# Newborn Screening

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## IHG, PNRI host 1st IAEA coordination meeting

*Delegates commit to a comprehensive newborn screening for congenital hypothyroidism in East Asia*

The **Institute of Human Genetics (IHG)** and **Philippine Nuclear Research Institute (PNRI)** hosted the **First International Atomic Energy Agency (IAEA) Coordination Meeting on National Programmes for Congenital Hypothyroidism in South East Asia** last October 16 to 20, 2000 at the National Institutes of Health.

Renowned pediatricians and scientists from 10 countries namely Thailand, Korea, Malaysia, Myanmar, Indonesia, Mongolia, China, Vietnam, Bangladesh, and the Philippines attended the meeting.

The IAEA spearheaded the five-day conference as part of its initiative to develop a regional program that will promote neonatal screening and to counter congenital hypothyroidism in East Asian nations. The agency pledged its support to the delegates in the form of expert missions, supply of reagents, laboratory equipments, technology transfer, and financial grants.

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***Toward a common goal of countering CH.*** *UP Manila Chancellor Alfredo T. Ramirez (seated left) poses with the participants of the IAEA meeting. Seated (from L) are PNRI representative Pilar Roseles and IAEA representatives Dr. Reyad Kamel and Dr. Soo Lin Ch'ng. Standing are (L-R): Dr. Nguyen Thi Hoan, Vietnam; Dr. Myint Aye Mu, Myanmar; Ms. Wiyada Charoensiriwatana, Thailand; Dr. Carmencita Padilla, Philippines; Mr. Frans Sardi Satyawiranan and Ms. Diet Sadiah Rustama, Indonesia; Mr. Shi Lixin, China; Dr. Fauzia Moslem, Bangladesh; Dr. Dong Hwan Lee, Republic of Korea; Dr. Rohanna Ismail, Malaysia; and Ms. Oyun Nanzad, Mongolia.*


## THE NEED FOR NEWBORN SCREENING LEGISLATION

Newborn screening in the Philippines started with a dream. A dream that all Filipino babies will be saved from mental retardation. That all Filipino babies will be given a chance to live normal lives and grow up to be productive and healthy individuals.

That dream began to be realized in 1996 when a group of obstetricians and pediatricians introduced newborn screening in the Philippines. The group named itself the ***Newborn Screening Study Group*** and the pilot project was called the ***Philippine Newborn Screening Project***. The objectives of the project are: (1) to establish the incidence of six metabolic conditions namely: congenital hypothyroidism (CH), congenital adrenal hyperplasia (CAH), phenlyketonuria (PKU), homocystinuria (HCY), galactosemia (GAL), and G6PD deficiency, and (2) to make recommendations for adoption of newborn screening in a nationwide scale.

Since then, newborn screening proved to be an important part of preventive newborn medicine. As of November 2000, there are already 153 hospitals offering newborn screening and has screened a total of 135,000 babies wherein 53 were confirmed to have one of the disorders included in the program. Based on the data collected from samples and studies conducted by NBS, early detection of metabolic disorders through newborn screening can save 1,500 babies a year from mental retardation and death.

However, the creation of the Newborn Screening Study Group and the initial implementation of the project is just the first step toward a bigger goal - the ***nationwide implementation.***

*(Continued on page 3)*

# 4TH Asia Pacific Meeting on Neonatal Screening to be held in Manila

The **4th Asia Pacific Regional Meeting of the International Society for Neonatal Screening** is slated on October 17 to 19, 2001 at the EDSA Shangri-la Hotel, Mandaluyong City.

With the theme, ***"Newborn Screening in the Asia Pacific Region: New Century, New Goals, New Opportunities,"*** the meeting will provide a potent venue for the exchange of medical knowledge, ideas, insights, and experiences on newborn screening and for the discussion of pressing issues, emerging trends, and recent developments on screening around the world.

The three-day conference, with target participants all over Asia-Pacific, will feature plenary sessions on current issues and developments on newborn screening, oral presentation of researches of experts and researchers, poster sessions, panel discussions, and a workshop on the latest screening technology.

Topics to be discussed in the plenary sessions include: application of Human Genomics to newborn screening; setting national priorities for newborn screening; management issues in inborn errors of metabolism: PKU, Galactosemia, MSUD; bio-ethical issues in newborn screening; new developments in newborn screening; monitoring outcomes in neonatal screening; quality assuarance in newborn screening; and country reports.

