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22nd Newborn Screening Convention

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Participants, speakers, and organizers of the 22nd Newborn Screening Convention at The Manila Hotel, Manila, October 08, 2024

featuring
OUR NBS CHAMPIONS

Arlene Abuan (second from left), mother of Fherbin, proudly shares the accomplishments of her child as they navigate the journey of managing Fherbin's condition, Galactosemia.

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National Newborn Screening Convention: Strengthening Resources and Outcomes for the Next Generation

"We are here today as persistent, as resourceful, and as resilient as the newborn screening program," said Dr. Eva Maria Cutiongco-de la Paz, executive director of the National Institutes of Health, University of the Philippines Manila, as she opened this year's highly anticipated two-day annual National Newborn Screening (NBS) Convention on October 7, 2024 at the Manila Hotel, 1 Rizal Park, Ermita, Manila.

Cutiongco-de la Paz recognized the small but significant beginnings that blossomed into a full-bloom national healthcare program and the most successful screening program today. Despite initial funding and logistics issues, the program has found success because of the different stakeholders. True to this year's theme "STRONG NBS: Strengthening the Resources and Outcomes for the Next Generation," the convention highlighted the humble beginnings of the program, the partnerships and strategies that strengthened its implementation, and how key program implementers are further improving the services provided to patients.

This year's convention was attended by over a thousand participants. Among them were experts, practitioners, and leaders from national government agencies, academic institutions, the private sector, and non-governmental organizations. It gathered managers, coordinators, and representatives from the

Dr. Ebner Bon G. Maceda, overall chair of the NBS Convention 2024, addresses queries from the participants during the open forum.

Board members and officers of the NSSPI award the Certificate of Appreciation to Senator Christopher Lawrence T. Go, keynote speaker, and chair of the Senate Committee on Health and Demography.

Centers for Health Development, Newborn Screening Continuity Clinics, Centers for Human Genetics Services, Newborn Screening Centers, and newborn screening facilities around the country. Patients were represented by mothers who recounted their experiences from the moment their children were diagnosed with a disorder up to up to the time when their child is exhibiting excellence in school or the workplace.

Dr. Ebner Bon G. Maceda, overall chair of the NBS Convention 2024, shared that this year's convention was committed to enhancing and improving health outcomes as the future of the children helps the stakeholders continue the program.

Senate Committee on Health and Demography Chair Senator Christopher Lawrence T Go, in his keynote speech, emphasized his commitment to public service and healthcare. He spoke about his initiatives and continuous efforts to enhance the healthcare system in the country. Introduced by Dr. Conchita Abarquez, head of the Newborn Screening Center Mindanao, Senator Go conveyed his appreciation for the chance to assist the Filipino people and noted the presence of participants from different regions along with notable medical figures who are the primary organizers of the NBS Convention in 2024.

Participants in the two-day event had the unique chance to learn from the experiences of the experts, health professionals in the communities, and affected families, and connect with other program implementers. The event also boasted an impressive lineup of speakers who delved into key topics relevant to powering up on current trends in newborn screening, upskilling on hemoglobinopathies, advancing newborn screening through genomics, revitalizing collaborative strategies for improved patient outcomes, strengthening roots and outcomes through updates and experiences, and energizing newborn programs toward new directions.

The first plenary session highlighted insights from National Scientist Dr. Carmencita D. Padilla, also one of the proponents of the newborn screening program in the Philippines. She walked the participants through the history of the Newborn Screening Program. Day 1 of the convention placed emphasis on the following topics: disease mapping; hemoglobinopathies, cystic fibrosis, and tyrosinemia type 1 disorders; genomic applications in newborn screening and management; and ethics of genomic newborn screening. The results of the following researchers were also presented: *Plasma Carnitine Levels of MSUD and Classical PKU on Protein Restricted Diets, Clinical and Biochemical Profiles of Patients Flagged for Increased Tyrosine via Expanded Newborn Screening,* and *Clinical, Biochemical and Genetic Profiles of Patients Screened with Fatty Acid Oxidation Disorders*.

