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OPEN PLATFORM FOR ORPHAN DISEASES

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No Disease Orphan

SDG # 18

Technology, Economics and Policy for Providing Equitable Healthcare Amidst Extreme Inequality

Sustainable Development Goal Number 18: NO DISEASE ORPHAN

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In September 2015, the UN General Assembly adopted the **2030 Agenda** for Sustainable Development that includes **17 Sustainable Development Goals (SDGs)**. Building on the principle of "**leaving no one behind**", the new Agenda emphasizes a holistic approach to achieving sustainable development for all.

Time to add #18 to the list – **No Disease Orphan**.

We need to promote an active dialogue among stakeholders in orphan diseases with a view to create a better world for persons with these disabilities. Most important to support the extraordinary heroic efforts by patients and their caregivers, such as the group at DART, that give us hope that the audacious goal of "no diseases orphan" is attainable

Problem Statement

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- In developing economies, rare diseases have received little to no attention in policy as well as practice, making them orphan diseases
  - Examples: rare cancers, hemophilia, hemoglobinopathies, ophthalmic, immunodeficiency, neuromuscular disorders, etc.
- In India, a policy for Treatment of Rare Diseases was released in 2017 and revised and published as the National Policy for Rare Diseases 2020 by the Ministry of Health and Family Welfare, India.
  - Disease burden in India ~50-70 Million children and adults
- How can we size the response in policy and practice when gaining the system level understanding of Equitable Access, Accurate Diagnostic, and Affordable Treatment remain illusive?
  - Total afflicted population
  - Prevalence
  - Frequency of occurrence

Model the Systems' Dynamics

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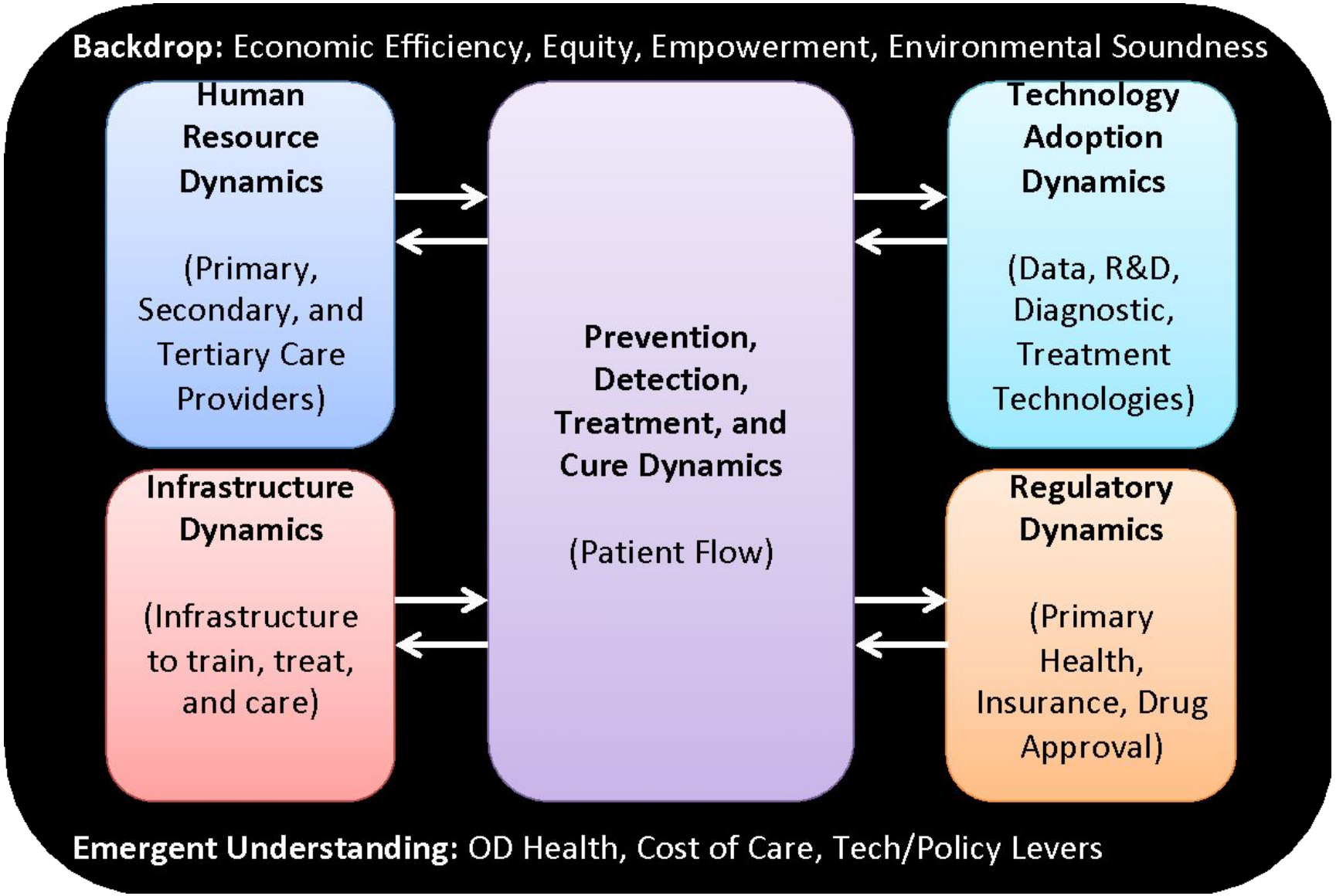

