



INBORN ERRORS OF METABOLISM: HEARTACHE-SAVING TEST TO GO NATIONAL

Relatively cheap early testing of infants for inborn errors of metabolism, a condition of which the consequences can be tragic and traumatic for families, has become nationally available in terms of a university agreement with the National Pathology Group (NPG).

The University of Potchefstroom acquired laboratory equipment for carrying out Tandem Mass Spectrometry testing earlier this year and the NPG has now agreed to collect and transport blood specimens, significantly increasing the reach of screening programmes.

With early diagnosis crucial to successful treatment (diet and medication), the implementation of universal screening could reduce nearly all the complications and fatalities resulting from metabolic disorders.

According to the NPG the testing is fast, accurate, reliable, inexpensive (R140 per test) and easy to initiate. It can detect more than 30 of the 200 identified metabolic disorders.

Although relatively rare (a pilot programme to establish the South

African incidence revealed it to match the 1/1 000 births global norm), the disorder is inherited in an autosomal recessive manner, giving it a 25% chance of recurring in subsequent pregnancies. This makes it advisable to test an older child for metabolic disorders even if it is too late to help that particular child.

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According to the leading force behind the testing innovation, Dr Jennifer Cartwright, of the Department of Paediatrics, University of the Witwatersrand and Johannesburg General Hospital, many doctors believe testing to be 'expensive and pointless' and that babies born with these disorders will die anyway.

She led the charge to secure the equipment and co-operation of the NPG after witnessing many children, 'come through my doors with unexplained brain damage'.

'As a paediatrician I found this frustrating and alarming and when I investigated possible causes, I found that a frequently missed explanation was an inborn error of metabolism.'

She is aiming to expand the newborn screening programme in the public and private sectors with NPG laboratories having already agreed to collect and transport blood specimens for a minimal fee.

Scientists at Duke University adapted standard Tandem Mass Spectrometry to newborn screening, taking a single spot of blood from the heel of a baby and placing it on filter paper. The sample is analysed for a lack or excess of the by-products of metabolism resulting from the absence of the enzyme required in the metabolic process.

Chris Bateman

HEALTH CARERS CRACKING UNDER HIV/AIDS WORKLOAD

The biggest survey yet on the impact of the HIV/AIDS epidemic on public health workers in this country shows 16.3% of them to be HIV-positive and nearly half to be 'exhausted and stressed' from a four-fold increase in AIDS patients over the past five years.

At least 6 000 health workers could be dying every year from AIDS-related illnesses, if projections based on death notifications compiled by Statistics SA between 1997 and 2000 are accurate.

Prescribed precautions to lower the risk of blood-borne diseases are being flouted and only 43% of public hospital managers and just 7% of private hospital managers have seen the national AIDS plan.

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These are the major and alarming preliminary findings of a survey of a cluster of 222 mainly metropolitan health facilities across all sectors conducted by a Human Sciences Research Council team and led by Professor Olive Shisana last year. Shisana is the HSRC's Executive Director, Social Aspects of HIV/AIDS and Public Health.

Just under 2 000 health care workers and just over 2 000 patients in private and public facilities were interviewed.