

Single Day Event

Innovations Advancing Rare Diseases Research

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Precision for Medicine



Rare Disease - Is it a paradox

7000+

Identified rare diseases, with more being discovered every day

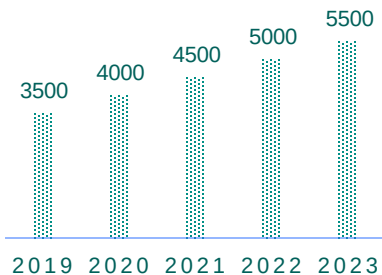


80% of rare diseases have a genetic cause; **70%** of which present in childhood; about **95%** lack approved treatments

Effect

350M humans worldwide

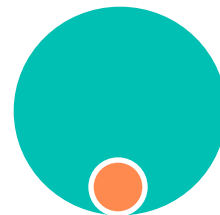
Studies



Market

Global clinical trials market 2022

\$77.27B



\$11.5B
Rare disease

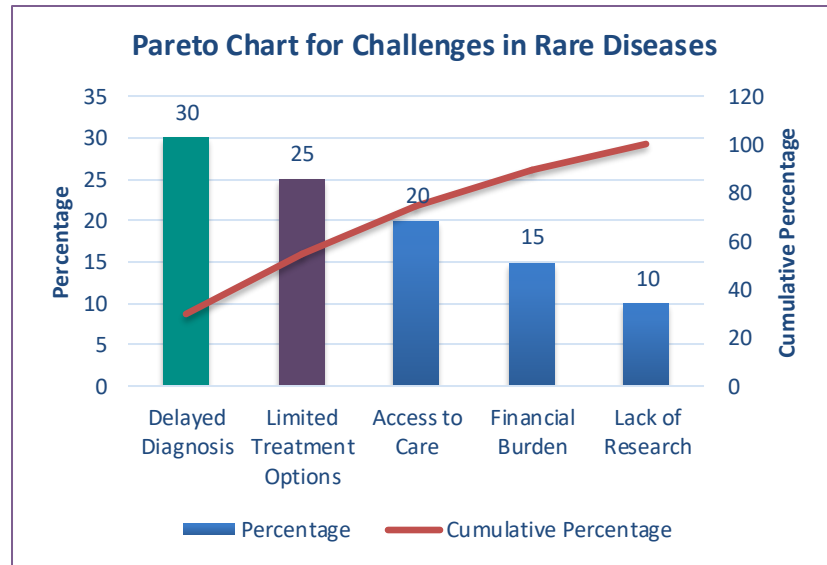
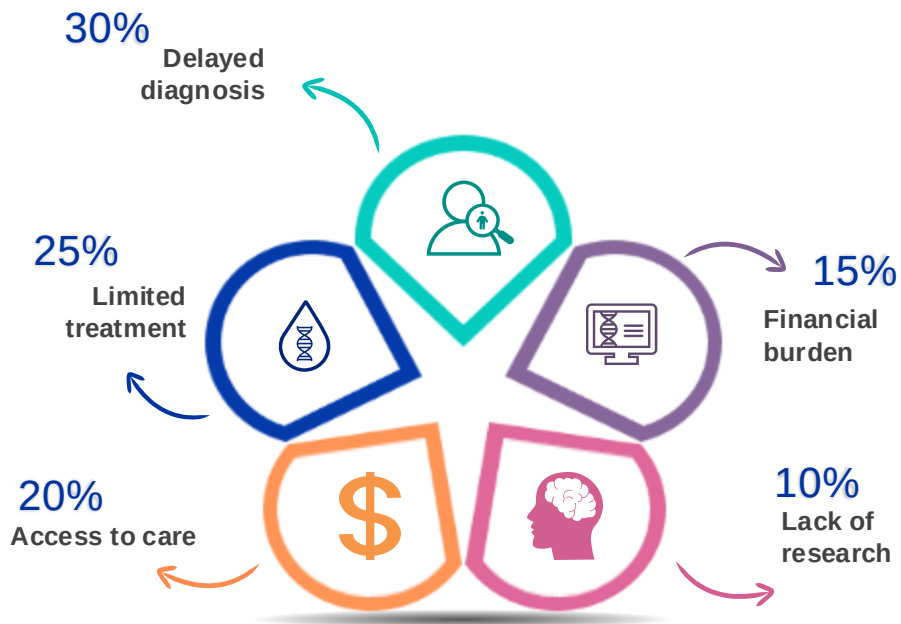
Top 5