Turning Levers in the Model: Progress, Insights and Questions

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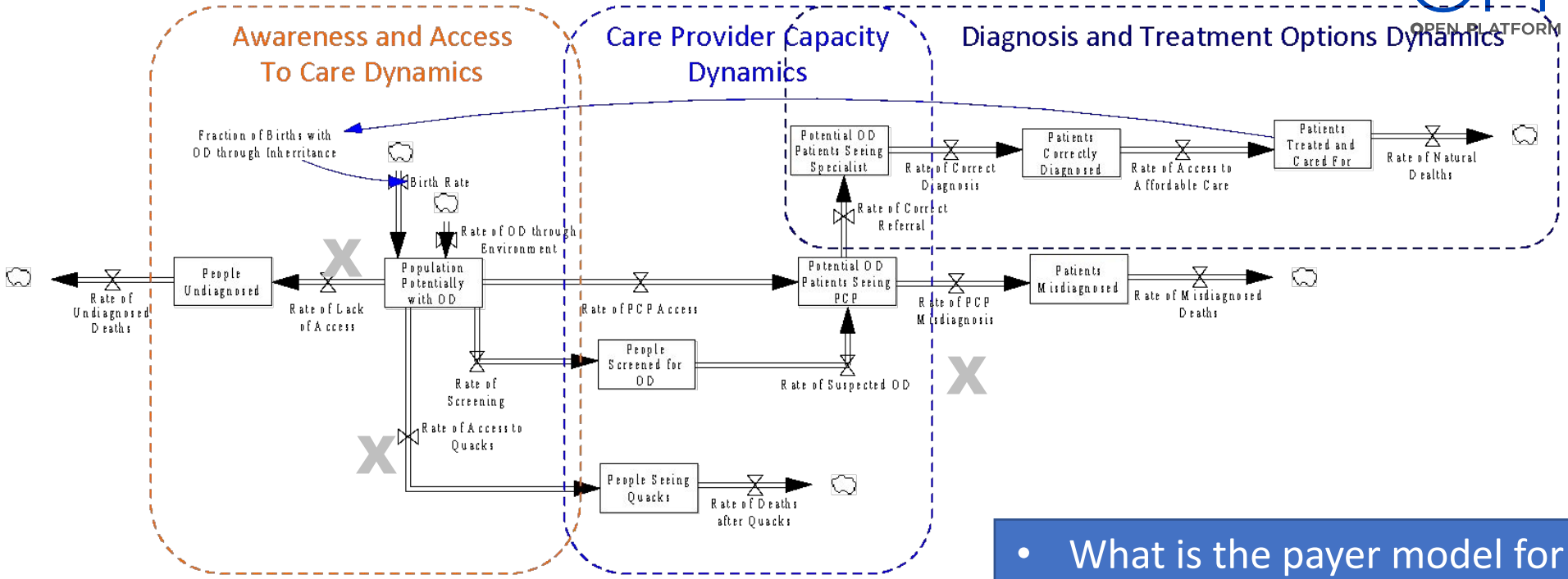

- **Three Foci**
  - Equitable Access
  - Accurate Diagnosis
  - Affordable Treatment
- **OPFORD Foundation**
  - Focus on haematalogical genetic disorders
  - Patient centred digital platforms

- What is the payer model for orphan diseases in your health system – for diagnosis, maintenance, cures by genomic medicine?
- What is the disease burden? Has Hardy-Weinberg Equilibrium been reached in genotype frequencies?

A PATIENT CENTRED CLINICAL DECISION SUPPORT SYSTEM

The system consists of a consumer part (green), a clinical part (turquoise) and a hybrid intelligent system (blue) that are able to communicate.

The framework is designed to enable patients and physicians to exchange health and research data, access valid medical knowledge and use reviewed reasoning technology while ensuring the quality of the components, auditability and a learning system.

