

# NEWBORN SCREENING IN AUSTRALIA AND NEW ZEALAND

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**Abstract.** Newborn screening began in Australia and New Zealand in the mid-1960's as local and pilot programs and implemented as country or state-wide programs around 1970. There are five programs covering all Australia and one for New Zealand. All screening programs are fully government funded, as is treatment for the conditions found by the screening programs and newborn screening is a universally adopted policy funded by the government. Some have additional involvement in program advisory committees. There are no major problems sustaining existing screening, however, some programs have financial problems with funding for new equipment. Other problems include storage and other uses of residual dried blood samples; consent issues; protocols for action after screening and introduction of expanded (tandem mass spectrometry) screening. New activities vary from program to program – working towards expanded newborn screening and collaborative projects for the evaluation of this screening and development of screening for lysosomal storage disorders. All programs are working towards automation of punching and testing and increased automated data handling and reporting.

## INTRODUCTION

Newborn screening began in Australia and New Zealand in the mid-1960's as local and pilot programs and was implemented as country or state-wide programs around 1970. There are five programs covering all Australia and one for New Zealand (see Table 1).

## SCREENING POLICY

The Newborn screening policy for the region was formulated by a committee of the Human Genetics Society of Australasia (HGSA) and the Royal Australasian College of Physicians, Division of Pediatrics. The

committee members include: Wayne Cutfield (NZ), Janice Fletcher (South Australia), Ivan Francis (Victoria, secretary), Lawrence Greed (Western Australia), Steve Kahler (Victoria), Barry Lewis (Western Australia), Jim McGill (Queensland), Enzo Ranieri (South Australia), Andrew Thomas (Queensland), Dianne Webster (NZ, chair), Bridget Wilcken (New South Wales), and Veronica Wiley (New South Wales). The role of the committee is to develop a newborn screening policy for Australasia and to share newborn screening experience among the six Australasian screening programs. In addition, some programs have local advisory committees.

The policy suits the Australasian programs but individual programs or groups of programs should develop their own policy taking local conditions (such as disease prevalence) into account. For example, it is unlikely that screening for cystic fibrosis would be recommended in a country without a significant Caucasian population.

The full text of the policy is available from the Human Genetics Society of Australasia (<http://www.hgsa.com.au>) and a summary follows:

**1. General recommendations** – The condition screened should benefit from early diagnosis. The benefit should be reasonably balanced against financial and other costs. There should be a suitable test and a system to deal with follow-up including diagnosis and treatment.

Table 1. Size of Australasian screening programs.

Program	Approximate annual births (babies screened)
New South Wales	94,000
Victoria	69,000
New Zealand	57,000
Queensland	49,000
South Australia	28,000
Western Australia	25,000

**2. Recommendations about how programs are organized**

– The screening program comprises everything from policy through specimen collection to treatment and audit. Programmes should be managed by a state-funded body of health professionals with the policy assistance of a multidisciplinary advisory committee. Screening laboratories should be large enough to minimise costs and accumulate experience and performance data. Laboratories should have appropriate accreditation. The HGSA will run a quality assurance program. Screening program audit parameters should include test sensitivity, specificity, positive predictive value, timeliness of reporting, and short and long-term outcome of diagnosed patients. Health care authorities have a responsibility to ensure that tests are available to all babies born in their region. Sample submitters should be notified of tests received. Information about the program should be available to the public and health professionals including written information for parents.

**3. Recommendations about legal and ethical issues**

– For each baby born there should be an identified individual responsible for ensuring that screening and follow up are done and that parents have information about the test. The screening program is responsible for correct interpretation and reporting of results, and ensuring that follow up occurs. Screening program participation should not be mandatory for individuals. The privacy of individuals should be protected. (Note: A separate HGSA policy exists concerning storage and use of samples and is discussed elsewhere in the volume).

**4. Research** – Research into the natural history of disorders (for which screening is actually or potentially available), and into possible new screening

tests should be funded.

**5. Disorders** – The committee recommends that all infants be screened for PKU, congenital hypothyroidism, and cystic fibrosis. There is a list of disorders for which good tests are available and for which screening is recommended if the local circumstances permit (including galactosemia and MCAD); and another list of disorders for which tests are available and screening is not recommended for various reasons. For the Australian States and New Zealand, screening is universal and the state or local government pays the costs of both screening and treatment. The disorders covered by screening are given in Table 2.

PROGRAM OVERVIEW

There are no major problems sustaining existing screening although some programs have financial problems with funding for new equipment. Issues under discussion include storage and other uses of residual dried blood samples; consent; protocols for action after screening and introduction of expanded (tandem mass spectrometry) screening. The committee has produced a policy regarding storage and the use of residual dried blood spots. New activities vary from program to program and include working towards expanded newborn screening and collaborative projects for the evaluation of this screening and development of screening for lysosomal storage disorders. All programs are currently working towards automation of punching and testing and increased automated data handling and reporting. Regional consideration is given to the benefits of screening and mutual areas of concern *ie* cultural, financial and developmental similarities. Programs share successful strategies, and occasionally form purchasing coalitions for better pricing on screening reagents and supplies.

Table 2. Disorders covered by the Australasian newborn screening programs.

	New South Wales	New Zealand	Queensland	South Australia	Victoria	Western Australia
PKU	yes	yes	yes	yes	yes	yes
MSUD	yes <sup>a</sup>	yes		yes <sup>a</sup>		
Galactosemia	yes	yes	yes	yes		yes
Biotinidase deficiency		yes				
Cystic fibrosis	yes	yes	yes	yes	yes	yes
CH	yes	yes	yes	yes	yes	yes
CAH		yes				
MS/MS <sup>b</sup>	yes			yes	soon	

<sup>a</sup>as part of expanded newborn screening.

