CASE STUDY: Pillar 2 Equipping Providers
UTOPIA (Unlocking Treatment Options Personalized In-Time Access)
Australia and Singapore

A digital platform enabling care and support for those with diagnosed and undiagnosed rare diseases

Context

Receiving an early and accurate diagnosis is imperative for informed clinical care, yet those living with a suspected rare disease will often spend more than five years on a diagnostic odyssey. Regrettably, for many children, the search for a diagnosis will ultimately be futile: **more than 50% do not receive a molecular diagnosis.**[1]

That is why devising care pathways for both diagnosed and undiagnosed patients is essential to improve clinical outcomes, provide emotional relief, and enable appropriate access to resources. Digital tools are emerging as quick, cost-effective solutions to help accelerate the path to diagnosis and care in many health areas, including in rare disease.

Photo credit to Perth Children's Hospital Foundation and Kylie Plunkett Photography

UTOPIA

Overview of program

In 2022, **The Rare Care Centre at Perth Children's Hospital** in Western Australia and KK Hospital in Singapore co-designed a digital platform to generate a semi-automated summary of a patient's condition called UTOPIA (Unlocking Treatment Options Personalized In-Time Access). UTOPIA's power is the ability to help determine the care a child needs even if a diagnosis has not been confirmed — streamlining the care pathway during the lengthy diagnostic journey. UTOPIA also advances equity by enabling children without access to more advanced diagnostics to begin receiving the care they need.

Based on an individual's clinical features (phenotype), the platform deploys a wide range of artificial intelligence approaches for analysis, including large language models and new concept recognition, which is more **accurate, faster, and less costly than other AI approaches**. The information generated is then filtered against large data sets of diseases. Medical experts review the output and produce a patient profile, including the likely trajectory of the disease's evolution and how it may affect the child physically and behaviorally.

This pairing of "doctor and machine" is enabling health providers to guide the type of care and support a child should receive while awaiting test results or when a diagnosis cannot be established. UTOPIA offers a targeted, informed and scalable approach to deliver individualized models of care for children living with a rare disease — regardless of whether they have a diagnosis.

Although early in its implementation, UTOPIA's major benefit is its ability to help families avoid unnecessary interventions and connect to care, support, clinical trials and research. Potentially, UTOPIA could help prevent the progression of a condition and reduce its burden even before an accurate diagnosis is possible.

In some cases, UTOPIA can bridge the gap in lack of access to full genome sequencing because it produces care plans that are based on an individual's clinical features (phenotype) — saving significant time and resources for families and health systems throughout the diagnostic odyssey.

For example, if a child has cerebral palsy, heart abnormalities and hearing loss, it is extremely likely that there is an underlying rare genetic disease. By capturing what is known about these individual conditions and how they inter-relate and evolve over time, UTOPIA is increasingly able to help create personalized plans for health, education, disability and social services — even without knowing the exact genetic cause.

Understanding the trajectory of a child's rare disorder, the associated phenotypes and the potential psychological effects can guide the development of comprehensive support systems to address a child's unique needs. For example, educators can adapt teaching strategies, provide necessary accommodations, and offer appropriate support to optimize the learning experience. Likewise, this knowledge can inform the development of appropriate rehabilitation strategies and assistive technology for children facing physical disabilities.

The reports UTOPIA provides are easy to understand because of accessible language and visual aids. As a result, UTOPIA facilitates communication between the care team and the child's family as well as between the family and other services providers, such as disability agencies and the education system.