DOH OIC Assistant Secretary Dr. Albert Francis E. Domingo discussed the state of NBS in Universal Health Care. He underlined the two considerations when doing newborn screening: 1) Will the newborn become well? and 2) How much money will be needed?

DOH OIC Assistant Secretary Dr. Albert Francis E. Domingo discusses the state of Philippine newborn screening in universal healthcare.

Dr. Barbra Charina V. Cavan, 2024 chair of the Convention Scientific Committee and chair of the Experts Committee on Metabolic Disorders of the Newborn Screening Reference Center, ended the convention by announcing that next year's theme will focus on outcomes, programs, progresses, and advances.

For the past 21 years, the National Newborn Screening Convention has been one of the major highlights in the celebration of the Newborn Screening Week held every first week of October. The convention is hosted by the Newborn Screening Society of the Philippines Inc. (NSSPI) in collaboration with the Newborn Screening Reference Center (NSRC)-National Institutes of Health Philippines, University of the Philippines Manila. The latter has grown to be a major partner since its founding in 2005, in providing continuing education for a range of health professionals, including nurses, doctors, midwives, medical technologists, nutritionists, and chemists.

For more information about newborn screening and related events, visit www.newbornscreening.ph.
VGMendoza

Voices of Strength: Mothers Championing Newborn Screening

A genetic disorder was detected in their children through newborn screening, allowing these mothers to take proactive steps toward treatment and management, transforming fear into hope

Three courageous mothers took to the stage on October 8, 2024 at the Manila Hotel their personal journeys of navigating the challenges of having children diagnosed with metabolic disorders. With the 22nd Newborn Screening Convention's theme "STRONG NBS: Strengthening the Resources and Outcomes for the Next Generation," their stories underscored the critical role that newborn screening played in saving their children's lives and ensuring that they grow up healthy, normal, and productive citizens of the community.

DISCOVERING THEIR CHILDREN'S DISORDER THROUGH NEWBORN SCREENING

"*Nang makatanggap po ako ng tawag mula sa Newborn Screening (Center) na diagnosed ng galactosemia ang anak ko, ang daming tanong na pumasok sa isip ko, 'Ano ba ang galactosemia? Ano ang mga pwede kong gawin?... Nasaan ang mga espesyalista na pwede kong lapitan?*" stated Arlene Abuan, mother of Fherbin who was confirmed to have galactosemia through newborn screening. Galactosemia is a rare genetic condition that prevents babies from processing galactose, a type of sugar found mainly in milk and other dairy products. If left untreated, children may develop cataracts, liver failure, developmental delay, and untimely death.

The fear and anxiety upon hearing the news of their child's rare disorder is not unique to Arlene. Julian Chelsea Galgana-Agito, a first-time mother with her daughter Athena, was overwhelmed after looking into her daughter's condition. "*The doctors told me that she is positive for GA 1... Nung nakauwi na kami, sinearch ko siya, What is glutaric aciduria type 1 (GA1)*" GA1 is a rare condition wherein the body cannot process certain amino acids that causes a harmful build-up of substances in the blood and urine. The list of complications that showed when Chelsea looked up GA1 on the internet dimmed the typically joyous moment of having her first baby.

While Fherbin and Athena were fortunate to receive newborn screening as part of the PhilHealth's newborn care package routinely done in thousands of health facilities today, extreme luck played at Janelle's side when her condition, congenital hypothyroidism (CH), was screened in 1996, a time when only 24 hospitals were offering newborn screening. Her mother, Mrs. Divine Barrameda, recalled that, "*When I found out that CH can lead to mental and physical retardation, I was both scared and worried. But at the same time, I am thankful that newborn screening was already available, and that I gave birth to one of the first 24 hospitals that offered NBS.*"

MANAGING THE DISORDER: CHALLENGES AND TRIUMPHS

Newborn screening is crucial in detecting genetic disorders in newborns so that they could receive appropriate care to avoid mental retardation and early death. However, all the efforts in catching the condition early could go to waste if parents or caregivers fail to comply with the treatment and dietary plan that the doctors prescribe. "*Mother knows best. But when it comes to health, I might have to rephrase it to 'Your doctor knows best,*" emphasized Divine as she discussed the treatment, challenges, and resolutions in managing Janelle's condition.

