Medical Genetics: ...still growing and expanding...

Today, it is not uncommon to read about novel treatments for conditions that were considered "untreatable" 30 or 40 years ago. Learning about biochemical or metabolic disorders then was challenging because there were no confirmed patients to speak of in the Philippines. Unexplained neonatal deaths were attributed to sepsis and there was no genetic test to challenge the diagnosis which had sound clinical basis. Multiple congenital anomalies were viewed as normal deviations or variants of an embryologic process. Did we even think then that these anomalies were part of a syndrome?

With the establishment and expansion of genetic services in the country[1], among them cytogenetics, newborn screening, molecular and biochemical genetics, our pediatric "unknowns" now have names of conditions that were only encountered in textbooks: maple syrup urine disease, PKU, methyl malonic acidemia, glutaric aciduria, mosaic Trisomy 13 and Tetrasomy 9p Syndrome.

The Philippines joined the rest of the world in December 2018 when the Department of Health implemented the expanded newborn screening nationwide.[2] This provided a platform to screen for at least 28 metabolic conditions, including hematologic and endocrine disorders. This ushered in a better understanding of the clinical course of conditions detected at birth and treated promptly. It opened doors for research collaboration between specialties within the country and overseas.[3] More importantly, it paved the way for better care for patients through sharing of expertise, best practices and the crafting of clinical practice guidelines.[4]

Acta Medica Philippina Genetics 6 highlights the diversity of applications and reach of medical genetics into various aspects of health care: preconception, early neonatal detection of treatable genetic metabolic conditions, disease risk detection and allelic associations, gaps in education of genetic conditions and genetic counselling. The case reports and case series describe various conditions and scenarios that offer opportunities to be acquainted with uncommon genetic phenotypes. May Acta Genetics 6 inspire the reader to explore the limitless possibilities of the applications of medical genetics.

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