- Rare Neoplastic disease – 37%
- Rare Neurologic disease – 18%
- Rare inborn errors of metabolism- 11%
- Rare Hematologic disease – 8%
- Rare Respiratory disease – 5%

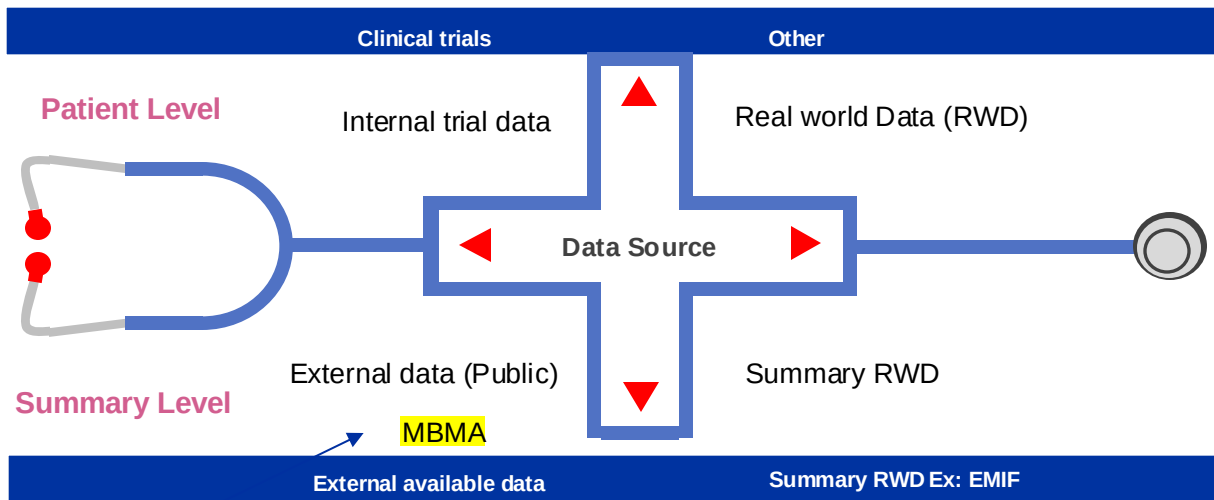


Challenges in Rare Disease

National Organization for Rare Disorders (NORD):



Model-based meta-analysis (MBMA) is a quantitative approach that leverages published summary data along with internal data and can be applied to inform key drug development decisions, including the benefit-risk assessment of a treatment under investigation. **Establishing a dose- or exposure–response (E-R) relationship is critical to inform early go/no-go development decisions**



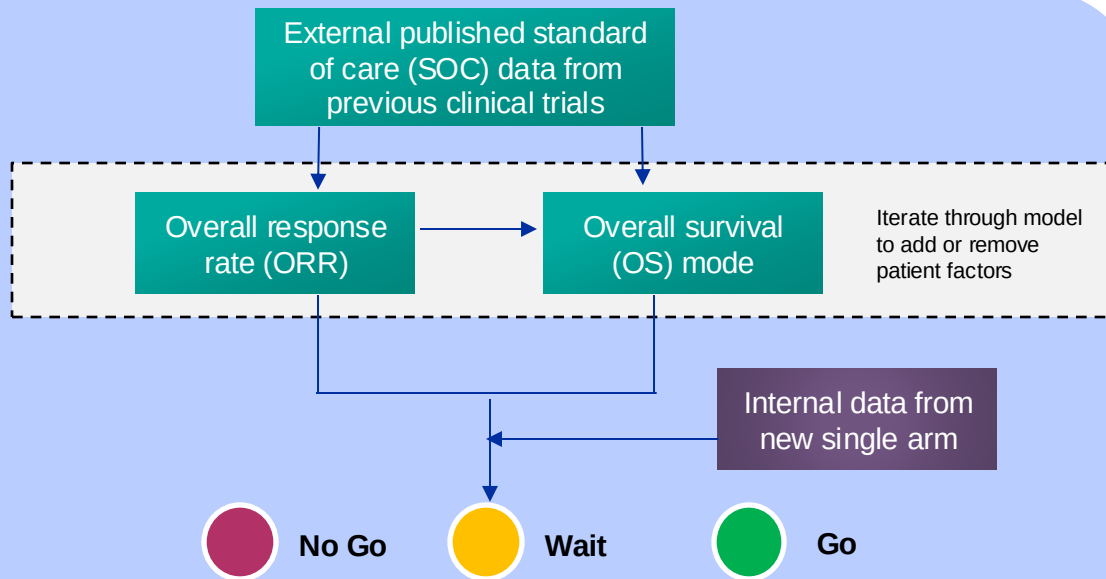
EMIF: External memory interface

Case Study 1: Model-based meta-analysis (MBMA)



Objective: To support early development decisions for PD-1 inhibitors and chemotherapy in metastatic NSCLC.

