Impact of my research

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Childhood neurodevelopmental conditions like Rett and Angelman Syndrome affect the trajectory of a child's and a family's development as challenges and uncertainties are constant in their lives. The world can be a confusing place for these children, who may communicate differently, find social interaction difficult or stressful, and experience sensory sensitivities. Parenting a child with disabilities often comes with greater worry, increased stress levels, financial insecurity, dealing with stigmatization and discrimination and many other physical, psychological or socio-economic challenges.

Rare diseases are complex. A patient will often present symptoms associated with more common conditions, which is why they can be so difficult to diagnose. Despite technological and medical advances in recent years, a misdiagnosis or a lack of knowledge remain significant challenges for people with a rare disease or undiagnosed condition. According to research conducted in 2014 by Global Genes, a patient with a rare disease will visit an average of 7.3 physicians and it will take 4.8 years from symptom onset to an accurate diagnosis. Being without a diagnosis is a very difficult place for families to be. Achieving one, even if there is no cure, can at least help the family understand what they are facing, and they can start to focus on how to manage the disease and their "new normal".

It is well understood that patients succeed with coordinated care and structured support but what is available can vary greatly. Access to support services and therapy, even if it exists, can be an enormous obstacle. Healthcare is often inaccessible for many families and therefore, they must navigate the needs of their loved one without any guidance. It has become a common phenomenon among Puerto Rican families where they are forced to leave the island in search of treatment options. Another ongoing challenge is the impact on the family and care givers. In many cases, it is the parents who are required to perform medical tasks without training.

I am passionate about transforming patients' lives through science. One of the major obstacles in developing treatments is the diversity and limited understanding of each disease. The pace of therapeutic development strongly relies on the quality and optimization of preclinical models. These models can ensure improved translation of various hopeful preclinical results into interventions that will ultimately benefit patients. Rodent models of these disorders are thus critical to uncover molecular abnormalities, improve our current knowledge of the disorder and discover therapeutic approaches. By better understanding these mechanisms, I hope to shorten the journey toward diagnosis and improve outcomes for these patients, providing them a better quality of life.