Health Ripples

The UP Manila Health and Life Magazine

Jan-Jun 2015 · Vol.1 · No.1

The UP Manila Health Ripples magazine is published bi-annually by the **UP Manila Information, Publication, and Public Affairs Office (IPPAO)**, 8th floor, Philippine General Hospital Central Block, Tel. nos. 554-8400 local 3842. Contributions are welcome. Please email to ippao@upm.edu.ph or send to the IPPAO. EDITOR: **Cynthia M. Villamor**; EDITORIAL CONSULTANT: **Dr. A' Erickson L. Berberabe**; **Vice Chancellor for Research and National Institutes of Health Executive Director Dr. Eva Maria Cutiongco-Dela Paz**; STAFFWRITERS: **Cynthia M. Villamor**, **Fedelynn M. Jemena**, **January R. Kanindot**, **Anne Loren Claire S. Santos**, **Charmaine Abing**; LAYOUT ARTIST: **January R. Kanindot**; PHOTOGRAPHER: **Joseph Bautista**; CIRCULATION: **Sigrid G. Cabiling**

Table of Contents

EDITOR'S NOTES:

Creating ripples of change in the health sector

Ripples are like actions of individuals and institutions that can have far-reaching effects on the community, nation, and society.

Over the years, UP Manila, as the national health university, has been creating ripples of positive and relevant changes in health that are contributing significantly to the development and achievement of national health goals and priorities for the Filipinos. The rippling effects are exemplified by the curricular, research, training, and community programs of the different colleges and the National Institutes of Health and patient care services of the Philippine General Hospital. These endeavors are done in partnership with other learning institutions, government entities, and other organizations to ensure greater excellence and relevance to the needs of the people. Through this bi-annual magazine, it is our wish to share with you the ripples in health that have been generated and still being generated by UP Manila that are benefiting the Filipinos. For new initiatives, we are hoping that through time, the ripples will create waves that will impact our health situation in significant ways. The magazine features incisive articles on the programs, projects, services, and individuals that make a positive difference and give pride and honor to the university.

For this maiden issue, the focus is on the role and importance of research and human genetics in the health and life of our country. We are honored to be graced by an article from no less than National Scientist, 2013 Ramon Magsaysay Awardee, University Professor Emeritus, and former UP Manila Chancellor Dr. Ernesto Domingo entitled "Reflections on UP as a University."

The issue includes other articles on the pioneering programs and institutions that were built in support for and promotion of human genetics, the groups and networks that were created to provide a clearer and deeper understanding of genetic disorders among Filipinos, the measures being taken to manage the conditions of such patients, and a glimpse of future endeavors in the field of genetics in the Philippines.

At the center of what human genetics has achieved is one of the country's top pediatricians and human geneticists who has been creating positive ripples of change in the health community with her achievements as a clinician, educator, researcher, leader, and administrator, Dr. Carmencita D. Padilla.

On the occasion of her investiture as UP Manila chancellor, we dedicate this issue.

Reflections on U.P. as a university

Dr. Ernesto O. Domingo
National Scientist, Ramon Magsaysay Awardee, and University Professor Emeritus
UP Manila College of Medicine

Dr. Ernesto Domingo is being conferred the Ramon Magsaysay Award by Supreme Court Chief Justice Lourdes Sereno in 2013.

The University of the Philippines System (UPS) is a vast network of autonomous universities located in the major islands of Luzon, Visayas and Mindanao. Since I have served only in the Health Sciences Center now appropriately called the University of the Philippines Manila (UPM), I will limit my reflection on this unit, not that I am unaware of the major changes in the system, but simply because the reflection is based on my personal knowledge and experience. After all, I have been Section Chief of Gastroenterology, Vice Chair of the Department of Medicine, Chair of the same department, Coordinator for Teaching and Research of the Philippine General Hospital (PGH), Associate Dean of the College of Medicine (CM), Acting Dean of the same College, Director of the Postgraduate School of Medicine and finally, Chancellor of UPM. With this almost inclusive academic-administrative positions held, I was not only a witness but a participant to the many significant events in the life of UPM. My continuing engagement with UPM spans six (6) decades as of this very moment, a period wherein

Dr. Ernesto O. Domingo, 3rd from left front row

UPS has had at least eleven (11) presidents and UPM at least nine (9) chancellors. Within this period, I, too, have changed not only in interest but also in outlook.

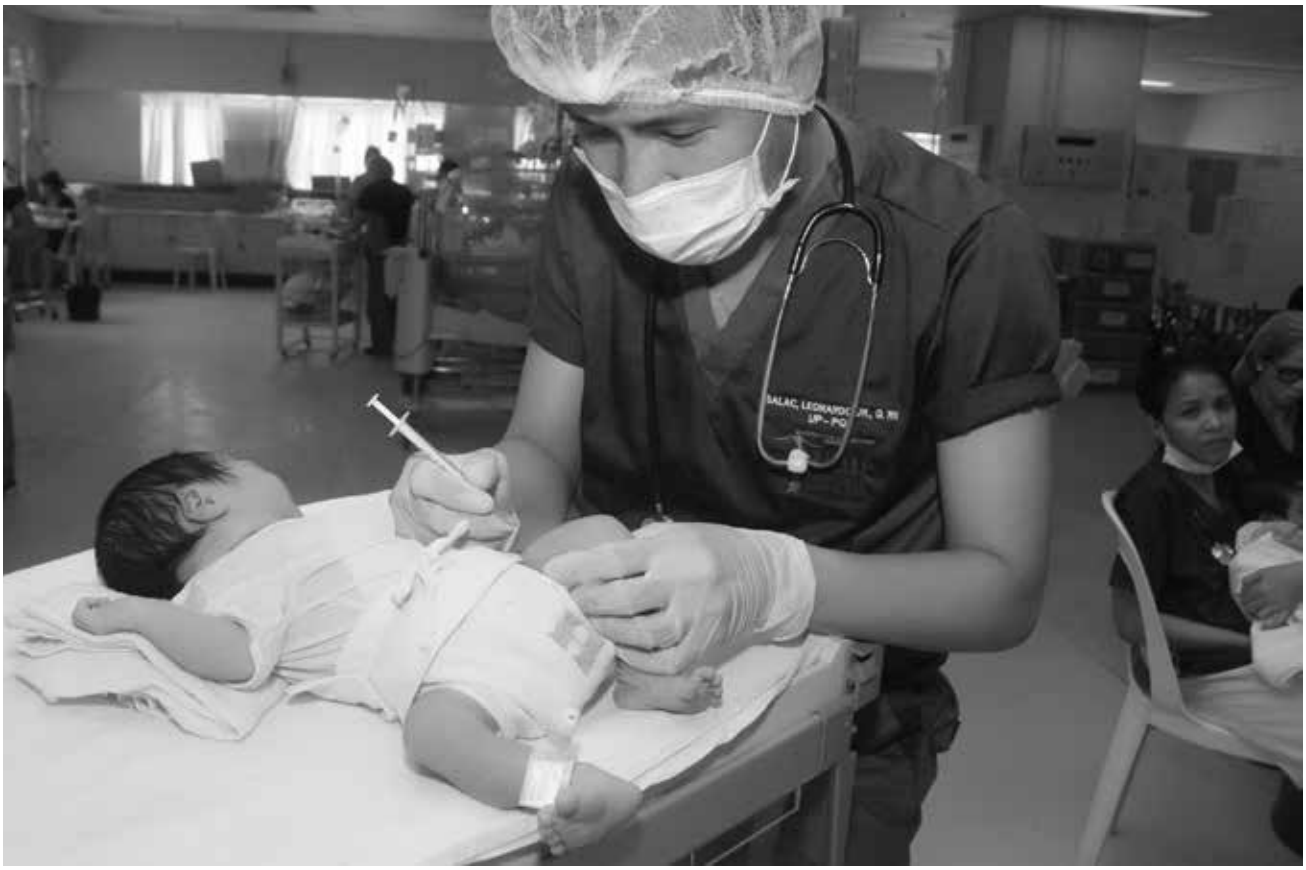
I have a wide choice of areas to dwell on in this reflection; UPM constituency which includes studentry, faculty, research and administrative personnel and non-academic support staff; the university's core functions like teaching and training, research and extension activities; academic administration, programs and processes, the added role of a national university like public service and advocacy. I will, however, choose research on this occasion and reserve at a future date a more comprehensive reflection on a wider topic.

Why do I choose research? First, the current administration of UPM is heavily focused on research what with the new National Institutes of Health building coming up soon. Second, at least half of my academic career in UPM was spent on research. Third, the incumbent Chancellor Carmencita D. Padilla is biased for research being an active excellent researcher and administrator. It also happens that research is an appropriate tool for reconciling excellence and relevance, a persistent dilemma in an institution of higher learning like ours. The reflection which I will subsequently narrate will serve to illustrate that research can be utilized to bridge the gap between excellence and relevance, should one exists.

In the early 70's, upon the prodding of a colleague, we organized the Liver Study Group (LSG) here in UPM. The motive force for the organization was the observation that we were admitting in the wards of the PGH an inordinate number of primary liver cancer (PLC) patients who all eventually died within months after falling ill. We did not know then what the main cause (etiology) was, therefore, we were not in any position to recommend preventive measures. This knowledge gap was confounded by the total absence of effective treatment modalities that were able to cure or extend the patient's life meaningfully.

Because of the paucity of knowledge regarding PLC, it was quite obvious to us that a comprehensive study of the diseases was needed. Such a study will need the collaboration of many investigators of various specialties, a requirement best served by a study group. Consequently, we established such a group and called it the Liver Study Group. LSG was composed of researchers coming from various disciplines as well as academic and research institutions outside UPM, and even outside the country like the United States of America and Japan. The organizational structure of the LSG as well as the working relationship were an ad hoc improvisation as there was no template for it. Whereas today, such a group as the LSG is very common in many biomedical researches, it was a novelty than as format for comprehensive research effort. I will return to this topic later.

To go ahead with the liver cancer research. Midway



A baby being immunized against Hepatitis B at the PGH Clinic

into the research on PLC, we found that the major cause of this malignancy in our country is a virus that attacks the liver principally, called hepatitis B. Incidentally, other viruses that infect the liver primarily (hepatotrophic) are designated by a letter of the alphabet, thus, hepatitis A, B, C, D, and E . Once we had this information, we shifted gears and launched another comprehensive study, this time on hepatitis B. The initial studies were epidemiologic in nature because we needed to understand, among others, the magnitude of this infection. Incidentally, acute hepatitis B has a very low mortality and morbidity compared with more common infectious diseases, hence, is not much of a public health importance, at least in terms of public awareness.

What is not easily recognized is that the infection results in a considerable number of infected people who proceed to become lifetime carriers of the virus, a state known as hepatitis B carrier. Presently, the prevalence of hepatitis B carrier is in the range of from 10 to 16 percent of the population, easily 10 to 16 million Filipinos. From this cohort of chronically infected people will emerge two dreaded diseases, PLC and cirrhosis.

From the national epidemiologic study of hepatitis B by the LSG, we came to know the following:

a) Its prevalence.
b) The incidence and attack rate.
c) How the infection is transmitted and which is the most important mode of transmission that results in chronic infection.
d) What the risk factors are.
e) Who are the subsets of the population that are at risk of getting infected.
f) What other liver diseases are caused or aggravated by hepatitis B.
g) What is the point in the transmission of the infection which can be targeted for interdiction with very high efficacy.
h) How to do a rapid epidemiologic assessment in the population regardless of geographical conditions, including the necessary technology required.
i) What are the modes of intervention and what are their comparative cost effectiveness, cost-benefit, feasibility and sustainability.