Method



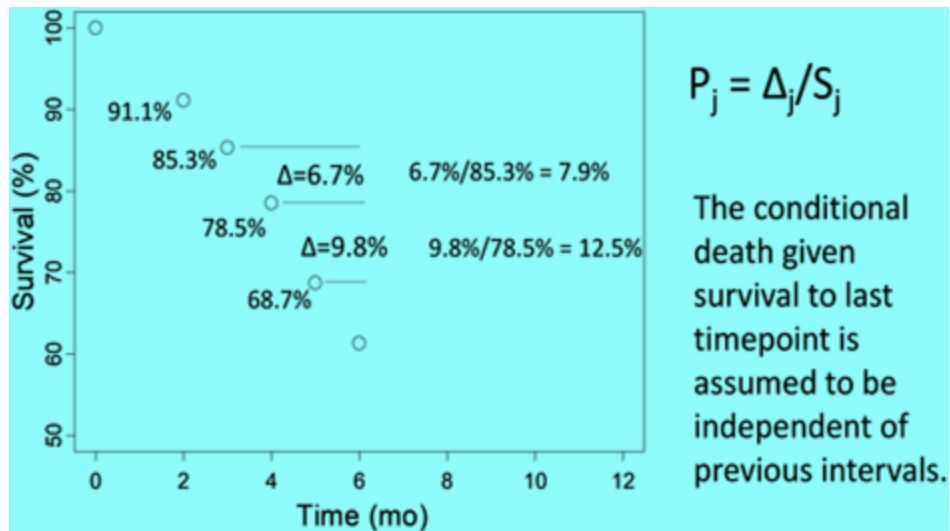
The MBMA simulations predicted OS hazard ratios as a function of ORR, adjusted for matched patient characteristics.

These predictions supported early development decisions, such as go/no-go decisions and futility rules for new treatments



Case Study 1: cont...

The graph illustrates the survival percentage over time (in months) for a specific population.



Y-Axis: Survival percentage (%), X-Axis: Time in months (mo)

Survival (%)

Conclusion

- At 0 months, the survival rate is 91.1%.
- At approximately 2 months, the survival rate drops to 85.3%, indicating a decrease of $\Delta=6.7\%$.
- At approximately 4 months, the survival rate further drops to 78.5%, indicating a decrease of $\Delta=6.8\%$.
- At approximately 6 months, the survival rate is 68.7%, indicating a decrease of $\Delta=9.8\%$.

These predictions supported early development decisions, such as go/no-go decisions and futility rules for new treatments



