

EARLY DIAGNOSIS OF CONGENITAL DISEASES IN CHILDREN (URU/6/022) E1 New

CORE FINANCING

YEAR	Experts		Equipment	Fellowships		Scientific Visits		Training	Sub-contracts	Misc. Comp.	Total US \$
	m/d	US \$	US \$	m/d	US \$	m/d	US \$	US \$	US \$	US \$	
1995	1/21	19,380	84,000	3/0	9,900	1/0	12,600	-	-	-	125,880
1996	0/21	8,400	20,000	-	-	-	-	-	-	-	28,400

First Year Approved: 95

OBJECTIVES: To extend the screening programme for the detection of neonatal hypothyroidism to the whole infant population; to introduce polymerase the chain reaction technique for the diagnosis of cystic fibrosis.

BACKGROUND: In Uruguay, the second principal cause of death in children under one year of age is currently congenital abnormalities (19.4% of infantile deaths). Congenital diseases not diagnosed in time can cause irreversible damage for the entire life of the child. Congenital diseases for which treatment is available nowadays, and treatment of which would result in clear social and economic benefits, include neonatal hypothyroidism and cystic fibrosis. If left untreated they result in irreversible and severe mental retardation in the case of neonatal hypothyroidism or repeated and severe respiratory infections and disorders of the digestive function in the case of cystic fibrosis. For the diagnosis of neonatal hypothyroidism, radioimmunoassay (RIA) techniques of blood samples are commonly used and detection of cystic fibrosis is achieved through the polymerase chain reaction (PCR) technique and phosphorus-32 labelled DNA probes. Uruguay has two ongoing programmes for neonatal hypothyroidism: one at the Nuclear Medicine Centre of the Clinical Hospital of the University of the Republic and the other at the Social Security Bank (BPS). Both activities are linked as part of a national screening programme. The programme of the University started in 1990, under the ARCAL project RLA/6/016, to produce RIA reagents of thyroid hormones (TSH) in blood

samples. The programme of the BPS started in 1992. By early 1994, 21,500 new born babies from Montevideo and other parts of the country had been tested at both centres. The present project will extend the screening programme to all newborns. The Nuclear Medicine Centre plans to establish two other diagnostic centres, one in the north, in the Department of El Salto, and one in the east, near Montevideo, in the Department of Maldonado. The Division of Epidemiology of the Ministry of Public Health, the National Vaccination Programme, the Faculty of Medicine and the Nuclear Research Centre will collaborate in the programme. The Government has made screening for neonatal hypothyroidism mandatory. The Cytogenetics Laboratory of the Institute for Biological Research (IBCE) has been conducting studies on cystic fibrosis but needs to establish the PCR technique in order to improve diagnostic capabilities to identify individuals affected and to determine the incidence of the disease.

PROJECT PLAN: For neonatal hypothyroidism, during the first year of the project equipment and reagents will be provided to implement a diagnostic centre in the northern part of the country and to set up routine sample analysis. A national training course will be carried out at the laboratory in Montevideo with participants from the hospitals of Salto and Maldonado as well as from Montevideo to establish RIA methodology and to discuss the organization of the national screening programme. A second diagnostic centre will be established in 1996. Co-ordination among the institutions involved in the national screening programme will receive special attention. For cystic fibrosis, equipment and reagents will be supplied during the first year, as well as expert services to establish the diagnostic techniques at the Cytogenetics Laboratory and to train the staff. During the second year, activities will focus on extension of the national screening programme for neonates and to establish a screening programme for adult carriers.

NATIONAL COMMITMENT: Staff, RIA facilities and operating funds at the Nuclear Medicine Centre; infrastructure and staff for the establishment of centres in the Departments of El Salto and Maldonado; laboratory facilities for cell culture, microscopy and molecular biology as well as staff and operational support at the Cytogenetics Division of the IIBC.

AGENCY INPUT: Expert services in organizing the national screening programme, establishing quality control mechanisms and implementing and optimizing techniques for the detection of cystic fibrosis; basic equipment for RIA for the new diagnostic centres and for PCR techniques; training.

IMPACT: The project will significantly improve public health, in particular paediatrics. A national screening programme for neonatal hypothyroidism and timely treatment will prevent mental retardation cases. Early diagnosis of cystic fibrosis will permit the establishment of control and prophylactic measures to prevent respiratory and digestive complications.