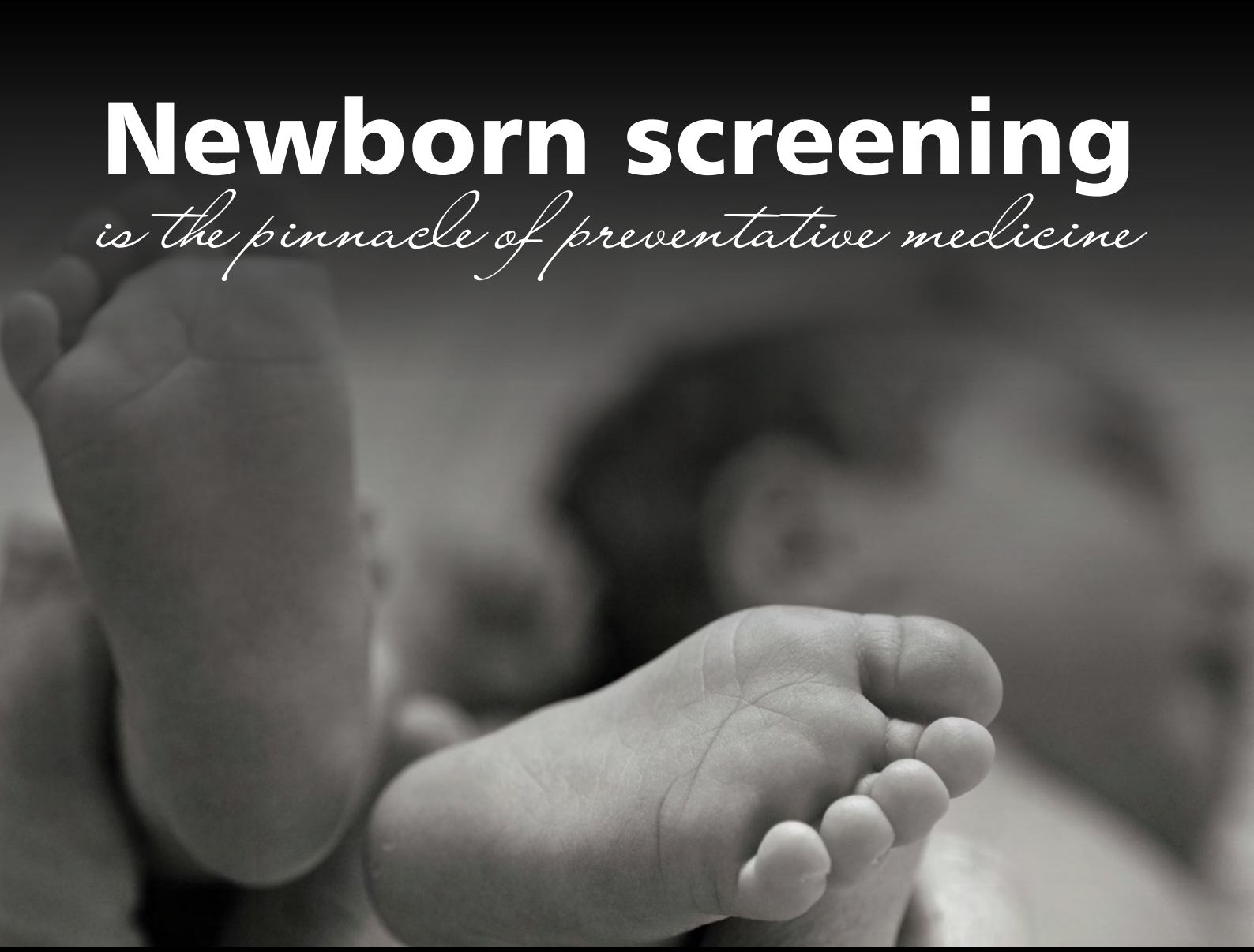
Newborn screening

is the pinnacle of preventative medicine



A pin prick on the heel of a newborn baby 48 to 72 hours after birth can be life changing.

ANALYSIS OF A FEW DROPS of blood can make all the difference between healthy thriving and debilitating ill health caused by an inherited metabolic disorder that might otherwise go undetected until it is too late.

