TC Project Screening Checklist – Early Planning Stage
Early diagnosis of congenital diseases in children
Uruguay – URU/6/022

What is the problem/need/opportunity?

Congenital abnormalities caused by diseases such as hypothyroidism and cystic fibrosis are the second most frequent cause of death for children under the age of one in Uruguay (19.4% of infantile deaths). Neonatal screening for hypothyroidism is established in the Montevideo area which accounts for about 40% of births. This capability needs to be extended to include the whole of the neonatal population. Screening for cystic fibrosis using radionuclide-based DNA techniques needs to be established.

What priority does the institute/Government assign to this problem/need/opportunity, and what information is this based on?

Government priority is high as it has decreed neonatal screening for hypothyroidism mandatory. The extension of hypothyroidism screening to areas beyond Montevideo is to be carried out under an agreement including the Ministry of Public Health, the Nuclear Medicine Centre and the Faculty of Medicine. A number of institutes are collaborating to implement a Multidisciplinary Study for the Diagnosis and Prevention of Congenital Anomalies in Pediatrics.

Ongoing programmes exist at the Nuclear Medicine Centre of the Clinical Hospital of the University of the Republic and the Social Security Rank. Significant progress has been made in the introduction of RIA through an ARCAL project. Preliminary work on establishing the incidence of cystic fibrosis has been carried out by the Cytogenics Lab of the Institute for Biological Research of the Ministry of Education and Culture.

What part of the problem/need/opportunity would the proposed project address?

- *The extension of screening for neonatal hypothyroidism to regions beyond Montevideo – the centres of Salto and Maldonado being targeted for establishment of diagnostic capabilities.*
- *The establishment of radionuclide-based DNA techniques at the Cytogenics Lab.*

What would be the impact of the project itself?

- *All infants would be screened for hypothyroidism and those positively diagnosed will be treated.*
- *The capability for diagnosing cystic fibrosis will be improved.*
- *Extension of these techniques to other congenital diseases such as phenylketonurea and Down's syndrome should be easily possible.*

Assuming success, what would be the impact of the project on the overall problem/need/opportunity?

The infant mortality rate will be reduced. The use of public funds to support retarded children will be significantly reduced.

What data or estimates support these impact projections?

For hypothyroidism, the incidence is estimated to be 1 in 3,500. Total births are in the order of 55,000 per annum. Only infants in the Montevideo area (21,000) are being screened. This translates into a number of neonatals not being diagnosed and treated. A cost benefit analysis indicates a ten-fold benefit.

Would the proposed project be supported by the institution/Government? If so, to what extent?

- *For the extension of hypothyroidism screening, basic infrastructure and qualified medical staff will be made available by the Ministry of Public Health and the Faculty of Medicine. Assistance to introduce the relevant technology and to organize a national training course on RIA techniques will be provided.*
- *Collaboration with the National Vaccination Programme is envisaged.*
- *For cystic fibrosis, staff will be made available for training in radionuclide-based DNA techniques.*
- *The Nuclear Medicine Centre will play a coordinating role.*

Is there – or is it expected that there might be – interest in this project in international development agencies or international institutes currently operating in the country? What agencies or institutes? What information indicates possible interest?

None are mentioned.

Would the nuclear technologies available through the IAEA be the best means available to achieve the goal of the project?

The use of RIA and radionuclide-based diagnostic methods are excellent applications of the peaceful uses of nuclear science and technology. The former is well-established for neonatal screening for hypothyroidism. The use of other techniques utilizing radionuclide-containing reagents for diagnosing other disorders could represent a significant opportunity.

Who would be the "end user" in the proposed project, i.e. the person or body who would ultimately apply the technology to bring about social or economic impact?

The "end user" is the entire future population of Uruguay as all will be subject to screening. Also, the institutions involved in the diagnosis will be strengthened to carry out this task. The Ministry of Public Health will benefit by the reduction in support for retarded children and could deploy funds for other purposes.