When all these epidemiologic information became available from the LSG study, quite fortuitously, a new vaccine against the virus was introduced commercially. The LSG got involved in testing the efficacy of the vaccine among Filipinos.

Throughout this period, from the inception of the epidemiologic study to the period when the vaccine became available, the Secretary of the Department of Health (DOH), was continually

informed of the progress in the research as well as new developments in the field. Eventually, he tasked the LSG, to develop for the DOH policy options for the control of the infection and inferentially eradication of PLC.

The LSG, using the data from its studies complied by giving the DOH Secretary policy options, including cost and effectiveness sensitivity studies of selective versus universal infant immunization. Because vaccine sufficiency was a major issue at that time, LSG proposed to the Secretary various approaches to solving the problem as a side study. The LSG evaluated the current capability of the DOH to give universal infant immunization, as is without adding resources and personnel.

We thought that the LSG's work (and responsibility) ended here. But no, it fell on our shoulders to campaign for the policy on universal infant immunization which we recommended. This meant that we, the researchers had to approach personalities, institutions, private business groups, local and international health agencies, line agencies of government, the legislature, as well as sundry groups to buy into the policy. It took decades of continuing efforts not only by the LSG but other groups later to secure the necessary support for the immunization program to be institutionalized.

It goes without saying that the advocacy part was difficult, frustrating, and sometimes irritating, but in reality, nobody told us it would be otherwise. The experience, however, highlights two lessons, if you will:

1) Scientific discovery and knowledge acquire more meaning if they result in some benefit to men.
2) Scientists and researchers may have to accept the possibility that they may step outside their comfort zone to bring to a desirable conclusion what they start out of curiosity.

While our experience in the LSG can be considered *sui-generis* at the time, it is standard practice now in our UPM research activity. Witness the few examples: newborn screening program, rare inherited diseases, aging, hearing loss during infancy, mental health, dental health, women's health, non-communicable diseases, primary health care, and many more I may not be aware of.

The conduit of research in the university has certainly metamorphosed from the simple pleasure of knowing to the more heroic imperative of ameliorating. While the search for the what, the how, and the why will endure in the university, every now and then, the question will be asked: For what and for whom? •

Dr. Ernesto O. Domingo with St. Paul University Manila students who attended his lecture as an RM Awardee.

Tackling greater challenges in the academe

by Cynthia M. Villamor

She brings to UP Manila more than 27 years of experience as health administrator, educator, researcher and advocate to transform the university as a policy hub and leader

Chancellor Carmencita Padilla is being sworn in by UP Pres. Alfredo Pascual in October 2014, witnessed by son, UP College of Medicine 2016 Student Patrick Jose Padilla.

Her assumption of the highest administrative and academic post at UP Manila could not have come at a more opportune time. After founding and leading several organizations and programs, her institution building and nurturing skills come to the fore as she steers the national health university towards being a major contributor to the country's health.

UP Manila Chancellor Carmencita Padilla cuts a "towering" figure in the health community and the academe. Beneath her slim, tall and exquisite frame are personal and professional qualities that have chalked up astounding achievements, particularly in children's health. She is dynamism personified, with an extraordinary drive and passion to achieve professional goals that she believes are contributing greatly to achieving better health for Filipinos.

She exemplifies her dedication and commitment to health improvement in several undertakings. She is a pediatrician, teacher, researcher, institution builder, policy setter, health leader and advocate, and, since November 2014, UP Manila chancellor.

Elected by the UP Board of Regents as UPM's ninth and third woman chancellor on September 29, 2014, Chancellor Padilla, Academician and University Scientist, sought the chancellorship on a vision of excellence, leadership, collegiality and service. During the forums for the search, she had emphasized the need for UP Manila to be the hub and leader of policies on health and development through its colleges and the National Institutes of Health (NIH).

"UP Manila will not be reactive to health issues but rather be pro-active through the generation of many policy statements that will assist

government and the private sector in the creative solution of health issues. UP Manila will gain the trust of society such that the public will always defer to UP Manila for valued opinion on health issues that affect Philippine society," she pointed out in her speech during the oath taking.

Her administration is now focused on every task that will contribute to the achievement of this goal. Cognizant that research is the main tool for relevant and sound policy making, an integrated program on research revitalization is being implemented. The program consists of infrastructure development, human resource capability building, strengthened publications and public affairs activities, operational efficiency, more and better research grants and incentives for the faculty and staff, encouraging student researchers and providing more forums for research presentations.

With bigger and better-designed offices and laboratories at the soon-to-rise NIH building, Chancellor Padilla foresees a major change in the research landscape at UP Manila. The new

Chancellor Carmencita Padilla with the members of her Management Team, from left, Vice Chancellor for Administration Dr. Arlene A. Samaniego, Vice Chancellor for Academic Affairs Dr. Nymia Pimentel-Simbulan, Vice Chancellor for Planning and Development Dr. Michael Tee, and Vice Chancellor for Research and NIH Executive Director Dr. Eva Marie Cutiongco-Dela Paz

Chancellor Padilla with the faculty, students and staff of the School of Health Sciences, Palo, Leyte during a visit in November 2014. Every medical student received a stethoscope, BP apparatus and "scrub suit'

facility will make research a more enjoyable and stimulating pursuit for faculty members and staff. Way before she even thought of leading UP Manila, Chancellor Padilla has been an active member of NIH and all the more when she became director of the NIH Institute of Human Genetics (IHG) and Newborn Screening Reference Center. She was actively involved in the planning and design of the 15-story building.

Apart from the NIH facility, Chancellor Padilla is working on plans for the renovation of the right wing administrative offices at the PGH Central Block 8th floor and the offices along Padre Faura Street, including the National Teachers Training Center for the Health Professions, the only academic unit without its own building.

The infrastructure beef-up is being matched by aggressive undertakings on capacity trainings, provision of more venues for publication and dissemination of researches and other scholarly works, maximization of information technologies and social media sites, and sustaining and building more academic partnerships.

With IHG, Chancellor Padilla demonstrated how a mere idea can be transformed into a significant program with hard work, perseverance, and a never-give-up attitude. She relates that IHG started with one staff from the UPCM, a microscope given by then UPM Chancellor Ernesto Domingo and a refrigrator given by then UPCM Dean Alfredo Ramirez. Today, it has grown and is considered the largest provider of genetic services in the Philippines *(please see separate article on page 10)*.

Each of these journeys at institution building tested her determination and patience. With every program initiated, she experienced challenges and rejections. But her resolve and faith in the nobility of her causes were far greater than any frustration and disappointment. Her firm belief that there is no problem that cannot be overcome when pushing for a quality and relevant program has kept her and her dreams afloat.

Constituents remember her as the nominee for UPM chancellor who responded on the issue of inadequate funds, thus: "Ayokong problemahin ang pera kasi magagawan yan ng paraan. Mas mahirap baguhin ang behavior at mindset ng tao." As UP Manila's top academic and administrative leader who has worked with the university for years and dealt closely with its system's strengths and weaknesses, Chancellor Padilla considers this a big challenge. This is

Chancellor Padilla welcomes the delegation from Teikyo University, Japan

why on her first day with the employees during the flag ceremony, she pounded on the need for culture change and gently pushed the staff towards having and living by a mindset of dedicated and excellent service Her first weeks were spent on addressing gaps in operational and academic efficiency and building trust in the system.

To ensure a cohesive and competent team, she chose the members of her administration judiciously based on academic qualifications, experience, competence, dedication to service and to her vision and goals for the university.

While other leaders would view lack of funds as an excuse for not pushing through with a plan or project, Chancellor Padilla sees this as a challenge. Apart from her charming persona and people skills reminiscent of her mentor, the late National Scientist and former chancellor, Dr. Perla Santos Ocampo, her resource generation ability is one of her strong suits. This is borne of her excellent record of achievements, performance, and goodwill earned from partner institutions and her knack for demonstrating the importance of program planning and strategizing its implementation, and delivering results as expected.

When she became chancellor, part of her vision was to strengthen the School of Health Sciences. She is the only chancellor who visited all three School of Health Sciences (SHS) campuses within two months of her term. The visit to SHS Palo was made two weeks into her term to witness firsthand the developments in the school after the damages wrought by Typhoon Yolanda and discuss with the constituents a rehabilitation program.

On May 7, 2015, Chancellor Padilla went back to SHS Palo to unveil the marker and launch the restoration of the Dr. Florentino Herrera Academic Buidling. Once completed, students, faculty, and staff will transfer there after enduring the difficulties of holding classes and offices in prefabricated tents that have been damaged by strong rains and typhoons during the past one a half years. It was an urgent decision by the administration that meant deferring a plan for the relocation of the School to a permanent site that will take several years. The trip to the other SHS campuses in Baler, Aurora and Koronadal, South Cotabato were made a few weeks after as part of her administration's efforts to strengthen the operations of the two extension campuses.

Chancellor Padilla recently led the conduct of a Strategic Planning Workshop to revisit the vision, goals, and programs of the SHS to increase their relevance to the needs of more Filipinos, particularly those in remote and underserved communities.

One of the issues that hounded Chancellor Padilla during the search was her ability to

Chancellor Padilla congratulates the new officers of the UPM University Student Council after their oath taking and dialogue.

straddle the demands of the chancellorship with the rest of her responsibilities. Used to multi-tasking, Chancellor Padilla has developed a remarkable work ethic in that she has learned to focus on several tasks at the same time, without one being sacrificed for the other. Because her roles and duties complement each other, she sees this as more of an advantage than a disadvantage. Her leadership in one builds on the other. Whatever accomplishments made and goodwill earned in one continues to rub off on others. Her multi-institutional involvement has been serving UP Manila in good stead.

Chancellor Padilla admits being fortunate to have been mentored by several health professionals, notably those from her department – the Department of Pediatrics led by Dr. Perla Santos Ocampo. In her inaugural speech, she acknowledged and thanked her mentors at the UPPGH Dept of Pediatrics where her leadership skills were honed. The Department stands proud in having three pediatricians as chancellors of UP Manila – out of 9 chancellors -- PSO, Professor Emeritus Dr. Ramon Arcadio and Dr. Padilla.

"I give credit to our mentors in the department who provide not only academic guidance but also ensure that all the graduates are leaders for the future. I consider them my gentle pushers. All of them continue to be my conscience, constantly reminding me of my responsibility to my home university campus, UP Manila."

Chancellor Padilla brings to UP Manila almost 30 years of experience and training and a high level of dedication as faculty member of the Department of Pediatrics and as administrator in various capacities – as Director of the Ugnayan ng Pahinungod, as Chair of the PGH Department of Pediatrics; as IHG and NSRC Director; and as PGC Executive Director that brings together UP Diliman, UP Manila and UP Los Baños to solve critical health and food security issues in the Philippines.

All of these endeavors have prepared her for the rigors and challenges of leading the national health university from which the country is expecting a lot and pinning their hopes on in improving the national health situation.

Through pioneering and relevant courses that are attuned to national needs and realities, evidence-based and policy setting researches, and community service programs that reach out to the larger society, UP Manila has set the path for Chancellor Padilla and her management team, with the support of the deans and directors, the administrative heads, and the rest of the constituents, to transform UP Manila as a stronger and leading institution and change agent for health. •

Pioneering and leading genetic services for the Filipinos' health

by Cynthia M. Villamor

After 25 years, the Institute of Human Genetics has grown and is now recognized as one of the leading genetic resource centers in Asia and the largest provider of genetic services in the Philippines.