Hirsch et al., Rare diseases 2030: how augmented AI will support diagnosis and treatment of rare diseases in the future BMJ 2020

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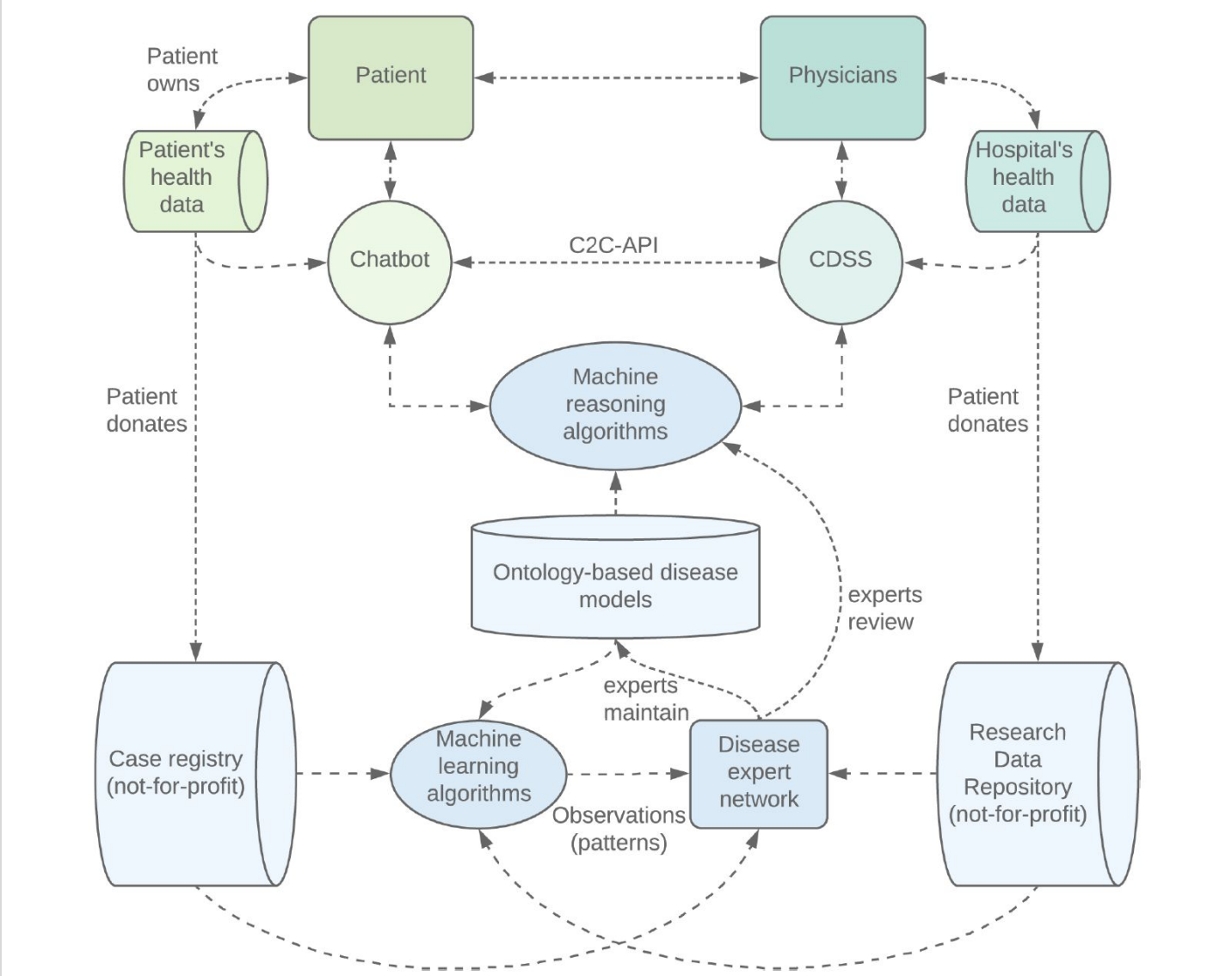

The future of Genomic Medicines

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Genomic Drug Discovery

A New Paradigm

Pioneered by Deep Genomics

University of Toronto

OligoTherapies selected by an Intelligent Engine

- **Training Data:** a comprehensive set of genetic variants and their predicted effects on human splicing across the entire genome
- **Deep Genomics:** Its first success story appears to be a treatment for Wilson disease in patients who possess Met645Arg, a mutation shown to lead to the loss of function of the ATP7B copper-binding protein. DG12P1 is the oligonucleotide therapy that caused approximately 70% skipping of exon 6. That exon skipping in turn resulted in frameshift and stop gain, which is expected to cause loss of ATP7B function.
- *It is important to note that the **ASO approach** taken by DART and its sister concern Hanugen can now be scaled to have a much larger impact using these methods.*