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RARE DISEASES AND ORPHAN DRUGS

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Lifting the burden of rare disease by providing access to next generation sequencing

From the Orphan Drug Act of 1983, a rare disease is a condition that affects fewer than 200,000 people in the United States. In the European Union, the condition must only affect fewer than 1 in 2,000 people. While the numbers seem small, there is an estimated 350 million people that suffer from rare diseases, with 25-30 million belonging to the US alone and so far there have been over 7,000 different rare diseases identified. To put this in perspective, there are more Americans affected by rare disease than for HIV, Heart Disease or Stroke combined. It is important to understand that by nature rare diseases are difficult to diagnose, and consequently are not tracked. Thus, it is hard to accurately determine the number of rare diseases and their impact on a population. The average length of tume from onset of symptoms to an accurate rare disease diagnosis is nearly 5 years, and patients see an average of over 7 different physicians before a diagnosis is made. This delay in diagnosis results in chronic physical, emotional and socioeconomic burden to both the patient and their family. A European Cost of Illness Study interrogating published literature on the cost of 10 selected rare diseases found that overall, the availability of data on economic burden for rare diseases was correlated with the availability of therapies, not the severity of the disease. Also, most rare diseases reviewed were found to have significant economic burden and indirect costs (many associated with loss of productivity) exceeded the level of direct costs. Rare Genomics has served over 500 undiagnosed patients since 2011, helping them access next generation sequencing to accelerate their pathway to a cure. We have seen the same patterns reported for rare diseases in our own patients including heterogeneous disease marked by a range of severity across a variety of biological systems. The most common systems affected are neurologic, respiratory, gastrointestinal, muscular and cardiovascular. The average RG patient has also seen a range of physicians, the top three specialties consulted are: neurologist, clinical geneticist, opthamologist and gastroenterologist. Lastly, undiagnosed/rare disease patients typically have already undergone a gamete of testing, the most common tests are: MRI, DNA microarray and single/panel sequencing. Because 80% of rare disease are genetic in origin, we hope that by providing support and access to next generation sequencing, we can help reduce the time and burden these families must undergo before identifying appropriate treatment for their disease.

Biography

Romina Ortiz completed her Bachelor's and Master's degrees at Johns Hopkins University in Neuroscience and Molecular Microbiology and Immunology, respectively. Her thesis focused on the interplay between the microbiome and hormones in autoimmune disease. She is a Co-founder of Rare Genomics Institute (RG), COO and VP of Patient Advocacy at RG. In 2016, she was awarded the Patient Advocacy Leadership award by Sanofi Genzyme. She trains volunteers on the topics of Genetics and Sequencing, directs a Patient Advocacy Program in next generation sequencing and research services to undiagnosed and rare disease patients and currently directs a philanthropic program with Illumina called iHope, offering free whole genome sequencing to children in need of a diagnosis. Her goal is to integrate phenotypic medical information with genomic data to identify the genetic cause to disease and accelerate the path to treatment and a cure.

Notes:

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