Symposia and parallel discussions will highlight topics on diagnostic modalities in newborn screening (old technology amidst newer methods; applications of molecular techniques, DNA chip technology); future directions in newborn screening; newborn hearing screening; strategies for a successful community implementation; emerging programs in the East Asia; screening for other disorders: infectious disorders and hearing screening; overcoming obstacles in newborn screening; congenital hypothyroidism (CH); congenital adrenal hyperplasia (CAH), galactosemia (GAL); phenylketonuria (PKU), Glucose 6-phosphate dehydrogenase (G6PD) deficiency; and free papers.

For inquiries, you may contact the 4th APRM-ISNS Conference Secretariat at telephone (632) 302-1626; fax 526-9997 or email at newbornasia@mydestiny.net. You may also visit the conference website at http://www.newbornasia.com.

*(IAEA Meeting, from page 1)*

The meeting served as a venue for participants to discuss problems on the implementation of newborn screening programs in their respective countries and to identify possible solutions and actions to address the problems. The participants likewise charted future plans and activities for their screening programs. The meeting was concluded with the commitment from all the delegates to achieve their goal of developing a comprehensive newborn screening program for congenital hypothyroidism in the region.

IAEA representatives, Dr. Reyad Kamel and Dr. Soo Lin Ch'ng facilitated the coordination meeting. Institute of Human Genetics director and local proponent of newborn screening, Dr. Carmencita Padilla, represented the Philippines in the said meeting. ■

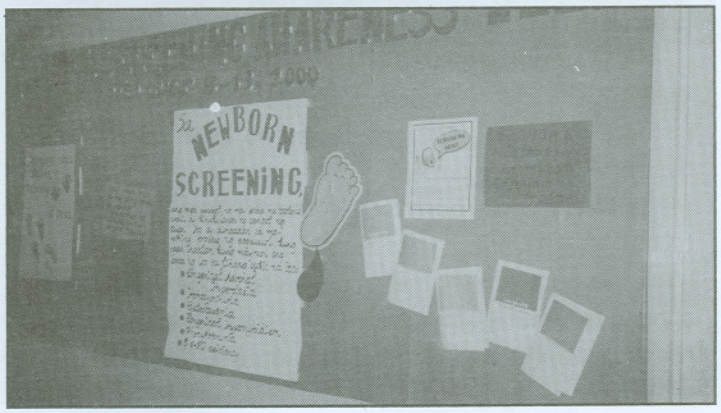
# HOSPITAL UPDATES

## NBS member-hospitals expand to 153

From 53 hospitals in 1999, Newborn Screening (NBS) member-hospitals increased to **153** as of November 2000.

The 188-percent increase is a result of the issued Department of Health (DOH) Administrative Order a-1, s 2000, which mandated that all DOH-retained and nationalized hospitals shall participate in the newborn screening project by year 2000. The growing number of member-hospitals will generate a more comprehensive and successful implementation of newborn screening.

## NBS awareness week celebrated

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**DOH Intervention**

Results of pilot studies on newborn screening which show the need for appropriate government assistance, prompted the Department of Health (DOH) to participate in the implementation of the project. The DOH issued Administrative Order a-1, s 2000 on the *"Policies on the Nationwide Implementation of Newborn Screening"* to encourage all hospitals to implement newborn screening.

The DOH appointed and trained regional coordinators to advocate and promote newborn screening and to monitor the performance of hospitals within their regions. Likewise, hospital coordinators were also identified and trained in all DOH retained hospitals to supervise the implementation of newborn screening.

Despite these efforts, only 10% of the 2 million babies born annually are screened through the project. This is because newborn screening is only conducted to hospital-born babies and not even all of them are being screened. Seventy percent of Filipino babies are delivered at home and these babies are not aware of or do not have access to newborn screening.

Leaving the decision to screen or not to screen on the physician or parent will not ensure the conduct of newborn screening on the baby. Still, most physicians and parents are not yet convinced of the merits of newborn screening. In some cases, parents view the tests as additional expenses and not as essential measures to ensure the health and nomal growth of their babies. In addition, hospitals are not obliged to implement newborn screening and do not have a financial scheme to cover the cost of the tests.

**Need for Legislation**

Legislation of newborn screening will fast track the implementation of newborn screening as an essential component of quality newborn care. It is the only way to ensure that all newborns will be given a chance to be saved from mental retardation and death.

It is the responsibility of the government to guarantee a productive and fruitful life to all Filipino newborns. It should provide for the cost of screening to eliminate the additional financial burden from the parents.

