All it takes is a pinprick

... and one very determined doctor

By Jillian Green
Health and Science Reporter

They look normal but within weeks, months or years, they collapse or go into a coma – resulting in brain damage or even death.

Yet simple blood tests can prevent this tragedy which leaves parents heartbroken and doctors puzzled.

These are babies born with errors of metabolism – a condition which is rarely diagnosed at birth.

For years, babies born in South Africa have not been tested for metabolic disorders. However, that is all changing thanks to a determined doctor at Johannesburg Hospital.

Dr Jennifer Cartwright of the Department of Paediatrics at the University of the Witwatersrand and Johannesburg Hospital, initiated the drive to introduce Tandem Mass Spectrometry (MS/MS) testing in SA.

"Many children came through my doors with unexplained brain damage. As a pediatrician, I found this frustrating and alarming. When I investigated possible causes, I discovered that a frequently missed explanation for this mental retardation was an inborn error of metabolism," she says.

And although the incidence of such disorders in the population is relatively low (1 in 1000), Cartwright believes screening is vital in that long term damage is preventable.

Newborn screening, testing infants for specific disorders such as genetic anomalies and inborn errors of metabolism, has received a boost through the acquisition of laboratory equipment dedicated to carrying out MS/MS testing, by the University of Potchefstroom

MS/MS testing will enable systematic testing of babies across all ethnic groups.

The test is a fast, accurate, reliable, inexpensive and easy to initiate programme that can test for more than 30 metabolic disorders.

It uses a single pinprick blood sample taken from a newborn baby's heel within three days of birth.

Planning to save many little lives ... Dr Jennifer Cartwright

MEDICAL TERMS

■ **METABOLIC DISORDER:** an inherited genetic condition in which the body is unable to produce a specific enzyme. People with metabolic disorders cannot properly break down the building blocks of protein, sugar and fat in food. The cells in the body are not able to turn these building blocks into the fuel that the body needs to run efficiently.

■ **TANDEM MASS SPECTROMETRY (MS/MS) TESTING:** the process involves taking a pinprick of blood from the baby's heel and placing it on a small filter paper. The blood sample is then analysed for a lack of, or excess of, the by-products of metabolism, resulting from the absence of the enzyme required in the metabolic process.

Once detected, most disorders can be treated through diet or medication, saving the child and a lifetime of unnecessary pain and disability.

"While most mothers will receive a normal result for their child, the cost to families, communities and government charged with raising, housing and caring for sickly, brain-damaged individuals runs into millions," Cartwright says.

She adds: "Although you can't cure these disorders, you can treat them with diet and medicine and stop them from having any long-term impact on the child's development."

Under the guidance of Cartwright and two other colleagues, a pilot programme was initiated to establish the incidence of metabolic disorders in infants.

The initial sites for testing were Johannesburg Hospital and nine hospitals in the Limpopo Province.

Results indicate that the incidence of metabolic disorders in SA is much the same as that in other countries throughout the world – about 1 in every 1 000 births – although the types of common disorders vary between countries, cultures and ethnic groups.

In SA, Galactosemia, a condition where a child has milk intolerance (mothers and cows) and sugar, seems most common among black children.

If the disease is not recognised and treated, the child develops cataracts, a huge liver and can become mentally retarded.

Cartwright says simply changing the child's diet to soya milk can prevent these complications.

Cartwright is looking to expand the programme and also make the tests available to parents using private healthcare facilities.

The National Pathology Group (NPG), a professional association representing the majority of private laboratories, has added its weight to the drive to make testing universal by offering its members' services to collect and transport blood specimens.