# RARE ANSWERS™

A system of innovative and sustainable tools that have potential to unlock the power of medical information to deliver answers for some of the most vulnerable rare disease patients: children.

## THE PROBLEM

### People with rare diseases often wait years to reach the correct diagnosis

**We have more medical data today than we ever have, but much of it is untapped, disorganized, and unusable by patients and physicians.**

**For healthcare providers, when it comes to sequencing data into actionable clinical data, it is easy to get wrong answers. The newest science is always developing and there is a shortage of key specialists. Even when a correct diagnosis is made, providers often do not have access to the best available medical information for rare diseases.**

Genetic disorders and congenital anomalies affect **~6% OF LIVE BIRTHS** and are the leading reason for hospitalization and mortality in infants.

While early diagnosis in infants enables optimal outcomes, it is exceptionally difficult to deliver for rare diseases since there are **MORE THAN 7,000** and presentations are often atypical from classical descriptions.

On average, it takes **4.8 YEARS AND 7.3 SPECIALISTS** before receiving a correct rare disease diagnosis.

## THE SOLUTION

**Developed in collaboration with leading children's hospitals and technology and data-science companies, RARE ANSWERS programs work by analyzing phenotypic and genomic data, incorporating precision software, medical information, and clinical insights to deliver actionable data to physicians.**

**CAPABILITY 1:**
**Provisional diagnosis**

**CAPABILITY 2:**
**NextMove: Guided approach for further patient assessment**

**CAPABILITY 3:**
**Gene-to-Treatment (GTRx): Delivery of expert-curated and validated clinical care pathway***

**Pilots of this approach have achieved world records in reducing the time to diagnosis and treatment for children in NICUs.**

POTENTIAL TO IMPACT RARE DISEASE CARE FOR THOUSANDS OF CHILDREN AND THEIR FAMILIES.

ALEXION HAS ACTED AS STEWARD OF THIS PROGRAM FOR **5 YEARS** INVESTING AS PART OF OUR COMMITMENT TO THE NEEDS OF ALL RARE DISEASE PATIENTS.

* Vetted clinical care pathways have been reviewed by scientific experts convened by our partner institutions. Alexion has had no role in the development of those recommendations beyond helping to curate evidence that has been provided to these experts for their review.

# RACE TO GET TO DIAGNOSIS AND PERSONALIZED TREATMENT PLAN

Treating physician may require access to specialist (e.g. genetics counselor, pathologist) to interpret diagnostic and/or confirm treatment next step (e.g. surgery, gene therapy)


**CAPTURE PHENOTYPE & GENOTYPE****

The diagnostic process begins with RARE ANSWERS extracting a patient's Human Phenotype Ontology (HPO) and Variant Call Format (VCF)

**PROVISIONAL DIAGNOSIS**

Combines HPO, VCF, and RARE ANSWERS™ Rare Disease Map, to gener-ate initial list of potential diagnoses

**CLINICAL NEXT BEST MOVE**

Guides healthcare providers to perform necessary tests and procedures until they reach a confirmed diagnosis

REAL TIME

The patient's evolving digital signature (genotype, phenotype, lab results) is matched against all rare diseases digital signature until clinically confirmed diagnosis. The patient's digital signature and confirmed diagnosis is then matched against vetted treatment options

**MEDICAL REVIEW PROCESS**

**GENE-TO-TREATMENT (GTRX)**

Takes confirmed diagnosis and provides clinical care pathways* that have been curated and validated by clinicians

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**A genotype is the genetic constitution of an individual organism. A phenotype is the set of observable characteristics of an individual resulting from the interaction of its genotype with the environment.