All the inherited things that set us alike or apart, including our predisposition to certain illnesses and how these can be managed to ensure optimum health is basically what human genetics is about. In the Philippines, it is taking more time for this science to be understood and its role in health recognized and applied. At UP Manila, the unit's precursor started 25 years ago with practically nothing.

Call it the "genetics" factor because of its siblings' similar success stories, but the Institute of Human Genetics (IHG) illustrates that an institution can literally be born from scratch and grow to become one of the more established and stronger entities in health. The siblings being referred to are the IHG and the Newborn Screening Reference Center (NSRC), both component institutes of NIH and both founded by UPM Chancellor and human geneticist Dr. Carmencita Padilla.

Historical Milestones

IHG started in 1990 as a small Medical Genetics Unit (MGU) of the UP College of Medicine under the Office of the Dean in response to the need for genetic services in the country. Initially housed at the Department of Pediatrics, the Dysmorphology Clinic (later renamed the Clinical Genetics Clinic) was opened at the PGH Outpatient Department in 1991.

Before this period, in the 1980s, genetic services were provided by, apart from the PGH Department of Pediatrics, the PGH Department of Obstetrics and Gynecology, the Department of Internal Medicine and the University of the East–Ramon Magsaysay Memorial Medical Center Department of Pathology and Microbiology. The field of genetics slowed down with the untimely demise of two geneticists and the retirement of another, according to a published article of Dr. Carmencita Padilla and Dr. Eva Cutiongco Dela Paz titled "Genetic testing and services in the Philippines" at the Journal of Community Genetics.

In 1990, coming back from training at the Royal College of Alexandria in Sydney, Australia, Dr. Padilla started a cytogenetics laboratory even with inadequate funds, staff, and equipment. All she had were a microscope, a refrigerator, one medical technologist, a table and a time slot for charity genetic patients at the Philippine General Hospital. Add to this an initial grant of P500,000 from the Philippine Council for Health Research and Development and P1 million from UP Manila under then Chancellor Dr. Perla Santos Ocampo in 1996. The Newborn Screening (NBS) Laboratory was opened in 1997.

When the National Institutes of Health (NIH) was established at UP Manila in 1998, all laboratory services were moved to NIH while the clinical services (in-patient and out-patient) remained at the PGH Department of Pediatrics. In 1999, IHG was established as one of the component institutes of the NIH. The IHG was tasked with promoting health by understanding the genetic basis of common diseases among Filipinos through research and services that will allow definitive diagnosis and early detection of potentially treatable genetic conditions and implementing educational programs to increase awareness of physicians and the general public on common genetic disorders. The Molecular Genetics Laboratory and the Biochemical Genetics Laboratory were opened in 1999 and 2001, respectively, as part of the IHG's expansion.

IHG Now

Today, IHG is recognized as one of the leading genetic resource centers in Asia and the largest provider of genetic services in the Philippines.

Institute of Human Genetics Director Dr. Mary Anne Chiong

"Most of our tests and services are not available in other facilities," says IHG Director Dr. Mary Anne Chiong. "The challenge would be how to stay abreast. We may be the largest genetic facility now but we need to expand or level up soon, so we can strengthen our position in the field of health."

IHG houses the Clinical Genetics Unit and the above laboratories. The Clinical Genetics Unit takes care of the diagnostic and therapeutic needs of patients with metabolic disorders. The Biochemical Genetics Unit provides diagnostic testing and physician-assisted consultative services to clinicians to promote adequate diagnosis and management of inherited metabolic disorders. The Molecular Genetics Unit provides an understanding of the molecular and genetic etiology of a range of single gene disorders, syndromes and complex traits in humans, develops new tests for diagnosis and monitoring of such conditions and translates research into improved care for children, adults, and families.

Of all the genetic services being rendered by the IHG, newborn screening is, by far, the most widely known and recognized. Researches conducted by Dr. Padilla and other pediatricians and geneticists in the 1990s generated the results that led to the setting of the policy on NBS years later. Republic Act 9288 or the Newborn Screening Act of 2004 was enacted, institutionalizing the National Newborn Screening Program in the Philippines. Dr. Chiong says that the IHG has been

the brewing cauldron of the Newborn Screening Program in the Philippines *(please see separate article on page 15)*.

Apart from the NBS program, the IHG has been promoting the field of genetics nationwide and in the world through its service programs, publications, and other advocacies. The Volunteer Youth Leaders for Health-Philippines, for one, was established and is continuously being supported by IHG *(please see separate article on page 25)*. IHG has also been providing counseling to families affected by genetic disorders. IHG faculty and fellows conduct lectures and orientations on different genetic conditions.

"We want to be recognized so that we can forge more partnerships and garner more support for our projects and programs. We want to bridge other service and product providers to our patients and beneficiaries, Dr. Chiong said.

To address the need for more genetic experts and counselors, particularly for regions and provinces, the Master of Science in Genetic Counseling (MSGC) was instituted during the term of Dr. Padilla with all geneticists at the Institute serving as lecturers and clinic preceptors. Dr. Chiong declares that the MSGC program will soon produce the first batch of genetic counselors who will help in promoting the field of genetics by educating the public and counseling affected families.

Advocacy Programs

In collaboration with the government and other health institutions and organizations, IHG participates actively in information campaigns to create a better understanding of genetic disorders and proper referral of patients with genetic conditions. It organizes workshops and hosts meetings for local and international participants.

The Institute assists in setting up support groups for families with members suffering from genetic disorders. It participates actively in the continuing education activities and medical missions being conducted by the Down Syndrome Association of the Philippines and the Philippine Society of Orphan Disoders, Inc.. A recent collaboration is the event dubbed "I Am Rare," *(please see separate article on page 20)* an awareness campaign that aimed to celebrate the strengths and struggles of children with rare disorders.

Research as Core

Without research, IHG would not have been here today and will not be sustainable in the future. According to Dr. Chiong, the IHG's Molecular Genetics Unit conducts researches in collaboration with both local and international clinicians and scientists on the genetics of monogenic and complex genetic conditions in the Filipino population, such as asthma,

Clinic for children with genetic disorders at the PGH Outpatient Department

cancer, cardiovascular disease, diabetes mellitus, thalassemias and metabolic disorders. Using molecular genetic techniques and researches, new molecular tools are applied for conditions seen in the clinics to earlier detect and manage them and to better understand how these genes cause disease.

She explained that the projects are closely linked to the Philippine Genome Center. For tests that are not available in the country, the IHG-NIH facilitates the DNA extraction and the transport of samples to overseas laboratories. Pre-genetic testing and post-genetic testing counseling are provided by the geneticists. Meanwhile, the Microarray Core Facility is involved in major genetic initiatives. Among its active roles in the investigation of various diseases, the facility has been in the forefront of researches on diabetes, cardiovascular disease and population research on the indigenous people of the Philippines.

"Recognizing that diseases of the heart and blood vessels and diabetes and its complications are prevalent and pose threats to Filipinos from all walks of life, studies are looking into creating novel ways of detecting these conditions through genetic testing. A genetic test would allow us to identify genetic markers that are present in those with strong predisposition for disease giving them more incentive to adjust their lifestyle and delaying, if not completely preventing, the diseases. These studies further hope to explain the occurrence of these diseases, which may help in the diagnosis and treatment of these conditions," Dr. Chiong explained.

The IHG director further said that the laboratory provides the necessary tools in the genetic characterization of indigenous groups in the Philippines. The study is part of regional initiative call Pan-Asian SNP that aims to investigate the interrelationships of Asian populations, explain movements, diversity, and, in the larger sense, their commonality. The study hopes to determine the very essence of being Asian, and in particular, of being Filipino.

As an offshoot of its research efforts, a bill dubbed as the "Rare Disease Bill" is pending in Congress.

Contributions recognized

In August 2010, IHG was conferred the PCHRD Outstanding Health Research Award for its contributions to the development of the field of human genetics in the country. The introduction and institutionalization of NBS and genetic services and research have made major breakthroughs that led to the early diagnosis and treatment of babies with serious inherited conditions. Also recognized was IHG's role in making genetic testing available and accessible to Filipinos.

Apart from this institutional award, IHG faculty members continue to reap honors from local and international bodies for their respective contributions to the development of human

genetics in the Philippines and the services provided to patients. Chancellor Padilla, IHG former director, has been conferred the rank of Academician by the National Academy of Science and Technology and was a recipient of the The Outstanding Young Men (TOYM) in 1994, The Outstanding Filipino Physician in 2007, Outstanding Pediatrician in 2013, one of the 100 Most Influential Filipino Women in the World in 2013 and the UPAA Distinguished Award for Health Education in 2014, among several awards.

Another former director, Dr. Eva Cutiongco Dela Paz, now UPM Vice Chancellor for Research and NIH Executive Director has been conferred the Dr. Jose Rizal Memorial Award in 2012 and the UPM Outstanding Researcher award and Research Productivity Award in 2011.

Future Projects

IHG has vowed to constantly seek technology and innovations to improve facilities and services. "We are continuously developing and building the capacity of our human resource in terms of skills, knowledge, attitude, and behavior. We are also planning to do some serious social marketing for the services because only the Newborn Screening Program is more widely known among our genetics services. We want to be recognized so that we can forge more partnerships and garner more support for our projects and programs. We want to bridge other service and product providers to our patients and beneficiaries," Dr. Chiong pointed out.

The soon to rise new NIH building will provide more opportunities and bigger challenges for the IHG researchers and staff. "The new NIH building will put all our units together for a faster, more efficient and smoother coordination and transaction. We will be occupying several floors in the building as we are currently the largest institute under NIH. It will have everything we need—from space requirement, technology, equipment, work environment, design and layout, and functions. What we are doing now is to address funding and sustainability issues because the maintenance of the new building will be more expensive," the IHG director added.

At present, IHG occupies most of the 2nd foor and 3rd floor attic of the decades-old NIH building. Because of lack of space, the Newborn Screening Center was moved to the UP-Ayala Technohub in 2013. More than 30 of the IHG staff are located in this temporary facility. They will return to NIH when the new NIH building is completed. The Newborn Screening Facility services 1,500 health facilities of the National Capital Region, Region 4B and Region 5.

The Institute is currently preparing for ISO certification in June this year, hoping to maintain the good quality of services, systems and processes that have been established during the past years. Dr. Chiong revealed the three priorities for the strategic thrusts under her term: Staff Development, Publications, and Systems and Processes.

"IHG will continue to develop the growth of the backbone of the organization—the human resource. It will intensify its research efforts and translate them to concrete outputs like publications, copyrights and/or patents, national policy, or other high impact services and programs. Lastly, it will institutionalize systems and processes that will enable the institute to become more accessible to its stakeholders and maintain high quality service and delivery so that the scope will be wider, and more and more patients will be reached by our genetic services," she stated.

Riding on a high level of confidence, support, and recognition borne by an outstanding track record of achievements and contributions in health and with a lot to look forward to, the IHG is certainly more primed to continue its mission of saving patients with rare diseases and genetic disorders and ensuring them and their families of healthier and productive lives now and in the future. •

The PGH OPD clinic for children with genetic disorders is open on Mondays, Wednesdays and Fridays, 9-12 noon.

