

Newborn Screening (Pilot Study) in South Africa: Four years of RAI and Tandem Mass Spectrometry

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Testing of newborns for Congenital Hypothyroidism and inborn errors of metabolism started in 1999 at Johannesburg Hospital, by means of the heel prick dried blood spot. The data is stored at a centralised database at the Potchefstroom University Biochemistry Laboratory. Collection of samples from the rural areas (Polokwane) was started in 2001. Testing for galactosemia is under evaluation since beginning of 2003. All results were sent to the second author as well as to the paediatrician in charge at the relevant hospital. Gradual expansion to the other 7 provinces is planned. Education of health professionals and the public about NBS was considered vital, so an intensive teaching program was commenced. Results: Of the 55 741 infants who were tested were tested for TSH, by RIA (ELSA-TSH-NN). Four cases of Congenital Hypothyroidism, were found. 9 cases of the other disorders were found in 12,000 samples giving an incidence of 1:1333, which is comparable to world experience. Two cases died before they could be treated. Approximately 5000 samples were tested for galactosemia, one case of galactose-1-phosphate uridyl transferase deficiency was found. Conclusions: Inborn errors of metabolism are present in South Africa, and should be identified and treated. Finding these cases and follow up in remote rural areas is possible. Cost effectiveness studies need to be done, as the National Health Department has many other commitments and remains to be convinced. The spectrum of disorders may be different, as CH is very rare, and disorders of fatty acid metabolism and galactosemia more common.