

# Tandem Mass Spectrometry Enables Fast, Accurate and Reliable Screening of Newborns for Metabolic Disorders

Newborn screening - testing infants for specific disorders called inborn errors of metabolism - has received a boost through the acquisition by the University of Potchefstroom of laboratory equipment dedicated to carrying out tandem mass spectrometry (MS/MS) testing.

Screening programmes are already in place, but their reach will expand significantly in both the public and private sectors from June, due to increased capacity at the university and the support of the National Pathology Group (NPG), which has agreed to collect and transport blood specimens within the private sector for a nominal fee.

Approximately 1 in every 1000 babies born suffers from an inborn error of metabolism. Although this incidence is low in comparison to other conditions, the consequences are tragic and include retarded physical and mental development, brain damage, and the need for lifelong care by a family member or institution if not identified in time.

The implementation of universal screening could reduce nearly all the complications and fatalities resulting from metabolic disorders as most are treatable through a combination of diet and medication. This would enable the patient to live a normal, healthy and productive life. However, early diagnosis is the key to successful treatment.

MS/MS is fast, accurate, reliable, inexpensive and easy

to initiate. It can easily detect more than 30 disorders from one drop of blood. The technique was developed by scientists at Duke University in the United States of America, who adapted standard mass spectrometry technology to newborn screening.

The process involves taking a spot of blood from the baby within the first few days of birth, typically obtained by pricking the heel, and placing it on a small filter paper card. 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 that metabolic process.



Dr Jennifer Cartwright, Department of Paediatrics, University of the Witwatersrand and Johannesburg Hospital, initiated the drive to introduce MS/MS testing in South Africa. "Many children came through my doors with unexplained brain damage. As a paediatrician, 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 said.

In cooperation with Mr D Knoll (University of Potchefstroom) and Prof P Venter (University of the North), Dr Cartwright initiated a pilot programme to establish the incidence of metabolic disorders in South African infants. The initial sites for testing were the Johannesburg Hospital and nine hospitals in the Limpopo Province. Results indicate that the incidence of metabolic disorders in South Africa is much the same as that in other countries throughout the world - about 1 in every 1000 births - although the types of disorders vary between countries, cultures and ethnic groups.

The project was supported by the three universities of the Witwatersrand, Potchefstroom and the University of the North, and also received funding from the International Atomic Energy Agency (IAEA) in Vienna, which provides the kits, needles, cards and training required to get the programme off the ground. However, the hospitals and universities have had to fund the salaries of the nurses required to administer the programme.

Dr Cartwright believes that there is a perception amongst healthcare professionals that testing is expensive and pointless - that babies born with these disorders will die anyway. Both assumptions are false.

She is looking to expand the newborn screening programme within the public sector and also to make the tests available to parents in the private sector. Laboratories under the umbrella of the National Pathology Group (NPG) have supported this objective by agreeing to collect and transport blood specimens for a minimal fee. This has enabled the University of Potchefstroom to provide the test to parents in the private sector at a charge of only R140.

Dr Tony Harrison, chairman of the NPG, said that the association was very pleased to be able to support the newborn screening initiative.

"Pathology services, of which newborn screening is one example, are at the cutting edge of medical science. The initiation of tandem mass spectrometry testing in South Africa is yet another indication of the pathology profession's commitment to ensuring that the best science the world had to offer is available locally, despite the challenges of operating in a developing economy, and in a sector where all equipment and materials have to be imported at great expense," he said.