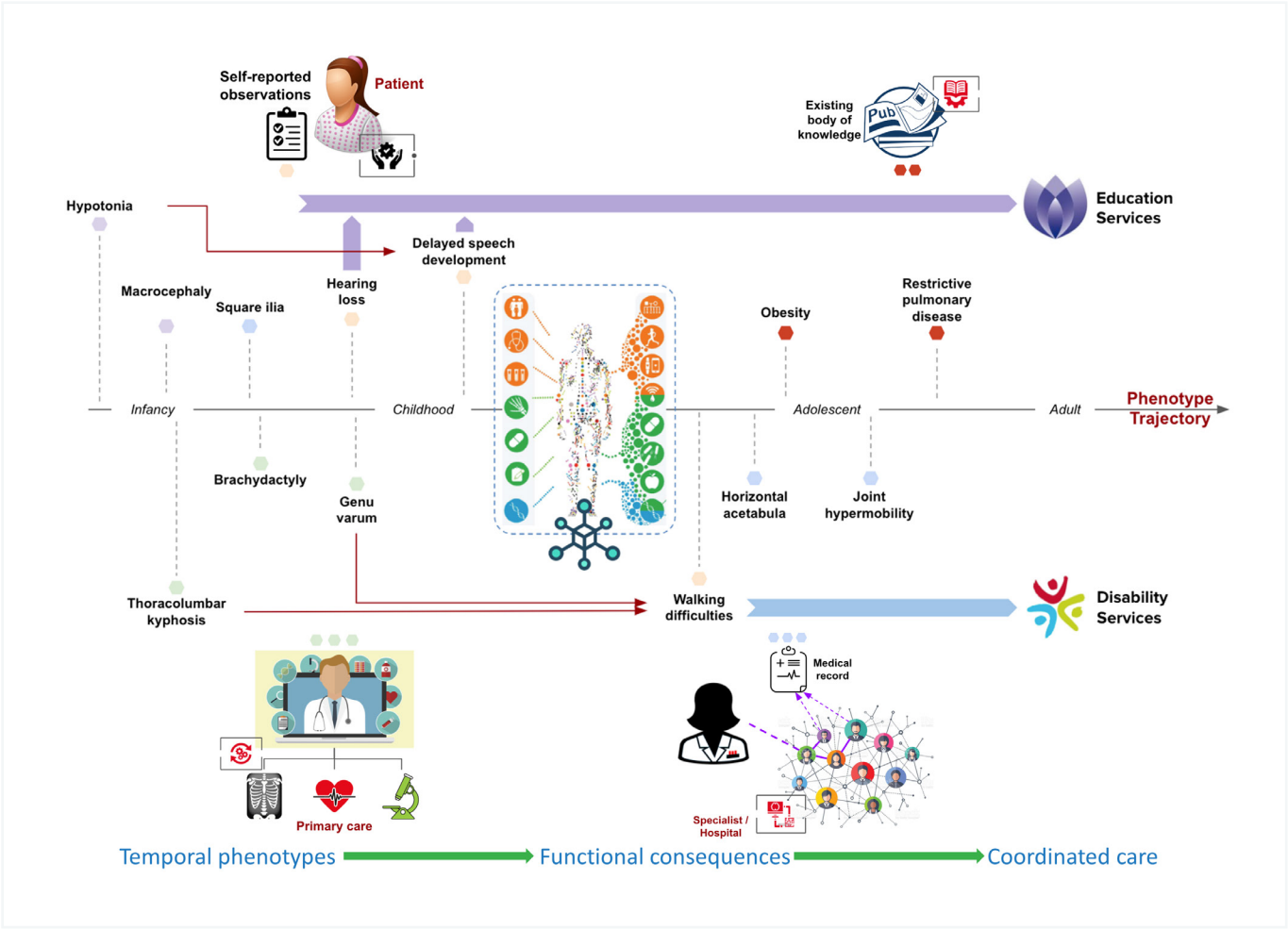


Figure 1: A "Phenotype trajectory" is a predictive mapping based on data from UTOPIA of how a particular rare disease for an individual evolves over one's lifetime and manifests externally. In this case, the patient starts with presenting with hypotonia (floppiness) as a newborn and progresses to show further clinical features (phenotype) over time, such as a curved spine (thoracolumbar scoliosis), developmental delay (motor and speech delay) and later obesity and lung disease (restrictive pulmonary disease). Knowing the likelihood and timing of development of these clinical features enables better prediction, prevention and treatment planning.

Both primary and specialty care clinicians have access to UTOPIA and can deploy it throughout the entire patient journey. The platform is currently active in the public health system in Australia (Rare Care Centre, Perth Children's Hospital) and Singapore (SingHealth, the KK Hospital for Women and Children). UTOPIA is also being integrated into Genomics 4 Kids in ASEAN (G4K), formally known as BRIDGE-ASEAN, a multi-country initiative in Southeast Asia to advance genomic diagnosis and care of patients with rare diseases across Southeast Asia.

Results

UTOPIA has cut in half the time to develop a personalized care plan by reducing the number of hours that clinicians otherwise would have to spend finding the relevant information, tailoring it to the individual child and presenting it in an easy-to-understand format. The platform's designers anticipate that UTOPIA will soon be able to create these care plans 10 times faster by transforming a traditionally manual process involving several health care professionals to a technology-driven, semi-automated system.

UTOPIA's speed in guiding the development of personalized cross-sector care pathways continues to increase with better access to relevant data sets, more advanced algorithms and greater integration of the platform into clinical workflow.