Janelle Barrameda, who was diagnosed with CH, shares with the audience why she thinks NBS is important.

The successful management of metabolic disorders relies heavily on the patient's dietary management and preventing triggers that could lead the patient into a crisis. Arlene took this to heart as she continually educated Fherbin on his condition, dietary restrictions, and possible complications. She recognized that community effort is needed in raising a child that requires special management, so she informed Fherbin's teachers, classmates, and friends about his condition. At the young age of 9, Fherbin displays understanding of his condition as he proactively reads into ingredients of food items while accompanying his mom for grocery shopping.

As the primary caregivers of children with metabolic disorders, these mothers are also the first to witness the milestones that their children achieved in their journey. Chelsea recalls the excitement and happiness she felt when she saw Athena being able to crawl, and eventually walk at 1.5 years old, something that she thought Athena would not be able to do because of GA1.

"My daughter excelled in her studies. She graduated salutatorian in elementary… She's a first-take-passer of the civil engineering licensure exam. She also actively participated in various extra-curricular activities in school and even in her work, bringing in lots of awards and recognition," shared Divine with hundreds of newborn screening practitioners in the convention.

Similarly, Fherbin displayed exemplary skills as his mother Arlene shared his outstanding performance in school, *"Yung makita ko yung anak ko na masigla, marunong bumasa at sumulat, okay na posa akin 'yon... Pero higit pa po yung binigay ng aking anak. Kasi simula po nung nursery hanggang Grade 3, siya po ay consistent honor student at siya po ay nanalo sa quiz bee competition in Science."*

A CALL TO ACTION

The mothers' stories were punctuated with themes of resilience, education, and the profound support they received from their communities and healthcare providers. Divine urged her fellow parents, "*magtiwala lang po tayo sa ating doktor at sundin ang kaniyang gabay hanggang dulo. Wag po natin palampasin ang pagkakataon upang mabigyan ng normal at magandang buhay ang ating anak. This is the greatest gift that we can offer to our child."*

Arlene echoed this statement, stating "*Nagpapasalamat po ako sa newborn screening dahil naging kasama ko sila sa pag-alaga, paggabay, at pagtulong sa pagpapalaki sa aking anak... Dati marami akong tanong pero ngayon ang tanong ko, 'Paano kung walang newbom screening, paano na kaya yung aking anak?'"*

Finally, Chelsea reminded parents to find beauty in small victories. She added "*Lumalaban siya (Athena) para sa akin, so wala akong nakikitang dahilan para hindi lumaban para sa kanya."* These mothers deserve the strong cheers and applause, respect for their strength and determination from the health practitioners. Their stories resonate beyond personal experiences; they illuminate a path for future generations, emphasizing that with newborn screening, education, and community support, children with congenital disorders cannot only survive but thrive.

Through the years, the coverage of the Philippine NBS Program has continually increased, reaching 96.9% of newborns screened in 2023. However, with limited funding and logistic constraints, the long-term care of patients remains a challenge to the NBS program which supports medication, food supplements, and consultation with specialists.

The collective call to action at the convention was clear: to strengthen newborn screening initiatives, support families, and foster a community where every child has the opportunity to reach his or her full potential. Together, these mothers have proven that hope, resilience, and love can indeed shape brighter futures for their children and inspire others facing similar challenges.

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Divine Barrameda encourages parents to trust their doctors in helping them to manage their child's condition.

Awarding of certificates to the three mothers who shared their NBS journey

Janelle Liean Javillo, who was diagnosed with Phenylketonuria, shares how she manages her disorder.

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