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New NSC Opens in Northern Luzon

International newborn screening accreditors, along with local experts and personnel of NSRC and the new Newborn Screening Center–Northern Luzon, gesture a sign of approval after the center underwent a two-day accreditation process held at MMMHMC in Ilocos Norte.

Ilocanos, rejoice!

The Newborn Screening Center–Northern Luzon (NSCNL) is open and now provides required laboratory tests and follow-up programs for newborns with heritable conditions in the regions of Ilocos and Cagayan. NSCNL is hosted by Mariano Marcos Memorial Hospital and Medical Center (MMMHMC) in Ilocos Norte.

The Department of Health (DOH), Newborn Screening Reference Center (NSRC), and international newborn screening experts from the United States and Australia granted the NSCNL three years of accreditation after passing the final extensive inspection on May 10-11, 2017. The new center is the sixth accredited NSC in the country.

Representatives from the Health Facilities and Services Regulatory Bureau evaluated the physical structure of the center and ensured that it complies with all rules and regulations governing health facilities. International accreditors Dr. Bradford Therrell, Jr,. and Dr. Veronica Wiley and local experts Dr. Carmencita Padilla and Dr. Anna Victoria Sombong conducted the final leg of accreditation. Representatives from NSRC—Charity Jomento, Jovy Ann Lising, and Dr. Maria Melanie Liberty Alcausin—also joined the team.

MMMHMC is a third-level referral tertiary general hospital located at Barangay 6 San Julian, Batac, Ilocos Norte. It provides medical and health services to Ilocos Norte and neighboring provinces. The accreditation of NSCNL is part of the expansion of services of MMMHMC. Dr. Ma. Virginia Paz K. Otayza is the unit head of NSCNL, while Dr. Modesty Leaño acts as Laboratory Head and Anthony James Almazan as Program Manager.

Dr. Bradford L. Therrell, Jr., of the US National Newborn Screening and Global Resource Center, Dr. Veronica Wiley of the New South Wales Newborn Screening Programme, and Charity Jomento (left photo, rightmost) of NSRC inspect the laboratory facilities of NSCNL.

NSRC Puts Up Exhibit for International Nurses Week

NSRC signed up as an exhibitor at the Philippine Nurses Association's 2nd Summer Conference on May 12, 2017, at Century Park Hotel in Manila.

Held in observance of the International Nurses Week, the conference carried the theme "Filipino Nurse: A Voice to Lead, Achieving Sustainable Development Goals." Some 400 nurses from all over the country attended the conference, where NSRC updated the nurses on the developments of newborn screening including the upcoming National Newborn Screening Convention in October.

NSRC offers brochures and newsletters and displays presentations to conference participants during the Philippine Nurses Association's Summer Conference in Manila.

The Case of Dave . . . *from page 1*

"The condition of my children tested our faith. However, eventually, my husband and I learned to accept the situation. We realized that our kids are still blessed. We saw children with worse conditions. I am thankful that Dana was screened."

Dana started the phenylalanine-free medical formula immediately after she was confirmed to have PKU. Fortunately, too, Lorelie was able to give her daughter a measured volume of expressed breast milk. She also worked closely with a dietitian from the Philippine General Hospital (PGH) to design a diet. A recipe book was prepared for them, as PGH does for patients afflicted with PKU.

Lorelie was not familiar with newborn screening until she gave birth to Dana.

"Newborn screening saved Dana's life," admitted Lorelie. "It is important for every parent to understand and realize the importance of newborn screening. Any baby can be born with a congenital disorder, and it's hard to take any chances."

Recipe book given by PGH to parents of patients with metabolic disorders like PKU

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DOH-RO 3 Brings Together Central Luzon's Saved Babies

More than a hundred saved babies and their parents shared a day of fun and laughter as DOH–Regional Office (RO) 3, in coordination with the region's NBS Continuity Clinic (NBSCC), spearheaded the Reunion of Saved Babies in Central Luzon at Jose B. Lingad Memorial Regional Hospital, San Fernando City, on May 9, 2017.

Representatives from NSC–Central Luzon (NSCCL), the provincial and city health offices, and the provincial DOH NBS coordinators prepared interactive games for both kids and parents/guardians alike. A Jollibee mascot also made a surprise appearance to the cheers of everyone.

The reunion was not only fun-filled, but it was also a learning opportunity for parents to gain more understanding about the condition of their children. Dr. Maria Victoria Macalino, NBSCC Head, discussed the proper management of the disorders included in the newborn screening panel, while Jaqueline Licup, NBSCC Nurse, shared the importance of proper compliance to the continuity clinic.

Some parents and guardians became emotional as they shared their experiences on raising babies positive with disorders screened by newborn screening, inspiring other attendees to become more supportive to the program by becoming advocates of Expanded NBS (ENBS). The parents and guardians were thankful to the program because with just a drop of blood, their children were saved from mental retardation and death. *NDelaCruz*

Region 3 Strengthens Referral and Management System

Organizers and participants of the reorientation for newborn screening coordinators in Tarlac and Aurora on May 31, 2017

To establish a proper and better referral and management system and strengthen the partnership between the NBSCC and the private sector, DOH-RO 3, in coordination with the NSCCL, conducted six batches of reorientation on ENBS, the management of the disorders that are included in its panel, and the system of referral for cases in April and May 2017.