Ensuring healthy citizens through newborn screening and management

by Charmaine Abing

Newborn Screening has grown from 24 participating health facilities in 1996 to the current and still growing 5,700, saved more than 120,000+ babies from mental retardation and death, and continues to save more babies each day.

Imagine what would happen to over 33,000 Filipino newborns at risk for mental retardation and death every year if newborn screening were still not available today. Parents would think that their healthy-looking babies would grow to be really healthy and well. Doctors would find it difficult to diagnose babies and children born with metabolic and genetic disorders and by the time they had done so, life-long irreversible conditions may have set in, or in worst cases, death may even have snatched them away.

In January 1981, Jenny (not her real name) grew up to be healthy and athletic although born with an enlarged clitoris and often feeling that there was something different about her; but she cast those thoughts aside. During high school and college, she could no longer avoid noticing the changes in her body. Her breast never developed and not once did she experience having menstruation. Her facial features, physical appearance, voice, external genitalia and even preference for the same sex were clearly manifestations of a male person. Jenny looked like a guy with long hair, often making her inconvenient and embarrassed.

Left: Genevieve, 22-years-old, Newborn Screening not done at birth. She developed mental retardation due to delayed diagnosis of MSUD with no NBS done at birth. Right: Andrew, 2-years-old, underwent Newborn Screening and was diagnosed and treated early from MSUD. He is developing normally.

Notwithstanding her odd situation, Jenny graduated from college and was hired as a teacher. One day, at the college library, she read about and researched on Congenital Adrenal Hyperplasia (CAH).

Newborn Screening procedure done in the first week of life. Using heel-prick method, drops of blood are collected in a filter card. Blood samples are then tested for the risk of genetic conditions.

At age 22, Jenny consulted an endocrinologist at the Philippine General Hospital (PGH) who diagnosed her with simple virilizing CAH. After consulting with some specialists, she decided to petition the Court to change her name in her birth certificate, public, and school records from Jenny to Jeff and her gender from female to male. The court granted Jeff's petition and the case was dubbed as the very first of its kind to succeed in the Philippines.

CAH is a genetic disorder that involves excessive or deficient production of sex hormones and can alter development of primary or secondary sex characteristics in some affected infants, children, or adults. There are many types of CAH and in one type, CAH leads to salt loss leading to dehydration and death of infants by 7-14 days of life.

In 1993, Genevieve was born. It was too late before her doctors took notice of the sweet scent of her urine and suspected she was afflicted with MSUD. Genevieve was already 11 days comatose. MSUD or Maple Syrup Urine Disease is a genetic metabolic disorder resulting from the defective activity of the enzyme branched chain alpha-keto-acid dehydrogenase complex, preventing the body to breakdown amino acids in protein such as, leucine, isoleucine and valine. Accumulation of these amino acids becomes toxic to the brain. Newborns with MSUD have sweet smelling urine (similar to burnt sugar), like the odor of maple syrup from where its name is derived. If left untreated, they may have convulsions, be in coma or may die within the first few weeks of life. Treatment is mainly dietary in nature by limiting intake of protein.

Now Genevieve is 22-years-old, and though she survived this ordeal, her disease left her crippled and in delayed mental state, while, Jeff spent a big part of his life struggling with identity crisis and social and legal difficulties. Genevieve and Jeff could have lived more like other children and adults if their conditions were diagnosed early on in their lives.

Advent of NBS Program

Hope came to newborns at risk from genetic disorders in the form of the Newborn Screening in 1996. Tragic stories became inspiring testimonials of survival and triumph.

In 1997, John Paul was born and was one of the newborns to be tested by newborn screening. Ten days after, his results came back positive for CAH, thus the reason for his coal-black skin and his enlarged clitoris mistaken for a penis. After chromosomal test, John Paul was proved to be a baby girl and became Joanna Paula. Unlike Jeff Cagandahan and other babies diagnosed late with CAH, Joanna Paula was fortunate to be tested by newborn screening and given medical attention before any complications occurred. She has shed her coal-black skin as a result of her medications and her penile-like genitalia was managed down to size by reconstructive surgery. Joanna Paula was given a chance to live a normal life.

In 2011, Jay-R, was born to Remy, not their real names, and one of the saved babies through newborn screening. He was diagnosed with Phenylketonuria (PKU), a genetic disorder characterized by abnormal levels of phenylalanine in blood. Phenylalanine is an amino acid naturally present in all kinds of dietary protein. Increased phenylalanine levels accumulate in the blood and are toxic to the brain leading to intellectual disability, seizures, and other serious medical problems. PKU remains incurable and may only be managed by dietary

limitation of phenylalanine. Remy recalls her struggles with knowing the specific amount of protein in each kind of food given to her son and religiously monitoring his phenylalanine level and watching closely the amount of protein he should eat daily, that is 6.5 grams divided in a day. She would text the nutritionist to check on the correct amount of protein. The special formula are expensive but our patients are fortunate that it is provided free to the patients.

All MSUD and PKU milk formula are currently being donated by Mead Johnson (MJ) as part of their corporate social responsibility. Since 1992 when the first MSUD patient was diagnosed at the Philippine Children Medical Center, MJ has remained a staunch partner of geneticists in saving lives through the provision of the life-saving milk formula.

It was December 2012 in Davao City when Matilda was born; her parents Joey and Ams Macaraeg were the happiest, finding her a beautiful and healthy baby. However, with newborn screening, Matilda was diagnosed with Glucose-6-phosphate dehydrogenase deficiency or G6PDD, a genetic condition characterized by an enzyme defect that leads to red blood cell destruction in response to a number of triggers, such as certain foods and medication. Newborn screening provided the opportunity for Ams to manage her daughter's condition, and was thankful for it. "During my time, NBS wasn't available yet. A lot of people did not have the chance to be properly screened and treated. NBS is such a great advancement in the field of medicine that gives us the chance to be provided with intervention at a very early age to be able to avoid possible effects later on in life, " added Ams.

Kristine Joy, mother of two, was introduced to MSUD when the newborn screening result of her second child, Andrew, was released. After diagnosing Andrew's condition, Dr. Sylvia Estrada, a pediatric endocrinologist at the Manila Doctors Hospital, explained to Kristine what MSUD is. Two geneticists, Dr. Mary Anne Chiong and Dr. Melissa Baluyot from the Institute of Human Genetics, helped Kristine understand the management of patients afflicted with the disease. When newborns have MSUD, they have to follow a low protein diet and drink medical formula that does not contain the amino acids that will harm them. Patients should also undergo regular check-ups and have regular blood tests to measure amino acid levels. There are a lot of responsibilities when a child has MSUD, so Kristine oriented the whole family to understand the disease and be careful about what they feed Andrew. Unlike Genevieve, who suffers the same condition, two years and two months later, Andrew appears normal and healthy and seems to be unaffected.

Today, Jay-R, Joanna Paula, Matilda, and Andrew like other children their age, are living healthy lives. Their parents were thankful that NBS was already here when their children were born. Their disorders were detected early and interventions given on time that prevented complications.

NSRC as Implementor of NBS

The Newborn Screening Reference Center (NSRC) created under RA 9288 oversees the implementation of NBS in the country and is responsible for the national testing database and case registries, training, technical assistance and continuing education for laboratory staff in all newborn screening centers. NSRC Director Dr. Maria Melanie Liberty Alcausin reported that at the end of 2014, there were more than 5,700 health facilities in the Philippines that provide newborn screening.

She stated that efforts are being intensified to increase further NBS coverage in the country. As of December 2014, the coverage has increased to 65% from 57% in 2013. The Center is planning to put up more NBS centers that will provide all required laboratory tests in Northern Luzon, Northern Mindanao and the Visayas. Currently, there are five operational NSCs in the country—one each in Manila, Central Luzon, Southern Luzon, Visayas, and Mindanao.

Ams Macaraeg and daughter Matilda, who underwent Newborn Screening at birth and diagnosed with G6PDD. She is developing normally.

There are 14 continuity clinics currently operational in the country, which are responsible for referrals to specialists, such as pediatric hematologists, endocrinologists, and geneticists, as well as, referrals to nutritionists, social workers, etc. However, there are only eight practicing medical geneticists in the country. To address the shortage, Dr. Alcausin said measures, such as the use of the Telegenetics Referral System that enables patients from remote areas to have real-time consultations with specialists using the Internet, are being taken. In response to the shortage of geneticists, an MS Genetic Counseling is currently offered *(please see sidebar on page 19)* by the Department of Pediatrics. The genetic counselors will assist the geneticists in the counseling of patients with genetic disorders.

In late December 2014, the Expanded Newborn Screening was implemented increasing the coverage of disorders from 6 to 28. Prior to this time, the NBS program screens only Congenital Hypothyroidism (CH), CAH, PKU, Galactosemia (GAL), G6PDD, and MSUD. The expansion was spurred by data taken from the California Newborn Screening Program from 2005 to 2009 that showed that Filipino newborns in California were at risk for more than 20 other disorders and confirmed for some of the rare disorders. The disorders added in the Expanded NBS fall under various groups of conditions, such as hemoglobinopathies, disorders of amino acid and organic acid metabolism, disorders of biotin metabolism, and cystic fibrosis.

Dr. Alcausin explains that NSRC is still in the initial stages of implementing the Expanded NBS in the country. Apart from its full implementation and achieving 100% coverage, future goals of NSRC include capability building to ensure adequate supply of geneticists, endocrinologists, genetic counselors and other specialists. NSRC hopes to field its MS Genetic Counseling graduates to the newly formed continuity clinics.

Beginnings and the Future

Today, the Newborn Screening Program has saved more than 120,000+ babies from mental retardation and death and continues to save more babies each day. It has grown from 24 participating hospitals and other health facilities in 1996 to the current 5,700 and still growing.

In November 1995, Dr. Padilla met with Dr. Carmelita Domingo and decided to work on the NBS project by inviting 75 hospitals in Metro Manila. Only 28 responded and eventually

Dr. Carmencita D. Padilla (left) and Dr. Ephriam Neal Orteza pay tribute to Dr. Rowena Pua (center) for serving as NSSPI President for 11 years.

24 hospitals joined the pilot project in 1996. By 1997, Dr. Padilla set up the first newborn screening laboratory in Manila. And in two years, Dr. Padilla and her team was able to gather data from the research. Coordinators from the 24 hospitals decided to expand the program and in 2003, NBS was already being implemented in 323 hospitals.

The difficulty of integrating NBS in the public health delivery system compelled Dr. Padilla to pursue the Master of Arts in Health Policy Studies (MAHPS) at the College of Public Health in UP Manila. This gave her the skills in preparing for the Newborn Screening Bill. In April 2004, after intense lobbying with both Senate and Congress. Republic Act 9288, also known as the Newborn Screening Act of 2004, was finally passed. The law is considered one of the fastest to be passed in the history of legislation in the Philippines.

Even after the enactment of RA 9288, Dr. Padilla continued nurturing the growth and development of NBS, her brainchild. She became a technical partner in the implementation of the national program. With her continued lobbying and vigilance, the NBS program is now 100% funded by the PhilHealth while her research efforts paved the way for the expanded program.

At present, apart from being UP Manila chancellor, she is devoting time on the promotion of and pushing for the institutionalization of the "Rare Disease Act"—a complementary bill of the Newborn Screening Act of 2004.