In addition, extensive studies have shown the merits of newborn screening by reversing mental retardation and death. Furthermore, studies also show that the cost of newborn screening is only a fraction compared to the productivity loss of a potentially normal person and the actual cost of diagnosing and caring for a retarded child.

Indeed, newborn screening legislation will surely make our dream come true - the dream of giving Filipino newborns a chance to live productive and normal lives. ■

**special ♋•♋•♋ report**

# NBS receives Outstanding Health Research Award

The **Philippine Center for Health Research and Development (PCHRD)** presented the **2000 Outstanding Health Research Award** to the **Philippine Newborn Screening (NBS) Project** last July 2000 at the Manila Hotel.

NBS received a trophy and a cash prize of P 500,000. The cash prize was utilized by the NBS to benefit all member-hospitals that participated in the project.

In recognition of the hospitals' participation in the success of the project, the amount was allocated to procure computers as part of the NBS' nationwide computerization and networking system in order to upgrade the communication system between hospitals and the central laboratory.

The computers are equipped with modems for easy internet access, primarily for the prompt release of screening results and recall of patients through e-mail, fast communication of problems and queries between the Secretariat's Office and the member-hospitals, and easier access to the data base of patients. It can also be used to retrieve current news and developments on newborn screening, important announcements and notices, project updates, hospital performance and statistics, and access to information and educational materials on newborn screening (e.g. NBS orientation slides, brochures, training manuals, and modules.)

The NBS shouldered half of the cost of the computers and the member-hospitals paid for the other half. This is to ensure that all hospitals offering newborn screening will be able to benefit from the cash prize awarded by PCHRD.■

*Institute of Human Genetics director and local proponent of the Newborn Screening Dr. Carmencita Padilla shared the joy and triumph of the PCHRD Outstanding Health Research Award for the year 2000.*

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# Parents' Support Group for Newborn Screening formed

A small group of parents with a common goal of promoting newborn screening was formed last September. During the seminar for parents with children affected with G6PD deficiency, a number of them volunteered to be a part of a support group that will help in the awareness campaign for newborn screening. The group, which decided to call itself the Philippine Association of Parents for Newborn Screening, Inc., has embarked on a three-pronged mission of (a) creating awareness on newborn screening among the general public through an intensive information campaign; (b) fast tracking the legislation of newborn screening through various advocacy activities; and (c) acting as a group who will provide emotional support top other parents who may have children affected with inborn errors in metabolism.

The Association, headed by Mario Mercado, is composed of parents whose children have been found positive of G6PD deficiency, congenital adrenal hyperplasia (CAH), and congenital hypothyroidism (CH).

The first activity of the association was a photo session featuring their own children portrayed as happy kids living normal lives. These pictures were used in an exhibit made by the parents themselves for the newborn screening awareness campaign last October 9-13, 2000, at the Philippine General Hospital lobby. The theme of the exhibit was " Our babies were saved. Thanks to Newborn Screening." To augment the message of the exhibit, flyers about newborn screening and its benefits were distributed.

The initial activities of the Philippine Association of Parents for Newborn Screening Inc. will be geared toward an information campaign about this screening modality.

If you are a parent of a child affected with an inborn error of metabolism and you are willing and interested to join the association, you may call the:

**Philippine Association of Parents' for Newborn Screening, Inc.**
c/o The Institute of Human Genetics,
3/F National Institutes of Health,
University of the Philippines Manila
Telephone: 5261710 or 5261725
Fax 526-9997/307-0788
Email: ihg@mydestiny.com

## PROJECT UPDATE
*(As of October 2000)*

| | | | |
|---|---|---|---|
| ***PATIENTS SCREENED*** | | | 135,906 |
| ***CONFIRMED POSITIVE*** | | | |
| *Congenital Hypothyroidism (CH)* | 32 | | 1:4,247 |
| *Congenital Adrenal Hyperplasia (CAH)* | 16 | | 1:8,494 |
| *Galactosemia (GAL)* | 2 | | 1:67,953 |
| *Classical Galactosemia* | | 1 | 1: 135,906 |
| *Galactokinase Deficiency* | | 1 | 1: 135,906 |
| *PKU/Hyperphenylalanemia* | 3 | | 1:45,302 |
| *Atypical PKU* | | 1 | 1:135,906 |
| *BH4 Deficient* | | 1 | 1:135,906 |
| *PBH* | | 1 | 1:135,906 |
| **TOTAL** | 53 | | 1:2,564 |
| ***G6PD SCREENING*** | | | |
| *Total Screened* | 61,595 | | |
| *Total Confirmed* | 856 | | 1:72 |