First PMM2-CDG Diagnosis in Mexico

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Since 1980 when Dr. Jaak Jaeken reported the first clinical description of a child affected by a Congenital Disorder of Glycosylation (CDG), which was subsequently found to be PMM2-CDG (formerly known as CDG-Ia; OMIM:212065), diagnosis of CDGs has spread around the world. Nonetheless, it's important to acknowledge that CDG diagnosis is still very limited and probably not being tested for at all in many countries. This is particularly true in low-income countries, from where scarce reports are issued regarding CDGs. Lack of awareness and diagnostic infrastructure are the primary factors that contribute to this dilemma. This situation, shared by many rare diseases, has a serious impact on health by avoiding the diagnosis of hundreds of children with CDG and denying them the specialized clinical follow-up they deserve.

In Latin America and the Caribbean, a region with 650 million people, only a few countries have reported CDG cases. These are mainly Argentina, Brazil and Mexico. In Mexico, we started in 2010 to offer free CDG screening in our laboratory, but it wasn't until 2014 that we reported the first cases of CDG in collaboration with Dr. Hudson Freeze (SBP Medical Discovery Institute, USA) and Dr. Kimiyo Raymond (Mayo Clinic, USA). These cases involved two children affected with Cutis Laxa Syndrome and who were diagnosed with ATP6V0A2-CDG (OMIM:219200).

However, in September 2020, in collaboration with Dr. Carmen Alaez, Head of the Genomic Diagnosis Laboratory of the National Institute of Genomic Medicine (Mexico) we confirmed the very first case, in Mexico, of a child diagnosed with PMM2-CDG, the most common type of CDG. This diagnosis was accomplished through exome sequencing, which is an especially useful technology that allows us to quickly screen the genome for mutations and find the answers we are looking for. Unfortunately, this technology is still not widely available in Latin America, limiting access for most patients.

Each CDG diagnosis is a great accomplishment because we can give families and clinicians the answers they have been looking for for many years. Having a confirmed diagnosis, allows us to set these patients on the clinical path to obtain the best quality of care and connect them with family organizations where they can find support, as has happened with Mexican families that are being connected with CDG CARE. It is also a great opportunity to increase CDG awareness in the clinical and research community, and society in general.

This recent diagnosis came at a time when we were invited to organize the first CDG Course in Mexico that was held on October 3, 2020 as part of the XLV Human Genetics National Congress (Mexico) and where international Leaders in CDG came together in an online format, including Andrew Edmonson MD PhD (Children's Hospital of Philadelphia, USA), Carla Asteggiano PhD (Cordoba University, Argentina), Eva Morava MD PhD (Mayo Clinic, USA), Francois Foulquier PhD (Université de Lille, France), Hudson Freeze PhD (SBP Medical Discovery Institute, USA), Kevin P. Campbell PhD (University of Iowa, USA) and Taroh Kinoshita PhD (Research Institute for Microbial Diseases, Japan).

We are hopeful that courses like these will allow us to increase awareness among physicians and bring hope to more children and their families by offering them free CDG screening and potentially the genetic diagnosis they have been searching for. While this is a difficult task to accomplish with very limited government funding, it has been made possible through personal commitment and collaborations. CDG has no boundaries and support shouldn't either. We are confident that communication and networking is key in achieving this for Latin American and the rest of the world.