The Newborn Screening Society of the Philippines, Inc. remains the major advocacy group of the newborn screening program. Its major activity is the Annual Newborn Screening Convention, which is attended by more than 1000 NBS practitioners - physicians, midwifes, nurses, policy makers, etc.

The Newborn Screening journey continues. There are a lot of things needed to sustain, improve and make the NBS services available to all future newborns. There are many stories to tell and many lessons yet to be learned. But the most important thing is, NBS is here to save lives and make these lives healthier and more meaningful. •

Master of Science in Genetic Counseling (MSGC)

by Charmaine Abing

The lack of medical geneticists prompted the need for genetic counselors in the country. In 2009, Dr. Carmencita Padilla, UP-Manila Chancellor, and Mercy Laurino, a genetic counselor from the University of Washington, USA, and a DOST-Balik Scientist Program Awardee collaborated in developing the Philippines' first genetic counseling program. The UP Board of Regents approved M.S. Genetic Counseling program in January 2011.

M.S. Genetic Counseling (MSGC) is a two-year program, training students to have in-depth knowledge in various genetic disorders, their clinical management, proper guidance and psychological support to the patients and their families. The Department of Science and Technology (DOST) provides scholarship grants. Graduates will have the opportunity to be part of the management team of Newborn Screening Program.

Ms. Laurino notes that since the inception of the program, MSGC students had published three researches in international journals, spearheaded the establishment of the Professional Genetic Counselors Society in Asia, and provided numerous genetic counseling to PGH and other specialty clinics as part of the student's clinical experience. MSGC will have its first set of graduates in June 2015.

"As we grow, our MSGC graduates intend to serve every hospital in the country and provide quality genetic counseling services. We will partner with our health care providers so that our Filipino patients will receive accurate information for informed decision making, while honoring their social and religious beliefs", said Ms. Laurino.

Apart from the MSGC, other activities are being undertaken to heighten understanding and recognition of the role and importance of human genetics. These include information campaigns, seminar-workshops, and hosting meetings for local and international participants. •

Care for 'rare'

by Anne Loren Claire A. Santos

The enactment of the Rare Disease Bill, an all-inclusive act that will address the needs of persons stricken with orphan disorders, would be a great encouragement and major lift to all orphan disorder patients and their families.

'Rare' is a word that denotes excellence, preciousness, and exception; but when it comes to diseases, rare takes on a different meaning. Rare diseases suggest a great level of suffering and misery. They are infrequent and affect only a tiny percentage of the population. In most cases, they are chronic, progressive, deteriorating, disabling, and capable of causing death.

Rare diseases, also called orphan diseases, bear different definitions from one country to another. In the United States, a disease is considered rare when it has fewer than 200,000 affected individuals. In Japan, a rare disease is affecting less than five per 10,000 individuals. In Europe, a disorder is rare if it affects one in every 2,000 people.

There are about 6,000 – 8,000 different types of rare diseases in the world, and the number continues to grow with new disorders being discovered each day. Of the known rare diseases, 80% have identified genetic origins. Therefore, in essence, even if symptoms do not manifest right away, a rare disease is present throughout an individual's life. The children are the most affected by rare diseases. Seventy five percent of orphan disorders afflict children. Moreover, 30% of children with rare diseases succumb before they turn five years old.

Other orphan disorders are caused by bacterial or viral infections, allergies, and environmental origins, for instance diet or smoking. Such environmental factors may cause rare diseases point-blank, or may interact with genetic determinants to bring about or advance the disease's sternness.

According to EveryLife Foundation for Rare Diseases, the approved drug treatments for the 7,000 known orphan disorders at present are only less than 400. This means that only 5% of the rare diseases have a certified and approved treatment.

Struggles in the local setting

A disease is regarded rare in the Philippines if it has a prevalence of at most one in every 20,000 Filipinos. The UP Manila – National Institutes of Health's Institute of Human Genetics (IHG) has recorded the names of almost 500 patients with metabolic rare diseases as of April 2015. Meanwhile, the Philippine Society for Orphan

UPM Chancellor Carmencita D. Padilla and PSOD President Cynthia Magdaraog join the VYLH volunteers, rare disease advocates, patients afflicted with orphan disorder and their families in raising hands for 'rare'.

Disorders (PSOD) has a record of 238 patients in their registry. However, it is noteworthy to state that their registries do not include those patients who have rare diseases that remain undetected, patients who are seen by private doctors, and those who wish to stay unidentified for fear of stigma.

At present, the most common rare disease in the country is the Maple Syrup Urine Disease (MSUD) with 145 patients based on the IHG registry.

However, just as the diseases are called 'orphan,' the drugs and healthcare products used to treat these disorders have also been referred to as 'orphan drugs.' The pharmaceutical companies have given very meager consideration in the development and production of drugs for use as medication for these diseases because of the expensive cost of production, which is due to the unavailability of considerable demand in the market. Also, since these diseases affect only a tiny percentage of the population, research groups and institutions have placed little involvement in understanding and finding out more about these disorders.

The available treatments and food supplements, which require lifetime administration, have to be imported from abroad and are very costly. For a regular Filipino family with a member suffering from a rare disease, the treatment may not be afforded or sustained.

According to Dr. Mary Ann Abacan, a metabolic specialist and clinical geneticist at the IHG, the way patients get treatment is through the kindness of other people. There are foreign and local benefactors – individuals or companies – who provide the needed treatment to some of the patients for free. "The family cannot afford it on their own. Definitely, the Institute cannot afford to give them this particular [treatment] for free," Dr. Abacan said.

At present, there are only eight practicing clinical geneticists in the country. This limited number of medical specialists who know how to diagnose and treat these disorders is another challenge.

The countenance of 'rare'

Having to deal with one member suffering from a rare disease is already agonizing for the family; but for Angelita Obeda, make it three times more. Among her six children, three have Hunter Syndrome. Also called the Mucopolysaccharidosis Type II (MPS II), this rare disease generally affects boys, wherein an enzyme that breaks down specific kind of complex sugars is deficient.

The Parco brothers perform with other kids with rare disease during the "I Am Rare" event on 25 April 2015 at the University of the Philippines Manila.

It was only in 2008 that the Parco brothers – twins Peter John and John Paul, and the youngest Vicente – now 18 and 17 years old, respectively, have been diagnosed with MPS II. Years prior to the diagnosis of their true condition, the siblings were almost always feeling ill. According to Nanay Angie, motherhood became more challenging for her when

she had the three boys. "Pag nagkasakit [ang isa], sunud-sunod na (When one gets sick, it becomes seriate.)," she said.

Unlike other boys their age, the Parco brothers stand at about three feet only. They have thick lips, clawed hands, hoarse voice, and have difficulty in enunciating the words properly and clearly. Because of these distinct features, they often become objects of mockery. Nevertheless, no matter how upsetting, the family has learned to graciously accept other people's prejudices.

In the midst of all these, Nanay Angie and her three elder children have been the constant power source from whom the Parco brothers draw strength and vigor. Moreover, the brothers themselves, obtain courage and boldness from each other. "Sabi nila kailangan sabay-sabay para malakas (They said they must stick together to be strong.)," Nanay Angie recalled her sons' conversation.

Like the Parco brothers, nine-year-old Pauline Martinez has been trying to live a normal life since she was diagnosed with Gaucher disease, a type of lysosomal storage disorder. For people with this kind of rare disease, the enzyme in their bodies that breaks down particular fatty substances is lacking or does not function accordingly. Hence, these fatty substances accumulate in certain organs, specifically in the liver and spleen.

Pauline, who is now in fifth grade, will have to battle with her condition permanently since Gaucher disease, just like all inborn errors of metabolism, has no cure. According to Dr. Abacan, one of the treatments for this disease is the long-term enzyme replacement therapy. In the case of Pauline, she receives an intraveous infusion of Cerezyme fortnightly. "Cerezyme contains a synthetic form of the enzyme na kulang sa kanya (that is deficient in her body), and she will have to receive it for life," Dr. Abacan explained.

At the moment, Pauline is one of four patients who receive the Cerezyme therapy for free as part of the International Charity Access Program of Genzyme, a biotech company that is dedicated to doing research, developing, and manufacturing Gaucher disease treatments. In administering Cerezyme, the dose is dependent on the patient's weight and the severity of the disease. Each vial costs around 600-700 USD, which is excessively unaffordable for a regular Filipino family.

Chancellor Carmencita Padilla (better known as "Tita Menchit" to the kids with rare disease) carries Gaucher disease patient, Pauline Martinez, in her arms.

Without external help, the therapy would be beyond the financial means of and cannot be sustained by Pauline's family. Knowing that his daughter will be under treatment all her life, Randy Matinez expressed his melancholy. "Yung gamot wala dito; tapos napakamahal pa nung gamot (The treatment is not available here, and is very expensive.)," the patient's father said in a sobbing voice.

Solace amidst anguish

Adding to the torment of the patients and their families is the feeling of social rejection due to the absence of organized support systems. Moreover, the lack of awareness among Filipinos about these diseases has rendered the patients as objects of ridicule.

It is in this light that the Philippine Society of Orphan Disorders, a non-profit organization, was organized with UP Manila Chancellor Carmencita Padilla as the founding chairperson. The PSOD relies on the basic premise that everyone has a right to quality life. Although Filipinos afflicted with rare diseases are insignificant in number, the organization still believes in the significance of guaranteeing the patients with better access to health.

Since its formal incorporation in July 2006, the PSOD has managed to clump its endeavors into four general programs – patient and family welfare, awareness and advocacy, resource generation, and program development.

The PSOD aims attention at finding mechanisms and channels so that the patients' medical and non-medical needs can be afforded. Aside from patient care, the organization also provides support to families with members suffering from orphan disorders.

According to PSOD President Cynthia Magdaraog, since rare diseases are lifelong,

Fifth-grader Pauline Martinez enjoys playing with other kids during the "I Am Rare" event.

the patients' families have to be strong. The organization provides families with home skills to care for the patients properly, such as physical therapy and communication augmentation. They also coach the families on the principles of saving and provide ways to improve the families' financial capacity. "If there is no help, it will increase the number of families who will be in poverty," Magdaraog said.

Moreover, the PSOD believes that the best way to provide rare disease patients with better access to medical care and health information is by including them in the universal health agenda. Hence, the organization actively participates in the research process for policy formulation and staunchly supports the passage of the Rare Disease Act. "Our Founding Chairman, Dr. Padilla, was the one who drafted the bill. The role of the parent organization is to do the legwork, to follow up with the senators, with the health committee, the ways and means committee, so that it moves," the PSOD President said.

The need for a policy

Given the country's current condition in terms of rare disease and the challenges and struggles faced by rare disease patients, the enactment of the Rare Disease Bill, an all-inclusive act that will address the needs of persons stricken with orphan disorder, would be a great encouragement and major lift to all orphan disorder patients and their families.

At the Senate, the bill was introduced by Senator Pia Cayetano during the 16th Congress as Senate Bill No (SBN) 2098. The bill specifies for the formulation of a comprehensive and sustainable health system for orphan disorders that will be incorporated within the current public health care delivery system. This is supported by SBN 1868 of Senator Miriam Santiago, an Act mandating the University of the Philippines National Institutes of Health to conduct research on pediatric rare diseases and conditions; SBN 2279 of Senator Cynthia Villar, an Act promulgating a comprehensive policy in addressing the needs of persons afflicted by rare disorders; and SBN 2383 of Senator Sonny Angara, an Act promulgating a comprehensive policy in addressing the needs of persons afflicted by rare disorders.

