Children with Rare Diseases – From Orphans of Medicine to Pioneers of Personalized Precision Medicine

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Children with rare diseases and their families face countless disadvantages. Many endure years-long odysseys before receiving an accurate diagnosis. Even though we now know of more than 8,000 rare diseases, for many of them, precise diagnostic tools are still lacking. Even more crucially, there are still no curative therapies available for most rare diseases. The clinical care situation and the holistic support for children with rare diseases are becoming ever more challenging, especially as resources in healthcare continue to shrink.

But there is hope: Thanks to major advances in genome sequencing technology and computer science, it is now possible to read and analyze all the genes of a child within just a few days. Tireless work in basic research has also produced innovative new methods in cell and molecular biology that can be used to correct disease-causing processes. Model systems for human diseases allow us to conduct essential preclinical studies in tissue cultures. These findings aren't just important for children with rare diseases—they also have a significant impact on adult patients with more common diseases.

Given all this, the hope for cures has never been as justified as it is today. However, children with rare diseases are now, more than ever, dependent on a wave of solidarity. Without active support from civil society, philanthropists, businesses, and non-profit organizations, pediatric medicine in Germany and around the world cannot fulfill the hopes of these seriously ill children.