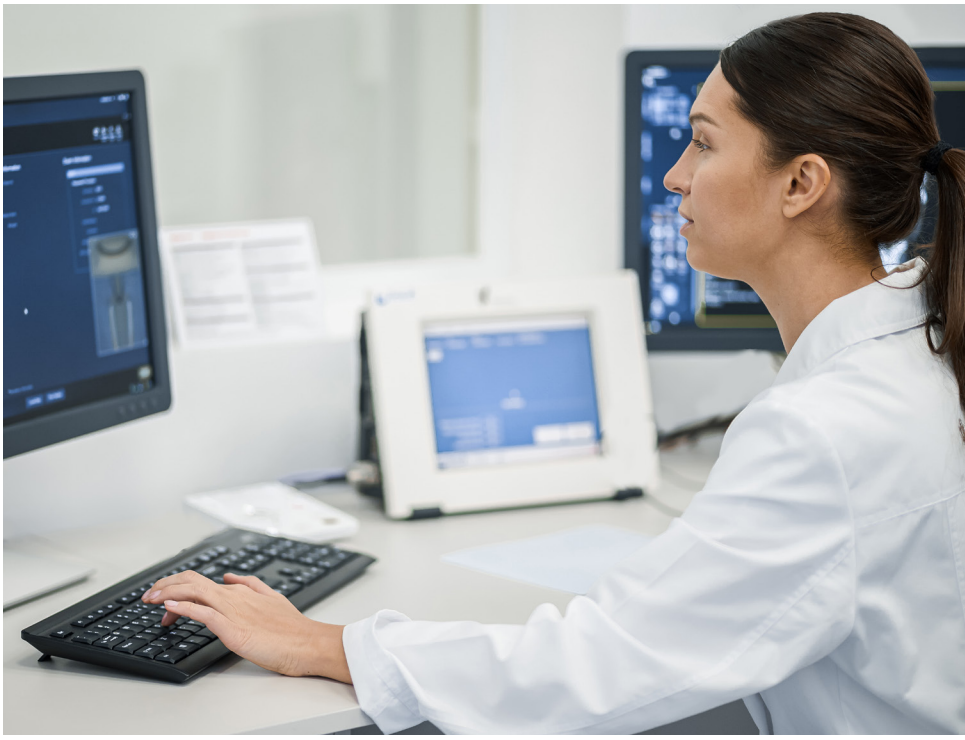
UTOPIA is being implemented for daily use in the clinical care of patients in the Rare Care Centre in Australia and approximately 180 patients, as of July 2024, have received reports that are guiding their individualized care plan.

In Singapore, UTOPIA is being adopted throughout the country. Currently, the platform is being used to better understand how a group of rare diseases called RASopathies evolve over time and how best to provide care and support to people living with these diseases.

Families have reported that UTOPIA-generated reports are the first time they have had access to an easy-to-understand and tailored plan that helps connect them to care across multiple systems, including health, education, disability and community services. A formal evaluation of UTOPIA's value to both patients and clinicians is in progress.

Learnings

- **Develop a solid understanding of the health care system before introducing a new tool:** Given vast differences in health systems, it is imperative — at the development stage — to have a clear sense of how a product and its implementation should be tailored to a health care setting to ensure smooth integration. Avoid developing a product externally and then trying to make it fit. UTOPIA was designed with a deep knowledge of the nuances of the Western Australian and Singaporean health systems where it would be tested.
- **Regional partnerships encourage more efficient implementation:** The UTOPIA team developed partnerships with multiple health jurisdictions, each with different strengths. The agility required to meet institutions' varied needs and capabilities ultimately led to implementing UTOPIA more quickly across different sites.
- **A tool intended for physicians should also have value for patients and families — and vice versa:** Unlocking the power of information from patients and physicians amplifies the impact. The UTOPIA team was intentional in designing ways for patients and families to provide their information and generating reports that patients and families could easily understand. Accessible language and visual aids based on health literacy principles have been important to ensure that medical information is communicated in a way that families can understand and interpret for non-health service providers who work with their children.