At the House of Representatives, the bill was introduced by Rep. Diosdado Macapagal-Arroyo as House Bill (HB) 2800, an Act to help persons afflicted with rare diseases by creating an office of rare diseases in the Department of Health, encouraging the conduct of research and

development activities on rare diseases, and providing for fiscal and regulatory incentives for the manufacture or importation of healthcare products for use by such persons. This bill was supported by Rep. Sol Aragones (HB 3258), Rep. Rose Marie 'Baby' Arenas (HB 3343), Rep. Alfredo Vargas III (HB 3634), Rep. Randolph Ting (HB 3896), and Rep. Gus Tambunting (HB 4002).

The Rare Disease community of physicians, families and supporters are hopeful that it will be enacted into law in the 16th Congress.

The bill ensures that rare disease patients have access to up-to-date health information and sufficient medical care to treat or aid them in coping with their condition. This includes the provision of early and sustainable care for patients; creation of a registry with data that will be used in policy formulation, program interventions identification, and relevant researches; and incorporation of instructional campaigns in the DOH's existing programs to raise awareness and foster understanding of the patients' special needs among the public.

The DOH, which shall be the principal agency in implementing the Act, shall set up a National Rare Disease Registry. The bill provides that "all healthcare practitioners and health institutions shall be required to report to the Rare Disease Registry of the DOH and NIH diagnosed cases of rare disease and provide reports on the status of patients". To ensure follow up by specialists, the bill provides that individuals diagnosed to have rare disease shall be referred to the Regional Continuity Clinics of the Newborn Screening Program, equipped to care for patients with rare diseases. Continuity Clinics have direct access to specialists through the telegenetics program.

In accordance with the Magna Carta for Disabled Persons, patients afflicted with rare disease shall be considered as persons with disabilities (PWDs).

The bills provide that the relevant national government agencies, such as the Department of Social Welfare and Development (DSWD) and Department of Labor and Employment (DOLE), shall make sure that rare disease patients be given the same rights and privileges as PWDs.
The bill also provides for funds to institutionalize the implementation of the program from diagnosis to treatment.

Hence, the rare disease patients, together with their families, the PSOD, and other orphan disorder advocates hold their hope fervently that their tiny voices be heard. The enactment of the bill would allow the patients to embrace a quality life, even if they have to endure their rare disease permanently. •

Photo credit: Ms. Josa Camille Bassig

The Parco brothers, despite having Hunter Syndrome, are grateful they have one another as source of strength and encouragement.

Empowering the youth for a healthier nation

by January Kanindot

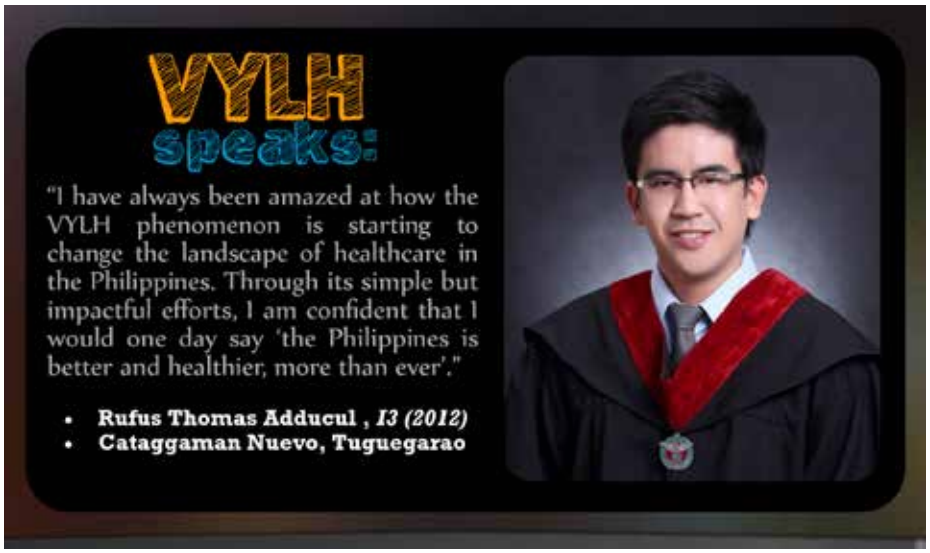
From its initial Folic Acid Awareness Campaign, the VYLH-Philippines' advocacy has grown to include two more critical health issues - Newborn Screening Promotion and Orphan Disorder Support – forging linkages with other health organizers and preparing to create international tie-ups.

The nation pins its hope on the youth. They serve as the foundation upon which our nation's vision is fulfilled. Their collaborative support can be pivotal in implementing successful and sustainable programs. The Filipino youth, with ages ranging from 15-30, constitutes 40% of our population. This figure is a huge reservoir of potential that can be harnessed to deliver real healthy changes in the nation. After all, health is wealth.

In a report released by Save the Children in 2014, the Philippines remains in the top 10 countries with the biggest inequality gap in newborn mortality rate between (1) the poorest and wealthiest; and (2) rural and urban communities. The studies revealed that more newborns die in poor and underprivileged households and rural communities. Therefore, the thrust for greater education, cooperation, accessibility, awareness, and early intervention is of utmost importance if we are to save the newborns of this nation.

With this, Chancellor Carmencita D. Padilla, then Director of the UP Manila-National Institutes of Health's Institute of Human Genetics (IHG) heeded the call of the March of Dimes - Global Network on Maternal and Infant Health (MOD-GNMIH) in 2008. It was a call to mobilize the youth in support of the Millennium Development Goals (MDGs), in particular, MDG 4: Reducing Child Mortality; and MDG 5: Improving Maternal Health by 2015. She pushed through with the project thinking that it was a one-shot deal. Never did she realize that she would be laying the foundation for an organization which would give its members a life-changing experience; that it would make a huge difference in the communities it would serve; that six years from its founding, the

VYLH-Philippines first batch



organization would grow by leaps and bounds and transform into a nationwide network of passionate, dynamic, visionary, pro-active, and dedicated youth leaders.

Chancellor Padilla started the ball rolling by handpicking University of the Philippines Los Baños' Genetic Society (UPLB-GenSoc) members to support the National Birth Defects Registry Project and Folic Acid Awareness Campaign. GenSoc, together with its partners at IHG, Jennifer Chiu and Aster Lyn Sur, and under the guidance of Dr. Merlyn Mendioro, the senior adviser of the UPLB Genetics Society, developed the program proposal and youth questionnaire.

On January 2009, the group presented the campaign to the UP Los Baños administration through Vice Chancellor for Instruction Dr. Rita Laude. Thereafter, they secured an endorsement from UPLB Chancellor Dr. Luis Rey Velasco.

The Folic Acid Campaign was officially launched at the 4th Annual Genetics Camp in UPLB, an event attended by high school students and faculty. A month after the success of the campaign, UPLB-Volunteer Youth Leaders for Health (VYLH) was formally instituted with GenSoc, UPLB Microbiological Society, UP Oikos, and UP Society of Human Settlements Planners (UP HSPSoc) as its core group.

The Volunteer Youth Leaders for Health's giant strides

By July 2009, the core group members were the pioneering facilitators of the Institution and Planning workshop of the Volunteer Youth leaders for Health (First National Youth Camp). Held in Dacha Hotel, Tagaytay, the three-day event aimed to create a network of volunteer youth leaders and youth organizations in universities and schools in major cities and communities. The event also endeavored to promote awareness on the role of folic acid in the prevention of birth defects, specifically neural tube defects, among the volunteer youth leaders. The participants in this youth camp became the founding members of the VYLH-Philippines, which was co-funded by the IHG and Department of Health (DOH).

The VYLH-Philippines, unlike what most people think, accepts youth volunteers who may not have a medical background. In fact, some of its members are communications, Information Technology (IT), and engineering majors. "We do not filter [the volunteers] because we are not leaning to a particular course of interest. So long as you have the passion and the drive," Rufus Adducul, cluster head of NCR South Luzon, shared.

Today, VYLH-Philippines continues to grow with almost 4000 volunteer members from its NCR South Luzon, North Luzon, Visayas, and Mindanao clusters of university- and community-based organizations. Serving as the main information arm of the Newborn Screening Reference Center (NSRC), VYHL-Philippines has pulled out all the stops in disseminating necessary, relevant, and up-to-date information that has saved countless lives.

They conduct regular lectures in schools and communities; hand out flyers even in public markets; give lectures to mothers in hospitals and health centers; and sponsor symposia and exhibits. To keep up with the digital age, they have also creatively used social media and other online-based tools that have effectively increased public awareness of their advocacies.

Champions of health

From its initial Folic Acid Awareness Campaign, the VYLH-Philippines' advocacy has grown to extend to two more critical health issues affecting the country's future generation. One is the Newborn Screening Promotion – a program where a baby is tested for congenital metabolic disorders, which if left untreated, can lead to mental retardation and even death. Another is the Orphan Disorder Support. The orphan or rare disorder is any health condition resulting from a genetic defect that affects only one in 20,000 individuals in the Philippines.

What makes rare disorders wistfully unique is that they are chronic, progressive, disabling, and life threatening.

In order for its advocacies to have greater impact, the VYLH-Philippines creates linkages with other societies and organizations. In March 2015, the VYLH collaborated with the Philippine Obstetrical and Gynecological Society (POGS) in the celebration of the 12th Buntis Day across the country. Volunteers were able to share the importance of folic acid supplementation and newborn screening with hundreds of pregnant women.

In April 2015, the VYLH held the 'I am Rare' event, a one-day program that gathered patients with rare disorders and their families. The event was a welcome reprieve to the afflicted individuals and their families. Not only was it a venue to relax and enjoy, but more importantly, it was a day of support and empathy as they were able to connect and share with fellow families the challenges and triumphs of a rare disease patient.

To date, one of the inimitable achievements of the VYHL-Philippines is the monumental success of the signature campaign for the RARE Disease Act of the Philippines. The organization was able to gather the most number of signatures in support of the bill. They were able to collect 14,000 signatures out of the total 19,000+. The legislation, if passed, will create a Rare Disease Program at the Department of Health (DOH). It will ensure that our society is mandated to take care of the individuals who are afflicted with rare disorders however few they are.

VYHL-Philippines has just recently conducted a number of integrated advocacy orientations and distribution of brochures on newborn screening, folic acid and orphan disorders in various groups and locations, namely, the IE Council of University of San Carlos, Cebu; at the Taculing Youth Summit in Bacolod City; and, at the Freshmen Orientation at Holy Name University - Dampas College of Business and Accountancy. As part of the post-camp activity of the Negros Oriental volunteers, they conducted the Operation Bigay Brochure to locals of Oslob, Cebu.

What lies ahead

To further strengthen its base volunteers, the VYHL holds a nationwide congress every two years which gives members from the different clusters the opportunity to interact with one another. It also becomes a venue where new breakthroughs of their advocacies are presented. Member volunteers are trained so that they may be more equipped to share information.

The VYLH-Philippines is poised to grow to an even bigger network. To share the organization's practices and passions, it is now looking into international partnerships. The organization may soon be able to accept 6-8 weeks internships from different countries. The dream to be a global network may be well within its reach.

The Philippines has still a lot of ground work to do in decreasing child mortality. Access to health service remains to be one of the country's greater challenge. But with a growing volunteer network, such as the VYLH-Philippines, the country is headed towards a more productive citizenry and a healthier Philippines.