<sup>b</sup>MS/MS refers to expanded metabolic testing.

# NEWBORN SCREENING: EXPERIENCE OF BANGLADESH

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**Abstract.** Newborn screening for early diagnosis of many preventable diseases is already an established program in developed countries as well as in some countries in Asia. With the control of infectious and communicable diseases, the Bangladesh government is now giving more attention to the preventive aspect of health. The country is now preparing to adopt newborn screening as part of the preventive aspect of health. Among the various activities are laboratory set up, expert visits, pilot studies on CH and training of personnel. A national project to screen newborns for congenital hypothyroidism is now under active consideration. A universal newborn screening program is a highly ambitious project for a country like Bangladesh. Funds to run such a program will be the main problem. The social pattern where still more than 80% of deliveries are done at home is another constraint. However, the enthusiasm of the professionals as well as of the government is a very encouraging thing. Now it needs the support of the international communities.

## INTRODUCTION

Bangladesh is a small country of 147,570 sq. km. and has a large population of about 130 million. The annual growth rate of the country is about 2.8% with an estimate of 3.0 million births in Bangladesh every year. Iodine deficiency is endemic in the country. Goiter and other iodine deficiency disorders (IDD) are very common and are known from ancient times. Both adult hypothyroidism and cretinism are quite frequent. With the increased attention on congenital hypothyroidism (CH), the concept of newborn screening has also come up.

Newborn screening for the early diagnosis of many preventable diseases is already an established program in developed countries as well as in some countries in Asia. Bangladesh, a developing country of East Asia, has many other problems to fight in the health sector. With infectious and communicable diseases being controlled, the government is now giving more attention to the preventive aspect of health. With that policy, the country is taking preparations to adopt newborn screening as part of the preventive aspect of health.

Ten years back, Bangladesh participated in an IAEA project using the filter paper technique for examining blood samples for TSH. The initial work revealed the presence of hypothyroidism in children (cretinism) in Bangladesh. However, due to lack of proper knowledge, the newborn screening program could not be continued at that time.

Efforts to establish a pilot project of neonatal screening for congenital hypothyroidism dated as far back

as 1999. After the Korea Meeting organized by the IAEA in May 1999, in connection with the project on neonatal screening for congenital hypothyroidism, (RAS/6/032) new enthusiastic ideas to establish the program of newborn screening emerged in Bangladesh. Several meetings and seminars were organized to focus on the problem of congenital hypothyroidism and the necessity of screening. A national seminar was held in Dhaka on the 18th of June 2000 to discuss the problem. The seminar was successful in creating awareness about the newborn screening program. The program gained much support from the medical community especially the pediatricians and obstetricians. The government of Bangladesh also showed keen interest on the program and approved a pilot project on neonatal screening in Bangladesh. At present, the newborn screening program to detect congenital hypothyroidism is ongoing under the patronization of two projects, one by the IAEA (RAS/6/032) and the other by the Government of Bangladesh.

## MATERIALS AND METHODS

For newborn screening of congenital hypothyroidism, TSH was adopted as the screening parameter. A TSH value of 20 mIU/L was used as the cut off value. Blood was collected through the filter paper technique. TSH was measured with filter paper technique by IRMA method. Both cord and heel prick blood were used. However, due to a shortage of hospital beds, most of the babies delivered by normal vaginal route were discharged on the first day. Cord blood was thus collected due to early discharge. The nurses attending the deliveries usually collected the blood in pre-supplied special filter papers.

A person working in the laboratory collects the samples from the specified hospitals of Dhaka city twice in a week. The samples from outside Dhaka were sent by post. The samples were analyzed in the in-vitro laboratory of Institute of Nuclear Medicine, Dhaka twice a week and the results were sent to the respective hospitals. Initially, due to lack of experience, a good number of samples were found insufficient. At present, almost all the samples were good enough for analysis.

### **Progress of the project**

Initial laboratory set up and necessary training for technologists were completed to serve the pilot project. Motivation of the concerned people was also reasonably achieved. Though in small number, the actual screening program was also started. At present, screening is ongoing to detect congenital hypothyroidism only. Blood samples from newborns were collected mainly from big hospitals of Dhaka City. However, some samples were also collected from outside Dhaka. Up to the present, some 10,000 samples have already been analyzed.

Prof Dong Hwan Lee of Korea visited the country in March 2001 under an IAEA program. He discussed many points about newborn screening with the concerned persons. His visit to Bangladesh gave a new momentum for the newborn screening program of the country.

### **RESULTS AND DISCUSSION**

The initial attempt to start newborn screening has been accepted by the government as well as by the professionals with a positive attitude. Screening is ongoing for the last three years under a project of the IAEA (RAS/6/032). One pilot project under the government of

Bangladesh has also been completed successfully. It is of great hope that the government is satisfied at the outcome of the project and has renewed the project for another year. The government is also actively considering the inclusion of the newborn screening project for CH in the Annual Development Budget.

### **Problems faced**

The main problem faced is the difficulty in the collection of samples. Lack of manpower and lack of orientation are two major factors. Funding is also a big problem. Even at present, no separate newborn screening center is available.

### **Future program**

The Government of Bangladesh has agreed, on principle, to run a project on newborn screening to detect congenital hypothyroidism. The government is actively considering the approval of a national project. Bangladesh is also interested to continue the IAEA project (RAS/6/032), which is supposed to go up to 2010.

### **CONCLUSION**

Bangladesh is a developing country with many other problems. The universal neonatal screening program is a highly ambitious project for a country like Bangladesh. Funds to run such a program will be the main problem. The social pattern where more than 80% of deliveries were done at home still, is another constraint. However, the enthusiasm of the professionals as well as of the government is a very encouraging point towards the establishment of a newborn screening program. Now, it needs the support of the international communities.