The series of reorientations was held in the different provinces of Central Luzon and brought together private practitioners from Bulacan, Pampanga, Tarlac, Aurora, Nueva Ecija, Bataan, and Zambales.

Continued on page 8 . . .

Reunion participants get warmed up for a fun day at Jose B. Lingad Memorial Regional Hospital, San Fernando City.

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DOH-RO 9 Holds G6PD Deficiency Forum

Dr. Conchita Abarquez (standing) discusses G6PD Deficiency to parents and guardians who attended the forum.

DOH-RO 9 Family Health Cluster, in coordination with the NSC-Mindanao (NSCM), conducted the G6PD Deficiency Forum for caregivers and healthcare workers in Zamboanga del Norte at Mibang Hotel, Dipolog City, on June 16, 2017.

Some 50 parents and guardians, 30 G6PD-deficient patients, and 20 healthcare workers from the different rural health units and city health offices attended the forum. The Integrated Provincial Health Office NBS Coordinator was also present.

Dr. Conchita Abarquez, NSCM Unit Head, discussed G6PD Deficiency and addressed queries, concerns, and misconceptions about the disorder. Parents, in return, expressed their gratitude to organizations that help them out in taking care of their children. *IRTomas*

Zamboanga NSFs Get ENBS Training

Twenty-six nurses, midwives, and medical technologists from the different newborn screening facilities (NSFs) in the region attended the ENBS Training conducted by DOH-RO 9 at Grand Astoria Hotel, Zamboanga City, on June 13-14, 2017.

NSCM Unit Head Dr. Conchita Abarquez delivered a lecture on ENBS updates. NBS Coordinators April Joy Calibot and Iris Roma Tomas, together with personnel from Zamboanga City Medical Center (ZCMC) and NSCM, facilitated the training. Practicum was held at the ZCMC where participants put into practice what they had learned during the lectures. *IRTomas*

CDO Hosts Refresher Course

As part of its annual routine, DOH-RO 10 conducted the Refresher Course on the NBS Program at the Pearlmont Inn, Cagayan de Oro (CDO) City, on May 12, 2017.

Thirty-five participants composed of nurses, midwives, and medical technologists who serve as program coordinators in their respective facilities attended the activity.

During the training, data relating to samples sent, unsatisfactory samples, return rates, transit time, and age of newborns during collection were presented to provide a glimpse of how the program was being implemented the region. Strategies and techniques on how to avoid common but perennial errors were also discussed to keep the number of oversights within the acceptable rate. *RAguilar*

Perly Bermudez (standing), NSCM Project Development Offer, speaks to participants about some aspects of ENBS.

NSC and NSRC Personnel Attend NSC Summit

Participants composed of personnel from all six NSCs and the NSRC pose for a group photo during the summit held at a resort in Laguna.

Laboratory managers from all the newborn screening centers (NSCs) lead the lectures during the Summit for Strategic and Directional Planning of the NBS Program at the Caliraya Resort Club Inc. in Laguna on May 26-28, 2017, to address the need for continuing information, education, re-education, and training among NSC personnel on NBS procedures, operations, and management.

The two-day event focused on the future of the NBS program, where participants discussed how to build better work relationships, benefit from a culture of collaboration, and achieve and maintain work-life balance.

The summit also included team-building activities to guide and encourage personnel from NSRC and the different NSCs to work together as a team toward achieving the program's national objectives.

A total of 191 personnel from NSRC and all six NSCs participated in the event. Employees who were with the program for more than five years were also recognized.

Dr. Carmencita Padilla, NSRC consultant and University of the Philippines Manila Chancellor, also graced the event.

IN THE KNOW: PHENYLKETONURIA (PKU)

What is PKU?

Phenylketonuria (PKU) is a disorder of aromatic amino acid metabolism in which phenylalanine cannot be converted to tyrosine due to a deficiency or absence of the enzyme phenylalanine hydroxylase. Phenylalanine hydroxylase requires the co-factor 6-pyruvoyltetrahydropterin or BH4 for activity in the hydroxylation to tyrosine, absence of this co-factor may present with an increase in plasma phenylalanine similar to phenylketonuria but is considered a separate disorder.

Clinical Manifestation

Patients affected with PKU appear normal at birth. The most important and sometimes the only manifestation of PKU is mental retardation. Patients may present with constitutional, intellectual, and neurologic abnormalities and signs as well as hypopigmentation of the skin and hair and iris rapidly develop due to impaired metabolism of melanin. Seizures occur in a fourth of patients.

The odor of the phenylketonuric patient is that of phenylacetic acid described as mousy, barny, or musty.

Pathophysiology

PKU results from a deficiency of activity of a liver enzyme, phenylalanine hydroxylase, leading to increased concentrations of phenylalanine in the blood and other tissues. Elevated phenylalanine interferes with myelination, synaptic sprouting, and dendritic pruning; in addition, it competitively inhibits the uptake of neutral amino acids in the blood-brain barrier, causing reduced tyrosine and tryptophan concentrations thereby limiting the production of neurotransmitters.