Share the fun and visit http://vylhphilippines.blogspot.com. •

VYLH Visayas wide camp teambuilding activities

Philippine Genome Center - government's commitment and investment in genomics science

by Fedelyn Jemena and Mary Ann Romero

"In these modern times where the existence of mankind is being challenged by nature, science has to come in to provide support and solution." – DOST Secretary Mario Montejo

The Philippine Genome Center is located in the University of the Philippines in its Diliman Campus in Quezon City. But before going into the creation and components of this new Center in UP, it is important to understand the word "genome" which may come as a big word especially for people with no science background.

A genome is the total genetic information contained in an organism, including its genes and DNA sequences. It can be relatively simple as those found in viruses and bacteria, or complex and composed of many copies inside the cells of an organism like in plants, animals and humans. The www.yourgenome.org of the Wellcome Trust Sanger Institute described the genome as the "recipe book of the body" (of a human, animal, or plant) since it contains the complete set of an organism's genetic instructions. The instructions are made up of DNA, which has, inside it, "unique chemical code that guides our growth, development, and health."

In 2008, the Philippines hosted the Human Genome Organization Conference in the Asia Pacific. After this conference, then University of the Philippines (UP) President Emerlinda Roman, created a committee to study the feasibility of setting up a Genome Center in the University. The committee consisted of then UP Vice President for Academic Affairs Amelia Guevara (Chair), Dr. Gisela Concepcion of the Marine Science Institute, UP Diliman; Dr. Carmencita Padilla of the UP College of Medicine, UP Manila; and Dr. Cynthia Saloma of the National Institute of Molecular Biology and Biotechnology, UP Diliman. The committee visited several genome centers in the region (Hong Kong, Taiwan, Singapore) and the United States to obtain insights towards the development of a Philippine Genome Center (PGC).

During the development phase of the PGC, several major events led to its establishment – the Dengue outbreak that reached alarming rates, the SARS outbreak in Hong Kong and the H1N1 outbreak in Mexico, reinforcing the importance of genomics in providing new solutions.An audience with President Gloria Macapagal-Arroyo in July 2009 paved the way for the first two grant awards of PGC – the Dengue diagnostic test

Building is located at A. Ma. Regidor St. in between Institute of Biology and College of Home Economics in UP Diliman

Dr. Raul Destura with the Dengue Diagnostic Kit

surveillance network through the Department of Science and Technology (DOST).

At the 1246th Board of Regents Meeting on July 31, 2009, the proposal for the establishment of the Philippine Genome Center under the office of the UP President was approved. With an institutional mantra of "Genomics for a Better Philippines", the Philippine Genome Center, a multidisciplinary institution that combines the strength of basic and applied research for development of health diagnostics, therapeutics, preventive products, and improved crop varieties paved the way for more genomics studies nationwide.

Why Genomics?

Genomics is the study of the complete set of genetic material (embedded in the deoxyribonucleic acid (DNA) or ribonucleic acid (RNA) and/or proteins) of an organism. It is a vital tool that can generate knowledge beneficial in unearthing or understanding the genetic composition of plants, animals, and microbes. Through genomics, local scientists may use this branch of science to better understand the biology of these organisms, and their lineage and evolutionary relationships. It can also shed light and provide the genetic basis of diseases.

For more than a decade, pursuing genomics has been a challenge for locally-based Filipino researchers and scientists as the country lacks the much-needed technology and experts in genomics and bioinformatics which are crucial in translating research results into research products poised to benefit the public/industry in the area of agriculture, health and the environment.

PGC's virtual beginnings

In 2010 the Center was operating virtually with Dr. Amelia Guevara, UP System's VP for Academic Affairs who concurrently served as the Acting Executive Director. It was then that the PGC's core group composed of distinguished UP professors namely Dr. Carmencita Padilla from UP Manila served as program director for health, Dr. Gisela Concepcion from UP Diliman as program director for biodiversity, and Dr. Cynthia Saloma, also from UP Diliman, served as the program director for DNA Sequencing Core Facility. Dr. Rita P. Laude, then Vice Chancellor for Instruction of UP Los Banos was also part of the team as program director for Agriculture, Livestock & Fisheries. In February 2011, Dr. Guevara was appointed Undersecretary of DOST and Dr. Padilla assumed the position of Executive Director of PGC. Dr. Eva Cutiongco-de la Paz became the health program director. Joining the group now were Dr.Ernelea Cao as deputy director, Dr. Corazon de Ungria as forensics and ethnicity program director, Dr.ArturoLluisma as program director for Core Facility on Bioinformatics and Dr. Peter Sy as program director for Ethics, Legal and Social Issues.

DOST Secretary Mario G. Montejo recognized the potential of genomics as a catalyst or game-changer given the appropriate equipment and availability of genomics experts.

Thus, in February 2012, the DOST approved PGC's program proposal "Capability Building in R&D Genomics – in order to support the two core facilities of the PGC. – the DNA Sequencing Core Facility, and the Core Facility for Bioinformatics.

PGC's Core Facilities

The two core facilities – the DNA Sequencing Core Facility and the Core Facility for Bioinformatics – crucial in pursuing genomics studies are now available to ultimately serve and equip the local scientific community in genomics research.

For a long time, local scientists send their samples to genomics laboratories abroad like Singapore, Taiwan, Korea, China and even as far as Europe and USA for sequencing. But now, the local scientific community may opt to avail of sequencing and genotyping requirements that the PGC's DNA Sequencing Core Facility offers. Program Director Cynthia Saloma said that aside from cost savings on shipping charges, and reducing turnaround time of sending samples abroad, the DNA Sequencing Core Facility has in-house researchers who provide guidance and consultation on experiment design as an

added service. PGC-DSCF provides a full range of DNA sequencing services including whole genome sequencing, transcriptome sequencing; and the sequencing of small genomes (of virus and bacteria). Bioinformatics analysis is as crucial as genome or DNA sequencing in fact, it is the process that will make the genomics research meaningful and useful. The massive amounts of data generated from any DNA or genome sequencing will need to be analyzed, and that is the vital role of bioinformatics.

Since April 2014, the Core Facility for Bioinformatics of PGC has been providing computing and storage resources, performing data processing and analytical services, and giving training and consultancies to individuals and groups wanting to get started in genomics and bioinformatics. Program director Arturo Lluisma shares that among the strengths of PGC-CFB are its human resource who were trained and equipped with the much-needed bioinformatics expertise, and the powerful computers and large storage devices required to facilitate the processing and data analysis from the sequencing of many genomes (remember that as a Facility, it needs to process and analyze many genomes from different research groups. Currently, CFB's current computing infrastructure consists of 3 SMP servers (each server having at least 512 GB RAM and 40 or 64 cores), data storage servers (>60 TB total usable capacity), and one IBM supercomputer (BlueGene/P, with 4,094 cores). A typical modern laptop has 4 CPU cores, 4 gigabytes of memory and 1 terabyte of storage. The facility's IBM Blue Gene/P Supercomputer and other large capacity servers enables massively parallel computation that can greatly accelerate the data analysis required of genomic data (comprising of billions of letters of DNA). This will consequently increase the number/probability/ chances of potential new discoveries, like new drug/medicine candidates, genetic markers for diseases, tracking evolution of harmful microorganisms, and other important scientific questions relevant to Philippine Society.

Front view, laboratory side of Philippine Genome Center

Next generation sequencing data being analyzed on a symmetric multiprocessor system to produce meaningful biological information

Apart from laboratory services, both core facilities provide training programs, seminars and workshops to fill in the talent gap in genomics and biotechnology.

Abaca and Sugarcane Genomics Projects Are Right on Track

Abaca (Musa textile Nee) is a plant native to the Philippines and is the country's most economically important fiber crop averaging US$90 million per year in exports. These exports consist of raw fiber, pulp, cordage, yarns, fabric and handicrafts and are expected to increase due to demands for natural materials such as fiber. The abaca industry suffers from low yield and poor quality fiber primarily due to the infestation of abaca by the bunchytop virus (BTV) disease, among others. Dr. Antonio Laurena, lead scientist of the abaca genomics project reports that it is the most advanced in the PGC Agricultural Program. Simple sequence repeat (SSR) based markers derived from genomics studies are now used in DNA fingerprinting and varietal identification of an abaca hybrid (BC2) with bunchy top resistance and enhanced fiber quality for pulp and paper applications. The project will be extended until December of 2015 to develop SSR markers for BC3 hybrids that will be used for rope and chordage applications.

The Philippines slipped down to No. 2 exporter of sugar to the US trailing behind Brazil. Brazil has heavily invested in emerging technologies including genomics in improving its production. Because of the

Christmas in January 2015, PGC Program leaders with research staff

urgency of obtaining novel and emerging technologies for the development of better sugarcane varieties in the Philippines, the sugar industry approached DOST to seek the assistance of PGC in developing new varieties using genomics as a tool.

Sugarcane breeding is a long and tedious process that can last for eight to nine years from hybridization to the release of a variety. The sugarcane genome is one of the most complex plant genomes with the commercial varieties having more than 100 chromosomes. The implementation of genomics in the breeding of sugarcane can result to the identification of novel genes associated with important traits and the generation of molecular markers that can be used to fast-track the breeding and improvement of sugarcane varieties. The sugarcane genomics project that started in August 2013 will end in December 2016. Dr. Laurena, lead scientist for the sugarcane project, proudly reports that the program is on the right track to generate at least 5 promising sugarcane lines for varietal development. This is indeed good news to the 60 000+ farmers of the country.

The 'Lab-in-a Mug' Project, Personalized Medicine and more

The Health Program of the Philippine Genome Center is as equally engaged in using genomics for better health in the Philippines. The main goal of the Health Program is to apply genomics for the promotion of Filipino health and the prevention and management of public health problems. This goal is being accomplished through research towards the development of diagnostic kits that can be used at the point of care for certain diseases.

One of the most anticipated projects on kit development is the Lab-in-a-Mug where all diagnostic kits are integrated and miniaturized in an isothermal unit as small as a "mug" which functions as a multi-infectious disease diagnostic device similar to a portable laboratory. The "Lab-in-a-Mug" was conceptualized and designed at the Institute of Molecular Biology and Biotechnology, National Institutes of Health (NIH), with Dr. Raul Destura, the current Deputy Director of NIH, as lead scientist. This Biotek-M Dengue Aqua Kit Commercial Prototype, which has been proven to have high specificity and high sensitivity, is already undergoing clinical trial in 67 hospitals nationwide. In the pipeline are testing kits for MDR and XDR tuberculosis, salmonella, leptospirosis, filariasis, schistosomiasis, paragonimiasis and chikungunya.

Together with the Department of Health, the Institute of Molecular Biology and Biotechnology led by Director Edsel Maurice Salvana, has developed a web-based Interactive Genome Library for Surveillance, Detection, Characterization and Drug-Resistance Monitoring of Influenza Virus Infection in the Philippines. Researches on non-communicable diseases such as cardiovascular disease and diabetes, among the top causes of mortality in the country, are also underway.

Dr. Edsel Maurice Salvana who is developing a web-based genome interactive library.

