screening Test should only be done 48 hours AFTER birth but BEFORE the baby is 7 days old.

The Newborn Screening Test can be done at any of our laboratories.

TO FIND YOUR NEAREST NEWBORN SCREENING LABORATORY:

BLOEMFONTEIN
AMPATH
Tel: 051 4000 700

CAPE TOWN
AMPATH
Tel: 021 596 3120

DURBAN
Dr Bouwer & Partners Inc.
Tel: 031 327 7500

JOHANNESBURG
Drs Du Buisson, Bruinette & Kramer Inc.
Tel: 011 797 3600

EAST LONDON
Drs Du Buisson, Bruinette & Kramer Inc.
Tel: 043 743 4313

PRETORIA
Drs Du Buisson, Bruinette & Kramer Inc.
Tel: 012 427 1800

PORT ELIZABETH
Dr's Swart & Maré
Tel: 041 363 2339

Pasgebore Baba Siftingstoets



**Toets Vandag
vir 'n Veiliger Môre**

Wat is 'n "Pasgebore Baba Siftingstoets"?

Met die Pasgebore Baba Siftingstoets poog ons om die babas te identifiseer wat 'n risiko het om sekere seldsame, maar ernstige, siektetoestande te ontwikkel. Die Pasgebore Baba Siftingstoets kan skadelike of selfs potensiëel dodelike toestande opspoor wat andersins nie duidelik sigbaar is nie in oënskynlik gesonde pasgebore babas. Die vroeë opspoor en behandeling van hierdie siektetoestande kan die gesondheid van die babas verbeter en erge gebreke of selfs dood, voorkom.

"Sifting" is nie dieselfde as "Diagnose" nie.

'n Siftingstoets is slegs die eerste stap in die identifisering van babas met 'n hoë risiko vir hierdie ernstige toestande. Indien 'n siftingstoets positief is, moet verdere toetse gedoen word om die diagnose te bevestig.

Wat is hierdie siektetoestande?

Verskeie van die toestande waarvoor gesif word is metaboliese siektes – ook genoem: "aangebore afwykings van metabolisme" – wat inmeng met die liggaam se normale gebruik van voedingstowwe om weefsel gesond te hou en energie te produseer.

Ander toestande wat deur die siftingstoetse opgespoor kan word, sluit in genetiese en hormonale afwykings.

Meer as 20 spesifieke siektetoestande word met die siftingstoetse opgespoor – insluitend afwykings van die Aminosaar-, Organiese suur-, Vet- en Ureum siklus metaboliese paaie; sowel as Sistiese Fibrose, Galaktosemie, Feniëlketonurie, Kongenitale Bynier Hiperplasie en Kongenitale Hipotireose.

Hoekom het my baba die Siftingstoets nodig?

Die siftingstoetse is ontwikkel om babas met die hoogste risiko vir sekere toestande op te spoor.

Die doel van die siftingstoetse is om babas met hierdie siektes te identifiseer voordat hulle simptome ontwikkel om sodoende te verseker dat behandeling so gou as moontlik kan begin.

Vroeë behandeling kan die gesondheid van babas met hierdie seldsame siektes verbeter, bv. by sommige babas kan erge verstandelike inkorting voorkom word.

Die siftingsprosedure kan 'n spanningsvolle proses vir families wees, maar dit voorkom die latere angstige geassosieer met die onsekerheid oor ernstige simptome voor 'n kliniese diagnose gemaak word en behandeling begin.

Hoe sal my baba getoets word?

Binne 48 tot 72 uur na geboorte sal 'n spesiaal opgeleide verpleegkundige of flebotomis van die laboratorium 'n paar druppels bloed van jou baba se hakskeen neem – die sogenaamde "hak-prik toets".

Hoe sal ek die resultate van die toets ontvang?

Die resultate van die Pasgebore Baba Siftingstoets sal by jou Pediater / Algemene Praktisyn afgelewer word wat jou van die resultate in kennis sal stel.

Indien die Pasgebore Baba Siftingstoets positief is, sal verdere toetse in konsultasie met ons paneel van kundige Spesialiste uitgevoer word.

Waarom moet sommige babas weer getoets word?

'n Tweede bloedtoets is somtyds nodig. Die toets word herhaal indien die eerste toets 'n twyfelagtige of positiewe resultaat gegee het. Hierdie tweede analise sal gratis gedoen word. Indien die toets herhaal moet word – laat dit asseblief onmiddellik doen.

Belangrike nota: Hierdie "Pasgebore Baba Siftingstoets" moet slegs gedoen word 48 uur NA geboorte maar VOORDAT die baba 7 dae oud is.

Die "Pasgebore Baba Siftingstoets" kan by enige van ons laboratoriums gedoen word.

OM JOU NAASTE "PASGEBORE BABA SIFTING" LABORATORIUM TE VIND:

BLOEMFONTEIN
AMPATH
Tel: 051 4000 700

KAAPSTAD
AMPATH
Tel: 021 596 3120

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