Goals & next steps

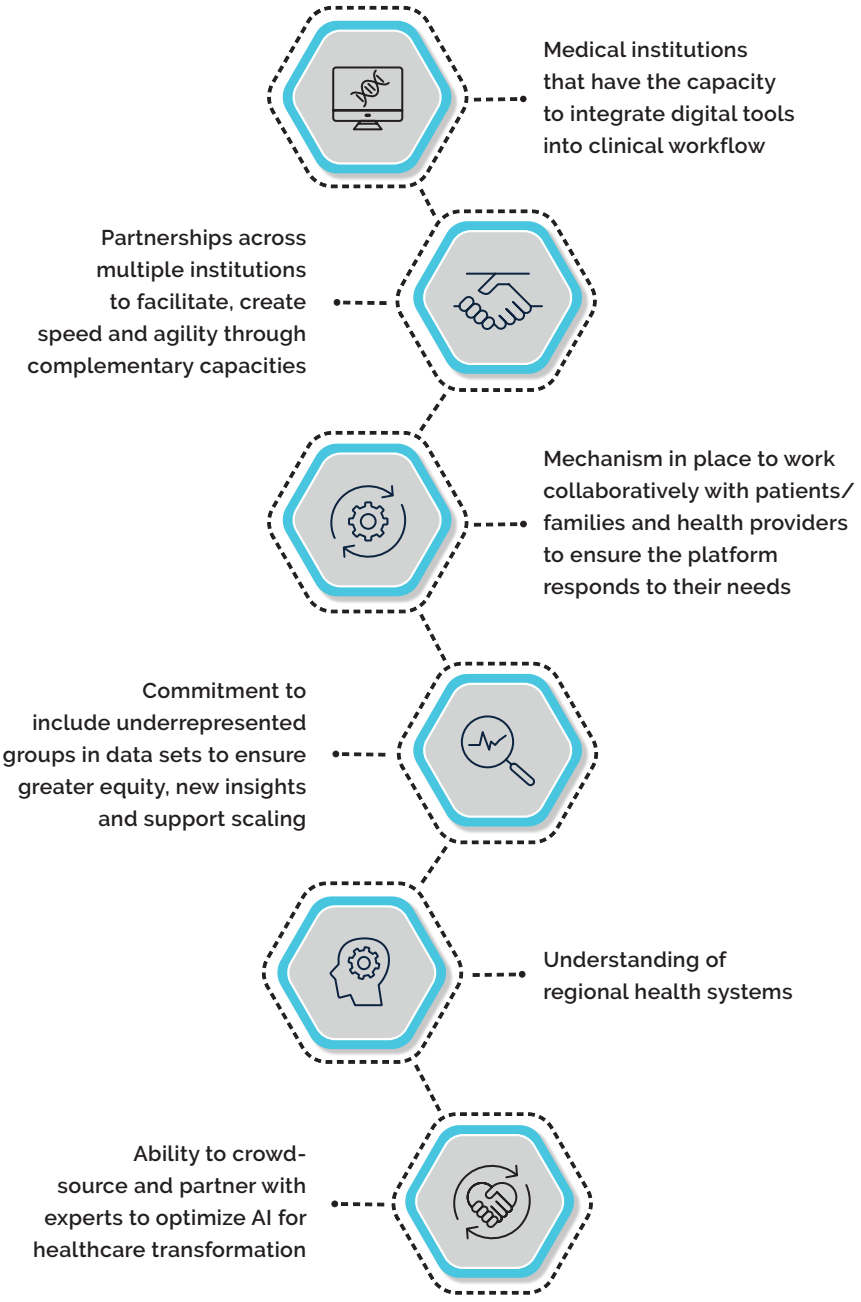
UTOPIA can be implemented in various ways, depending on data systems and resources. The platform is built to be compatible with electronic health records while also being flexible to adapt to settings in which hospital data sets are disparate and not integrated into one electronic health record.

The UTOPIA team aims to continuously strengthen the platform by increasing access to more datasets from genomic testing results and tailoring its approaches to be more culturally safe and responsive. Given that language is central to culture, UTOPIA will integrate with **Lyfe Languages**, an Indigenous medical translation solution. UTOPIA is also partnering with **Cliniface**, a 3D facial analysis software that supports diagnosis and treatment monitoring of rare diseases.

In the long term, the team at UTOPIA plans to apply learnings from its AI model on rare and undiagnosed diseases to a broader set of rare and common diseases.

In terms of geographic expansion, a goal is to enhance regional capacity – including at the community, state and national level – to use the tool to provide more timely care for people living with a diagnosed or not yet diagnosed rare disease.

Success factors & considerations



References

1. A three-year follow-up study evaluating clinical utility of exome sequencing and diagnostic potential of reanalysis | npj Genomic Medicine (nature.com)