IBM Blue Gene/P super computer of the Core Facility for Bioinformatics

Dr. Rody Sy, Chair of the Department of Medicine, UP College of Medicine is the Program Leader for the Program entitled, "Genomic Researches on Hypertension, Coronary Artery Disease and Dyslipidemia Towards the Development


Dr. Rody Sy, Program Leader of a research on genomics research on coronary diseases.

of Individualized Diagnostic and Therapeutic Strategies." The Cardiovascular Genomics program seeks to ascertain the most important genetic variations present in the population that predispose Filipinos to hypertension and coronary artery disease and their complications that lead to early death. Included in this program is determining genetic differences that are present among Filipinos that can be used to predict whether a medication will be effective for a particular person or not and to help prevent adverse drug reactions.

Dr. Elizabeth Paz-Pacheco, former Head of the Section of Endocrinology, Department of Medicine, UP College of Medicine, is the Project Leader of the "Genomic Association Studies in Filipinos on Treatment, Diagnosis and Risk Assessment of Type 2 Diabetes Mellitus and Its Related Medical Condition." Similar to the cardiovascular genomics researches, the diabetes project also seeks to determine genetic variations present in the population that predispose to diabetes and its devastating complications such as diabetic nephropathy and retinopathy as well as study of how genes affect a person's response to commonly used drugs in the treatment of diabetes. Data generated from these researches will allow the creation of a pharmacogenetics database that can assist physicians in choosing the right drug, the right dose for the right person based on that person's genetic profile.

According to PGC Health Program Director Eva Cutiongco-de la Paz, concurrently UPM Vice Chancellor for Research and NIH Director, knowledge that comes from these genome-based researches aims to prepare the Philippines to an emerging practice of medicine called Personalized Medicine, an approach to health care which uses a person's genetic profile to guide decisions on prevention, diagnosis, and treatment of disease.

PGC's Final Home

Just like any other new program, Executive Director Carmencita Padilla relates that PGC has its own share of birthing pains – budget, staff items, procurement issues, etc. PGC has 26 program/project leaders and 85 research assistants spread out in UP Diliman, UP Manila and UP Los Banos. Its primary budget source is DOST through its 3 councils – Philippine Council for Health Research and development (PCHRD), Philippine Council for Agriculture, Forestry and Natural Resources Research and Development (PCAARRD) and Philippine Council for Industry and Energy Research and Development (PCIEERD). The PGC administrative office and core facilities are temporarily housed at NIMBB while waiting for the completion of its building.

PGC is fortunate to have the unwavering support of President Alfredo Pascual, who believes in the significant role of PGC in the provision of creative solutions to old problems of the country. He lobbied for the funding of its permanent home, a complex of 3 buildings being constructed in phases. The first building will be opened to the public at the end of 2015, the second building in 2016 and the last one in 2017.

For inquiries visit www.pgc.up.edu.ph or contact 981-8742, 981-8744 or via pgc@up.edu.ph •

Newborn Screening

Ano ang PHENYLKETONURIA o PKU?

Josephine D. Agapito*

Kung programang pampublikong tungkol sa kalusugan
Dapat ang Newborn Screening ay alam ng mamamayan
Tungkol ito sa mga sanggol na kasisilang pa lamang
Kung mayroong magiging sakit ay agad na malalaman

Maaaring sanggol ay walang senyales o sintomas
Normal ang itsura pero ang genes ay problemadong tiyak
Kaya kung susuriin ang dugo lalo at pagkasilang agad
Malalaman ang dapat gawin at kayang mailigtas

Dahil kapag hindi ito naagapan kaagad
Minsan ang pagkamatay ang unang sintomas
O kaya ay panghabambuhay na abnormal ang labas
Kaya napakalaking tulong kung ma-iiscreen ang lahat

Taong mga 1960s ito nagsimula sa Estados Unidos
Two thousand seven naman ng maibatas ang utos
Pero sa ating bansa ay taong 1996 nagsimula ang study group
Taong 2004 naman ng maisabatas dito sa bansang lubos

Ang kaunahang sakit na sadyang nadiskubre
Phenlyketonuria ang pangalang sinasabi
Mental retardation ang epekto nito kasi
Kung maalalaman agad ay mapaghahandaan siempre

Kaya sa ilalim ng RA 9288 ay sadyang ipinapatupad
Na lahat ng kasisilang na sanggol ay dapat maeksamen kaagad
Batay ito sa karapatang mga tao ay dapat na ligtas
Mula sa pagkasilang at sa paglaking ganap

Manggagamot mula sa 24 na ospital sa MM
Mga pediatricians na eskperto sa bata sila
Obstetricians din na nagpapaanak sa mga ina
Sila ang bumuo nitong nasabing programa

Isa sa mga nauuna sa listahan ng layunin
Anim na kondisyong metabolic ang iintindihin
Incidence o ilan ang nagkakaroon nito ang aalamin
Nasa sunod na stanza ang dapat kilalanin

Congenital o mula pa lang sa pagkakaanak
Hypothyroidism, Adrenal hyperplasia ang tawag
Phenylketonuria, galactosemia, hemocystinuria ay idagdag
Maple Syrup Disease at Glucose -6-phosphate dehydrogenase
deficiency ay tiyak

Bukod sa layuning maisagawa sa buong bansa
Sila rin ang nagpaplano at nagbibigay panukala
Mula sa mga pananaliksik na kanila ring isinasagawa
Para mas mapagbuti pa ang serbisyong ito sa madla

Sa kasalukuyan ay may Newborn Screening Centers sa mga lalawigan
Kung saan ang dugong kinulekta ay susuriin ang kalagayan
Kasama na sa PhilHealth package kung iniisip ang babayaran
Resulta ng mga pagsusuri ay agad namang ipinaaalam.

Kaya naman patuloy ang programa sa pagseserbisyo
Kaakibat ng maraming pananaliksik tungkol dito
National Institutes of Health sa UPManila ang nangungunang totoo
Walang hinahangad kungdi ang kapakanan ng bawat Pilipino.

Serye ng tula na inilalathala
Tungkol sa "Genetic Disorders" ang paksa
Ibabahagi ang impormasyon sa madla
Nawa ay makadagdag yaman sa isip, puso at diwa

Sakit ito na sadyang namamana
Naisasalin sa lahi kumbaga
Deteksyon pagkasilang ay mahalaga
Kaya mag- Newborn Screening na

Katawan kasi natin ay binubuo ng mga kemikal
Isa na rito ang protinang laging kinakailangan
Kung may depekto ang gene na siyang aasahan
Sa paggawa ng enzyme para Phenylalanine ay pakinabangan

Phenylalanine kasi ay isang uri ng amino acid
Na siya namang bumubuo ng protinang nais
Di ba't katawan natin ito ay pinoprocess
Pinapasimple para sa metabolismo ay magamit

May problema kung hindi makagawa ang katawan
Nitong enzyme para Phenylalanine ay magamit naman
Tumatambak o dumadami sa dugo iyan
Siya na ngayong nagdudulot ng mga karamdaman

Dahil di agad nakikita ang sintomas sa bagong anak
Mga ilang buwan pa makikita itong ganap
Kaso ang epekto sa utak ang sadyang masaklap
Mental retardation ang siya kasing katapat

Siempre ay marami pa ang kaugnay na problema
Pagdebelop o paglaki ay maantalang talaga
Bukod pa sa maraming sikolohiyal na suliraning kasama
Mga pangingisay, paghina ng buto, maliit ang ulo ay makikita

Kaya buong buhay ang tutok ay mahigpit
Kailangang ang masusing pagsubaybay sa diet
Pagkukunsulta sa doktor ay kailangang higit
Kasama na ang pag-aaral tungkol nga sa sakit

Hindi pare-pareho ang antas ng pagkagrabe
Lalo kung naagapan kaagad ay mas mabuti
Kaya Newborn screening ay inirerekomendang maige
Matutukan kaagad at mga payong gagawin ay masasabi

May mga nagbubuntis din na may ganitong peligro
Kaya kailangang nagpapacheck up ng regular mismo
Para ang anak sa sinapupunan ay di rin mamiligro
Lalo kung ang ina mismo ay namana nga ito

Kailangang istatistika ay sadyang iintindihin
Sa Pilipinas ay 1: sa loob ng 102,000 mandin (PSOD Aug 2011)
Kaya naman napakahalaga ng newbor n screening
Di naman lingid sa inyo ang dami ng populasyon natin

Sa huli ay inirerekomenda ang newborn screening
Ang PKU ay isa lamang sa nasabing anim
Kung saan ito ay isinabatas na rin (RA 9288)
Para mas malawak ang matutulungang magaling.

* Josephine D. Agapito
Guro sa Department of Biology, College of Arts and Sciences, UP Manila; nagsusulat at bumibigkas ng mga tula tungkol sa agham sa DZEC tuwing Linggo, 5 pm sa programang "Pinoy Scientist."

University of the Philippines Manila

National Graduate Office for the Health Sciences

Pioneering graduate programs in the health professions

Continuing recognition as CHED Centers of Excellence and Development

Shaping the minds of scientists through DOST-PCHRD scholarships and other support mechanisms

Fostering a wide array of academic and research opportunities through flourishing local and international linkages

Pursue graduate education in the country's national university.

MD-PhD Molecular Medicine
PhD in Biochemistry
PhD Nursing
Doctor of Public Health
MA Health Policy Studies (Health Science)*
MA Health Policy Studies (Health Social Science)*
MA Nursing
MS Biochemistry*
MS Bioethics*
MS Clinical Medicine
MS Dentistry (Orthodontics)
MS Epidemiology (Clinical Epidemiology)
MS Epidemiology (Public Health)*
MS Genetic Counseling*
MS Health Informatics (Bioinformatics)*
MS Health Informatics (Medical Informatics)*
MS Medical Anthropology*
MS Pharmacology*
MS Pharmacy
MS Physiology*
MS Public Health*
M in Clinical Medicine
M in Medical Anthropology*
M in Orthopedics
M of Basic Medical Sciences
M of Clinical Audiology*
M of Health Professions Education*
M of Hospital Administration*
M of Management*
M of Occupational Health*
M of Physical Therapy
M of Public Health*
M of Rehabilitation Science
M of Rehabilitation Science-Speech Pathology
Certificate of Proficiency in Orthodontics
Diploma in Dental Public Health*
Diploma in Health Professions Education*
Diploma in Preventive Ophthalmology
Basic Course in Health Professions Education*

*open to any baccalaureate degree holder
(with specific qualifications per program)

For inquiries, call 02-526-5870/ 523-1495/ 523-1498
National Graduate Office for the Health Sciences
University of the Philippines Manila
3/F Joaquin Gonzales Hall, Padre Faura corner Ma. Orosa Streets
Ermita, 1000 Manila

visit www.ngohs.upm.edu.ph for the downloadable forms

Subscribe to: www.youtube.com/upmanilachannel
Log on to: www.upm.edu.ph
Like us on: www.facebook.com/upmanilaofficial
Follow us on: www.twitter.com/upmanilaonline

Information, Publication, and Public Affairs Office
University of the Philippines Manila, 8/F Central Block Building, UPM-PGH, Taft Avenue, Manila 1000
Tel. No. 554.8400 local 3842 • Email: ippao_upm@yahoo.com

A genome is the total genetic information contained in an organism, including its genes and DNA sequences. It can be relatively simple as those found in viruses and bacteria, or complex and composed of many copies inside the cells of an organism like in plants, animals and humans. The www.yourgenome.org of the Wellcome Trust Sanger Institute described the genome as the “recipe book of the body” (of a human, animal, or plant) since it contains the complete set of an organism’s genetic instructions. The instructions are made up of DNA, which has, inside it, “unique chemical code that guides our growth, development, and health.”