

## Perspective

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# Navigating Rare Diseases

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Rare diseases have more similarities than differences. Even though there may be different etiologies and symptoms, the care and needs of the patients, caregivers, medical and allied health professionals are extensive and very similar.

Many rare diseases are faced with the challenge of low prevalence and therefore are positioned as a low priority. They suffer from a lack of funding, education and experience by the medical and allied health communities. Because of these limitations and the complexities of diagnostics and insurance restrictions, many professionals may only be able to offer shrugged shoulders and referrals instead of answers and treatments. In many cases doctors are only treating patient's symptoms and not the cause, and far too often when a scientifically supported diagnosis is confirmed it is followed by the emotionally debilitating comment; "There is no cure."

An estimated 30 million people in the US (1 in 10 Americans or 10% of the U.S. population) along with 30 million Europeans are living with rare diseases. And with an estimated 350 million people worldwide suffering from rare diseases, demanding that we reframe the way we look, define and treat the rare disease community in an uncertain healthcare landscape, has become a necessity.

An increased need for partnerships between patient advocates and the medical community in the world of rare diseases is greater today than ever before. According to the National Organization of Rare Disorders (NORD), 95% of rare diseases do not have one single FDA approved drug for treatment. During the first 25 years of the Orphan Drug Act (passed in 1983), only 326 new drugs were approved by the FDA and brought to market for all rare disease patients combined. 1000 newly identified compounds will be narrowed to 100 in 3–6 years via pre-clinical investigations. In 6–9 more years, those 100 will be narrowed to 10 via clinical trials and then the FDA review process will approve one of those ten in the next 2-3 years with a post mandatory surveillance that goes on indefinitely. I am not inferring that the process is not necessary, but only that the results of the process, and the fact that from drug discovery to developing a new medicine takes 10–15 years and \$800M-\$1.2B is not meeting the needs of the rare disease community.

Rare diseases by their nature are complex medical cases and complex medical cases demand a more focused and coordinated team approach. An approach that goes beyond standard diagnostics and treatments. Patients, caregivers and physicians need to have an interest and willingness to dedicate the time to understand these diseases and they need the tools to develop standard diagnostics and more effective treatments.

Many diagnostic labs have differing methodologies and matrixes that present differing results and opinions on test samples; they are not wrong and they are not right, they are in need of standards. We must bring all the providers together to share and communicate ideas and experiences in order to reach more accurate diagnostics and effective treatments.

Sydney Harris said, “Communication and information are often used interchangeably, but they signify two quite different meanings. Information is giving it out and Communication is getting it through.”

The journal, *Translational Research in Rare Diseases (TRD)*, represents a major step forward enhancing communication and disseminating accurate information for both the lay and medical communities. It is a major resource supporting and benefitting the rare disease community and TRD is an effective tool helping to bridge the gap between shrugged shoulders and accurate diagnostics and effective treatments.

I applaud and thank the efforts of the editors and contributors to this journal helping patients, families, caregivers, researchers and physicians navigate through the complicated maze of rare diseases.