"Newborn screening is probably the pinnacle of preventative medicine, and international practice recommends testing for all newborns," says Dr Chris Vorster, head of the Centre for Human Metabonomics at the Potchefstroom Campus.

This is so even in developing countries such as Thailand and Brazil, where an estimated 97% and 80% of newborns, respectively, are screened shortly after birth.

The situation is starkly different in South Africa: in 2012 fewer than 1% of newborns were screened. "Newborn screening is not a health priority in South Africa and no legislation or even recommendations exist for testing," says Dr Vorster, a chemical pathologist by profession.

South Africans are not immune

The main reason why newborn screening is not a priority is that inherited metabolic diseases – which impair the body's ability to process certain nutrients – are perceived as being extremely rare in South Africa. Hence, the thinking is that newborn screening would divert funds from more pressing health priorities.

"There is little evidence to suggest that South Africa is spared from inherited metabolic diseases," says Dr Vorster. "Based on the experience obtained from our own screening programme, it appears that some of these disorders are quite common."

Examples are isovaleric acidemia, propionic acidemia and glutaric acidemia type 1, which impair the body's ability to process certain proteins. This can be lethal. It causes the build-up of harmful substances in the blood and urine, which can lead to mental retardation and early death.

The sheer number of inherited metabolic defects that can be passed on by parents unaware that they are carriers is daunting. To date, as many as 2 000 different defects have been identified. Taking into account only the more commonly occurring defects, the combined incidence is between 1 : 1 000 and 1: 3 000.

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Filling the vacuum

In the absence of a government-run screening programme in South Africa, the Centre for Human Metabonomics has stepped into the breach. Its Potchefstroom Laboratory for Inborn Errors of Metabolism (PLIEM) has been operating since 1983 under the leadership of its founder, the legendary Prof Japie Mienie.

In the past 30 years, PLIEM has tested more than 50 000 babies, toddlers, teens, adults and elderly people referred to it by public and private hospitals for suspected metabolic diseases.

Just over 1% of all referred patients have been diagnosed with inherited metabolic disorders. Of these, about half have been treated effectively.

"The earlier the diagnosis, the better the chances of successful treatment," says Dr Vorster.

This, of course, is where newborn screening comes in.

In 2000, PLIEM helped establish the Newborn Screening Laboratory, which is a dedicated facility that tests for selected, mostly treatable inherited metabolic defects in babies within seven days of birth.

"Babies born with metabolic disorders often look normal at birth," says Dr Vorster. "While most affected infants show no obvious signs at first, the onset of the disease may be so rapid, or so insidious, that irreversible damage has often set in when the diagnosis is finally established."

Although inherited metabolic disorders cannot be cured, they can be treated, some more effectively than others. In most cases, treatment consists of dietary modifications and dietary supplementation. Some patients also require medication. In many instances, an early treated baby will grow up to live a normal, healthy life.

It all starts with a quick, relatively painless prick on the heel of the newborn. RD

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Helping parents CARRY THE LOAD

As most medical aids still do not cover testing for inherited metabolic disorders, parents of babies born with defects often face massive medical and other expenses. The Centre for Human Metabonomics does what it can to assist them.

An example is the free follow-up testing for treatable metabolic defects. This entails providing the initial diagnostic service at a market-related price but, once a child is diagnosed with a metabolic defect, all subsequent tests are conducted at no cost. The follow-up test is needed when metabolic crisis occurs, as well as to evaluate the therapeutic management of the disorder, which is unique for each patient.

In addition, the PLIEM laboratory may facilitate in prenatal testing and testing of siblings of the index patient. The economic implication of prenatal and sibling testing depends on the family structure and financial abilities of the parents. Each case is unique and limited costs will be applied to ultimately support families in uncertain times.

PLIEM also collaborates with various metabolic and genetic laboratories in Europe and America to provide additional specialised diagnostic tests at no or minimal costs (depending on the selection of tests for a specific disorder).

While providing a vital, one-of-a-kind testing service in South Africa, the laboratory also – with the informed consent of parents – uses residual blood samples for further research on inherited metabolic disorders. The greater the knowledge base about these disorders in South Africa, the greater the prospect of assisting generations not yet born. RD