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International Atomic Energy Agency

Atoms for Peace

September 2008

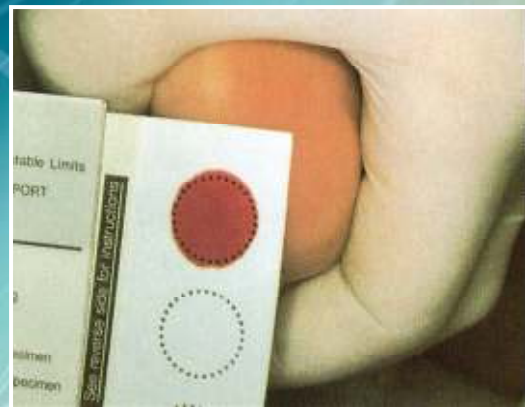
Screening for congenital abnormalities in newborns, Uruguay

The challenge...

In Uruguay, the second principal cause of death in children under one year of age is congenital abnormality. Failure to diagnose and treat some congenital diseases can cause irreversible, lifelong damage, such as mental retardation in the case of neonatal hypothyroidism, or repeated severe respiratory infections and impaired digestive function in the case of cystic fibrosis. Many developing countries are initiating newborn screening programmes for these conditions, but funding problems slow their progress. Startup finances and technical information are the principal obstacles.

The project...

The programme for neonatal hypothyroidism at the Nuclear Medicine Centre at the University of the Republic, Montevideo started in 1990 under the ARCAL project RLA/6/016. It has been continued in another three projects, which extended the screening programme to all newborns and supported the implementation of two diagnostic centres in the northern and eastern parts of the country with equipment and training.



Blood from the umbilical cord or heel stick is transferred to a specimen collection form.

The impact...

- All newborns in Uruguay now receive screening for hypothyroidism through the cord blood screening programme. The Presidential Decree that made the testing mandatory and the policy of sharing existing resources (the personnel and logistics of the Immunization Programme and the Social Security Bank) have been instrumental in the programme's success.
- Babies that have been detected as positive are usually able to receive treatment within a few days, or within 3–4 weeks at the latest, which will virtually eliminate mental retardation resulting from congenital hypothyroidism. A long term follow-up national programme to monitor treatment compliance and assess intellectual development has been organized since 2001.
- Once a programme for hypothyroid screening is organized, the expansion to other disorders is granted. Expansion to other life threatening conditions (phenylketonuria and congenital adrenal hyperplasia) has been developed. Two more projects were developed to give further assistance in the detection of cystic fibrosis in newborn infants and provide diagnostic confirmation and also to establish a national screening capability for congenital adrenal hyperplasia.

URU/6/022: *Early Diagnosis of Congenital Diseases in Children*; URU/6/023: *Screening Neonates for Cystic Fibrosis using RIA*; URU/6/024: *Screening and Diagnosis for Congenital Adrenal Hyperplasia*