Solution - Access to Care: Decentralized trials

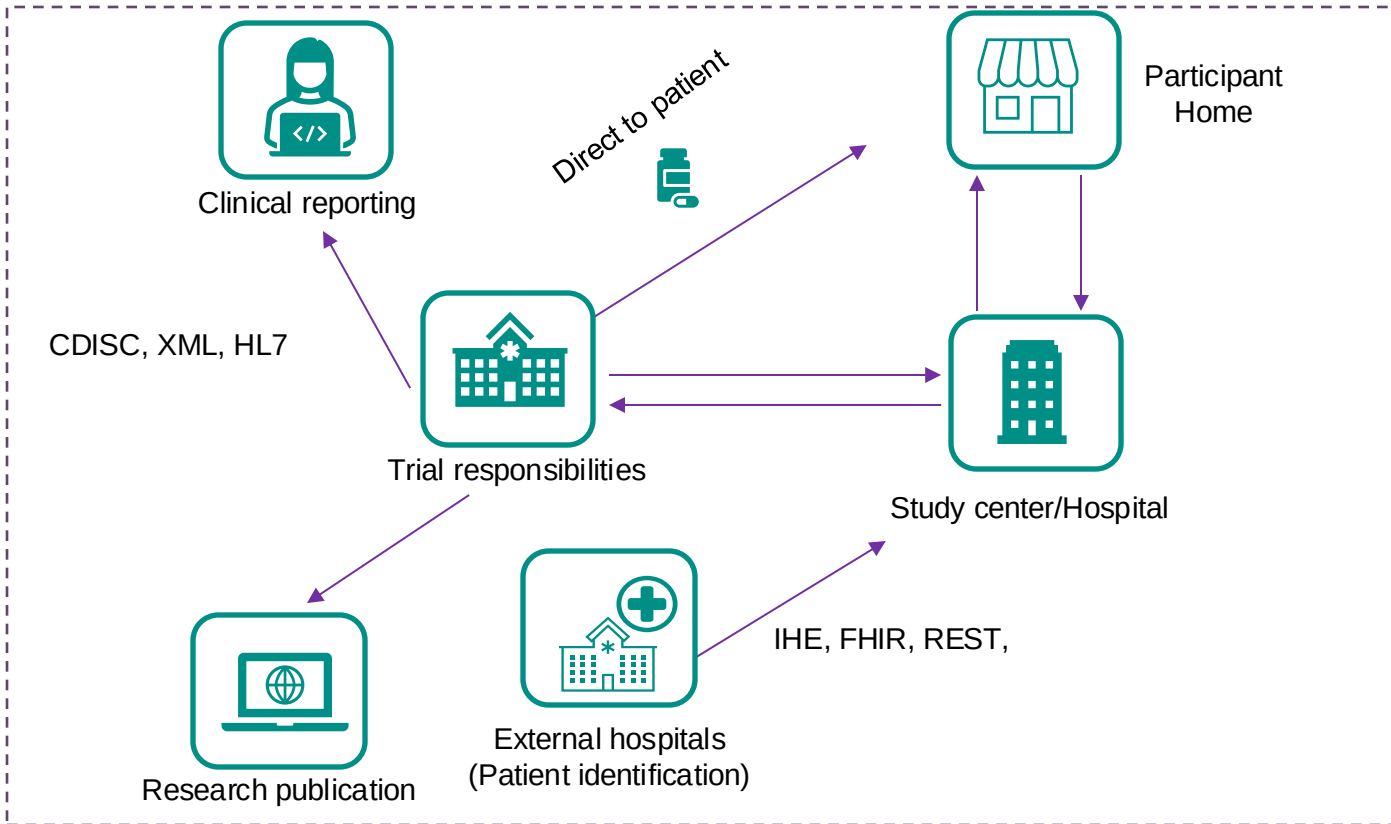
2012

250
studies

2022

1425
studies

Decentralized trials





Case study 2: Patient engagement and care



Address the challenges of patient engagement and retention in a clinical trial for an ultra-rare and aggressive cancer. The trial faced significant hurdles due to the small, dispersed patient population and the complexity of the disease



Predictive Analytics: Predictive analytics tools forecasted which patients were at higher risk of dropping out, enabling proactive engagement with targeted interventions



