

RECOMMENDATIONS TO ADVANCING RESEARCH IN RARE DISEASE WHITE SPOTS

INTRODUCTION

Rare diseases, defined by their low prevalence, pose significant challenges in research, diagnosis, and treatment. Despite advancements, most rare diseases lack effective therapies, underscoring the necessity for a strategic overhaul in the approach to research and development. To address the multifaceted challenges in rare disease research, a unified framework that encompasses the recommendations across clinical trials, diagnostics, and translational research is proposed. Central to this framework is the role of public-private partnerships in bridging gaps in research and development. By aligning efforts, leveraging resources, and fostering innovation, significant strides can be made towards erasing the "white spots" in rare disease research.

RECOMMENDATIONS CO-CREATION PROCESS

The publication of research needs recommendations for clinical trials, diagnosis, and translational research is the result of several months of dedicated collaboration among members of the rare disease community. The stakeholders consulted during the drafting process included patient representatives; global pharma/biotech companies; small innovative enterprises; academic translational research; biobanking, non-profit clinical and fundamental research communities; biobanking community; and non-profit PPPs. This approach highlighted our collective commitment to advancing research outcomes and addressing the challenges of rare diseases.



RECOMMENDATIONS

OPTIMIZING DIAGNOSTIC RESEARCH

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- The journey towards a definitive diagnosis for rare diseases is often long and challenging, known as a 'diagnostic odyssey'. To
 address this, diagnostic research should focus on increasing diagnostic accuracy, improving time to diagnosis and ensuring wider
 access to cutting-edge diagnostic tools
- Discovering and validating novel biomarkers is crucial, including, molecular biomarkers for identifying disease-specific genetic markers, clinical biomarkers for insights into disease manifestation and digital biomarkers for understanding disease progression
- Advances in technologies are instrumental (genomics, multi-omics) as well as deep phenotyping
- Epidemiological studies should complement these efforts to better understand disease prevalence and distribution and facilitate targeted screening and prevention strategies
- Embracing technology for good can further enhance diagnostic capabilities, including AI and machine learning for data analysis and pattern recognition and enabling earlier and more accurate disease identification

ENHANCING CLINICAL TRIALS

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- Conducting clinical trials for rare diseases faces inherent challenges such as small patient populations, dispersed expertise and complex regulatory pathways
- Addressing these challenges requires a multifaceted approach by bridging translation and regulatory approvals and leveraging regulatory science to develop new trial designs
- Building capacity for implementation is crucial, including infrastructure development, skills development and scaling up and Drug Development Tools (DDTs) in R&D practice
- Maximizing the utility of existing resources is vital through the standardizing and FAIRifying patient-generated data, and the integrating patient data into regulatory-grade sources in R&D plans
- Public-private partnerships (PPPs) serve as a strategic model to catalyze progress to facilitate collaboration across sectors, optimize clinical trials and enhance the probability of success in developing therapies for rare diseases

ACCELERATING TRANSLATIONAL RESEARCH

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- Translational research in rare diseases faces the challenge of navigating the 'valley of death' and potential therapies often fail to progress from discovery to clinical application
- Overcoming this challenge requires a robust collaborative framework involving public-private initiatives by leveraging complementary strengths of stakeholders across the spectrum, developing a shared vision focused on patient-centric approaches and ensuring structural access to comprehensive expertise across the value chain
- Investing in innovative methodologies and fostering standardization and awareness efforts are crucial by creating cross- sectoral
 accelerators and enhancing Technology Transfer Offices (TTOs) which can facilitate effective technology transfer and innovation
 processes to bring scientific breakthroughs closer to clinical application
- Engaging with regulators, payers, and clinicians structurally and in a timely fashion ensures research design is optimized for translation into a healthcare setting
- Adopting targeted public-private interventions can significantly improve the translational research ecosystem for rare diseases to foster deep collaboration and facilitate the journey of scientific discoveries from the bench to the bedside