Inheritance: autosomal recessive

Confirmatory Testing

The demonstration of decreased enzyme activity is confirmatory. However, in the presence of increased phenylalanine levels, it is important to differentiate phenylketonuria from a BH4 deficiency. This is accomplished through the administration of tetrahydrobiopterin (doses of 2 mg/kg intravenously and 7.5-20 mg/kg orally), which leads to a prompt decrease to normal in the concentration of phenylalanine. Pterin metabolites in urine are likewise useful, demonstrating a very low biopterin and high neopterin levels.

Overview of Disease Management

Dietary management is key to treatment. The diet of patients has four components: (1) complete avoidance of food containing high amounts of phenylalanine; (2) calculated intake of low protein/phenylalanine natural food; (3) sufficient intake of fat and carbohydrates to fulfill the energy requirements of the patient; and (4) calculated intake of phenylalanine free amino acid mixture supplemented with vitamins, minerals, and trace elements as the main source of protein.

Prognosis: When treatment is started early and performed strictly, motor and intellectual development can be expected to be near normal.

Preliminary/Initial Management during Metabolic Crisis: Metabolic crises may be caused by illness, prolonged fasting, or stressful situations such as surgery and severe infection. The goal of the treatment is to reverse the catabolic state, correct the acidosis, and prevent essential amino acid deficiency.

What to Do

If unwell and cannot tolerate oral intake:

- Nothing per orem
- Ensure patient's airway is secure
- Insert IV access. Collect dried blood spot for phenylalanine levels. May request for investigations (i.e., CBC, blood gas, etc.) as needed.
- May give fluid boluses if patient requires.
- Start D12.5% 0.3NaCl at full maintenance. Assess patient clinically; if there is a need to increase fluid, may do so up to 1.2x or 1.5x the maintenance.
- Monitor input and output strictly (q6 hours).

If unwell and can tolerate oral intake:

- Insert oro- or nasogatric tube and start continuous feeding with PKU milk formula or protein free formula at maintenance rate.
- Insert IV access. Collect dried blood spot for phenylalanine levels. May request for investigations (i.e., CBC, blood gas, etc.) as needed.
- Start D12.5% 0.3NaCl at 5-10 cc/hr
- Monitor input and output strictly (q6 hours)

* Children should not be protein restricted for longer than necessary (24-48 hours).

*Inform metabolic doctor on call for further guidance regarding ongoing management.

Source: Fact Sheets for Doctors (May 2016) available at www.newbornscreening.ph.

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Region 3 Strengthens Referral . . . *from page 4*

Maria Elissa Veronica Benipayo, NSCCL Program Manager, gave an introduction to newborn screening, its history, and highlights of the Newborn Screening Act of 2004, while Dr. Crislyn Samia, NSCCL Follow-up Manager, discussed the updates on ENBS, the disorders included in the panel, and their management. Maria Regina Yutuc, NSCCL Follow-up Officer, explained the process of recall and follow-up of positively screened patients and the referral system.

Participants were also able to raise questions regarding the issues and concerns they faced in the implementation of the program. *NDelaCruz*

Organizers and participants of the ENBS Reorientation Seminar for the provinces of Bataan and Zambales on May 16-17, 2017

Your Feedback Is Important to Us!

Thank you for reading our newsletters!
We would love to hear your feedback. Please let us know how we can improve our bimonthly newsletters by answering our feedback form at www.newbornscreening.ph. If you would like to write an article, please contact us at info@newbornscreening.ph. We will do our best to keep you informed about current and relevant newborn screening issues.

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The Case of Dave and Dana: Caring for Children with PKU

WHAT'S INSIDE

"My son had a late diagnosis; he was diagnosed at age 2," narrated Lorelie De Guzman during an interview conducted in Batangas. "Not until my daughter was born, screened, and detected to have Phenylketonuria (PKU) that my son was diagnosed to also have the same disorder."

Lorelie found out about PKU when Dana, the younger of the two, was diagnosed with the disorder through newborn screening. After her daughter was confirmed to have PKU, her firstborn, Dave, was also tested and diagnosed to have the same disorder.

"We had no idea and we did not see the signs then. Dave had speech and motor skills development delay. We thought it was just normal. He was not able to undergo newborn screening so his congenital disorder was not managed immediately," added Lorelie.

PKU is a rare metabolic disorder that causes severe irreversible mental and developmental disabilities if not managed from birth. A person with PKU cannot properly use one of the building blocks of protein called phenylalanine, which is present in most food. Excessive accumulation of phenylalanine in the blood causes brain damage. Individuals with PKU need a closely monitored diet including special phenylalanine-free formula from the point of diagnosis to get the necessary protein they need. PKU diet mostly consists of fruits and vegetables, and all portions are carefully measured.

It proved to be a challenge for the De Guzman family after learning that both Dave and Dana were afflicted with PKU.

"It became more challenging when they started schooling. We cannot always watch what they eat. We were worried so we informed their teachers of their condition. Dana is also now aware of her restricted diet."

Continued on page 3 . . .

Number of Babies Screened as of June 2017 **9,799,124**