Engage

Outcome

**Personalized
Communication**

Support Programs

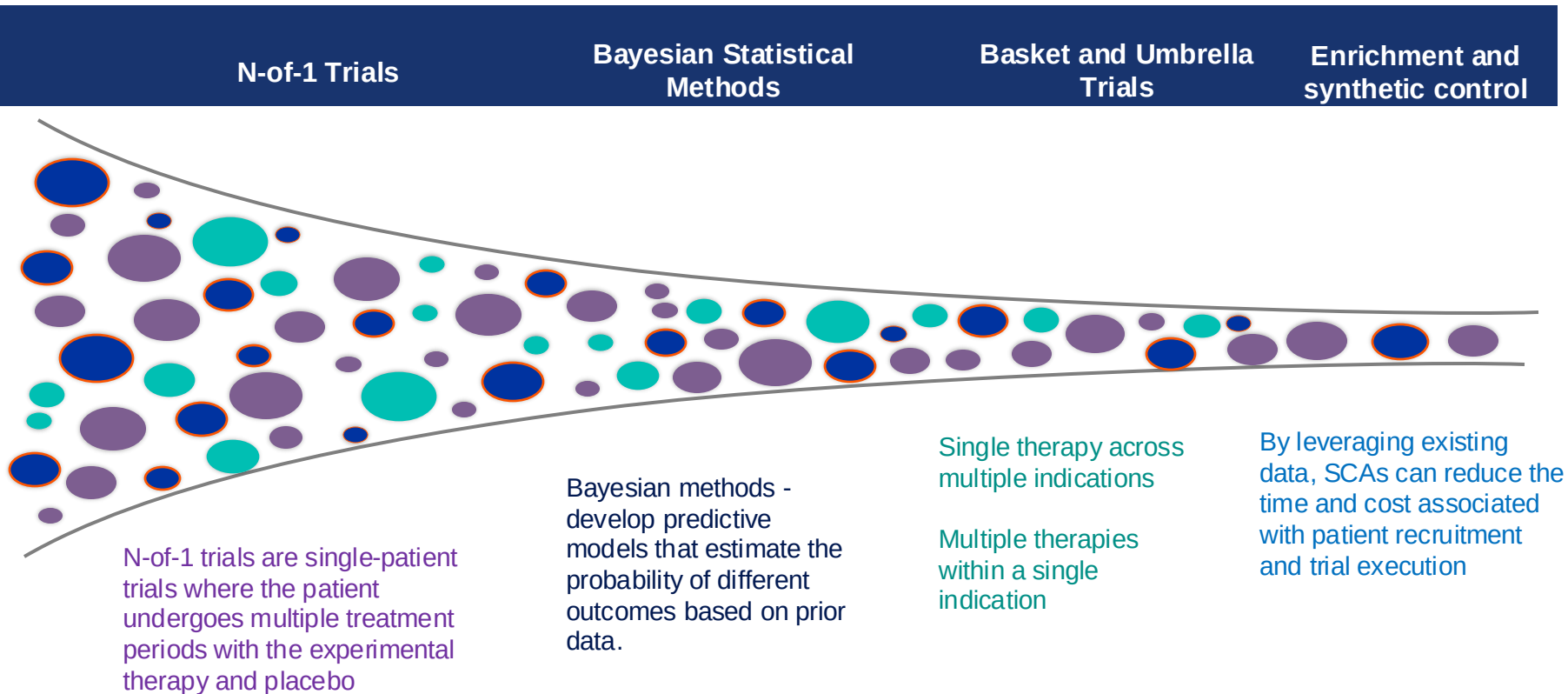
Flexible Scheduling

The tailored interventions and real-time monitoring significantly improved patient retention rates.

Timely interventions, reducing dropout rates and ensuring the collection of comprehensive data



Other strategies for rare disease





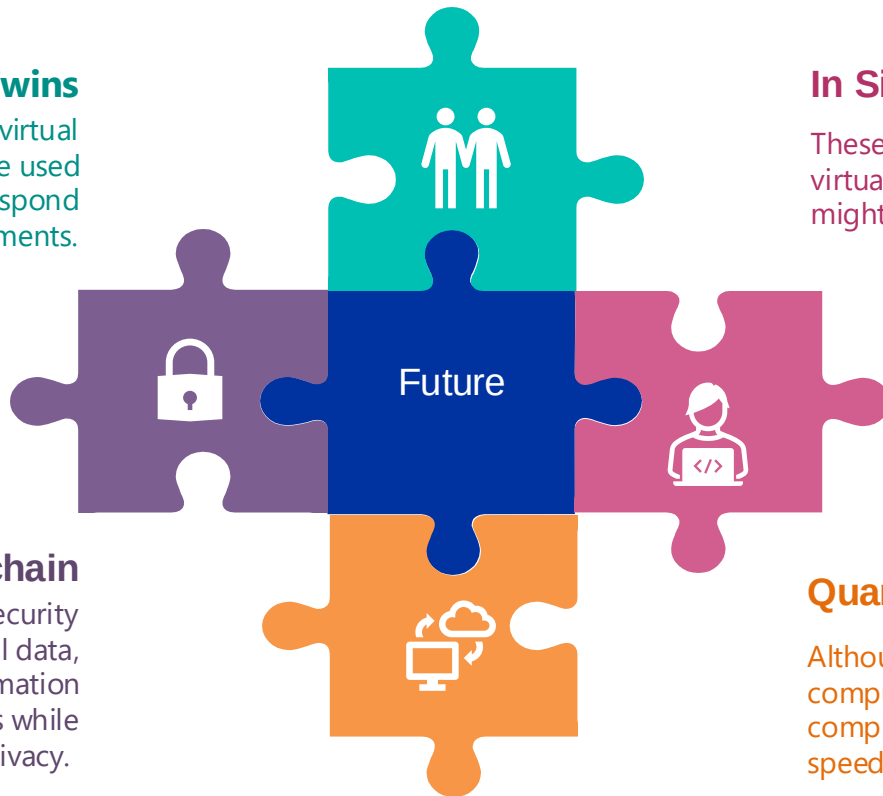
Future trends paving way to solve rare disease mysteries

Digital Twins

This involves creating a virtual model of a patient that can be used to simulate how they might respond to different treatments.

In Silico Trials

These are computer-simulated trials that use virtual patients to predict how real patients might respond to treatments.



Blockchain

Blockchain can ensure the security and integrity of clinical trial data, making it easier to share information across different stakeholders while maintaining patient privacy.

Quantum Computing

Although still in its early stages, quantum computing has the potential to process complex biological data at unprecedented speeds.

Making Data the Drug...?

Thank you 😊



**The Global Healthcare
Data Science Community**

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