

Rare Disease Research: The Importance of Precision Medicine and Real-World Data (RWD)

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Innovative, disruptive and emerging applications of Real-world Evidence (RWE) are required to advance the quest for new treatments and care guidelines for patients with rare diseases. While the numbers of rare disease patients with a specific recognized condition are small, there are estimated 350 million diagnosed individuals around the world.



One in ten people are affected by a rare disease



50% of people with a rare disease are children



95% of rare diseases lack approved treatment



Over 7000 rare diseases smaller disease populations with disbursed geographic locations

This is where RWD comes in. Using high quality data that is based on globally accepted data standards. This often involves conducting small clinical trials, albeit ideal, the feasibility depends upon finding patients and classifying these patients into subpopulations that differ in susceptibility to a disease or their response to a specific treatment [1]. Precision medicine requires an exactness of science to best target populations that can benefit.

The Quest for Real-World Evidence (RWE) to accelerate research and drug development

Create a new, convenient, and seamless pathway for capturing valuable data, from multiple data sources to develop a comprehensive view of the impact of medical conditions



Patient reported outcomes and natural history

Real-World Evidence Requirements:

- ✓ High quality data
- ✓ Input from a wider range of patients
- ✓ Patient engagement for continued input to natural histories
- ✓ Integrating multiple RWD sources

Real-World Evidence Use Cases:

- ✓ Enable decentralized digital studies
- ✓ Accelerate research and Access
- ✓ Advance new product development and clinical guidelines



Disease/clinical specific data



Genetic & Lab information



EHR



Claims & Billing

An end to end solution to foster collaboration and answer complex research, statistical and economical questions



- Accelerate scientific research and advancing the delivery of new products to market
- Promote global community alliance, enabling collaboration through data sharing across boundaries
- Gain support and interest for funding drug trials.



- Integrate a wide variety of healthcare data sources, providing greater insight into the disease burden of patients living with rare diseases
- Simplified data entry workflows including wearable integration to increase patient engagement & data completion rate
- Supporting natural history studies, clinical disease and patient reported outcomes and journeys for the acceleration of product development



- Generate clinical trial ready data that is GDPR, HIPAA and 21CFR part 11 compliant
- Identify new biomarkers for a condition or across conditions. Assess the viability of drug repurposing for rare diseases
- Establishing reimbursement, cost of therapy, and pricing strategies



References

1. Ginsburg GS, Phillips KA. Precision Medicine: From Science to Value. Health Aff (Millwood). 2018 May;37(5):694-701. doi: 10.1377/hlthaff.2017.1624.