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Precision health for children and adolescents:

RARE DISEASES

Collaboration with Harvard Consulting on Business and the Environment

The Challenge

50-75 percent of rare diseases affect **children**.[1]

70-80 percent of rare diseases are **genetic**.[2]

30 percent of children with a rare disease die **before the age of five**.[3]

Key Takeaways

- There are between 6,000 to 8,000 life-threatening and chronically debilitating rare diseases that affect less than 1 in 2,000 people, according to the European Commission.[4] Only about 5% of rare diseases currently have approved treatments.[5]
- The genomics revolution over the past decade has led to great strides in rare disease research, with gene-editing treatment for sickle cell disease now commercially available.[6]

Context

- There are between 6,000 to 8,000 life-threatening and chronically debilitating rare diseases that affect less than 1 in 2,000 people, according to the European Commission.[7] Only about 5% of rare diseases currently have approved treatments.[8]
- The genomics revolution over the past decade has led to great strides in rare disease research, with gene-editing treatment for sickle cell disease now commercially available.[9]

Findings

- The therapeutic options for rare diseases are limited because diagnosis is difficult and underreported. Rare diseases are characterized by a wide diversity of symptoms and signs that vary from disease to disease and from patient to patient suffering from the same disease. Children with rare diseases tend to be geographically widely distributed. Medical professionals often lack the appropriate training to recognize them.
- While many rare diseases remain incurable, early diagnosis and specific treatment can improve the quality of life of affected children and their caregivers who may otherwise spend years navigating the diagnostic maze, which is both financially and psychologically draining.

Innovation

- Genetic therapies are considered the most promising treatment for rare genetic diseases.
- Genomic technologies, such as the use of whole-exome sequencing (WES) or whole-genome sequencing (WGS) have the potential to curtail the diagnostic process for children with rare diseases, often avoiding the need for invasive and expensive investigations (Figure 1).[10]
- The Generation Study, part of the National Health Service (NHS) England Newborn Genomes Program is sequencing and analyzing the genomes of 100,000 babies at birth to identify a specific set of rare genetic conditions that affect babies and to test whether genomics can speed up the diagnosis of about 200 rare genetic diseases and access to treatment.[11] The two-year study also evaluates the cost-effectiveness and parental acceptance of the program and works with the public, people with lived experience of rare diseases, researchers, and health professionals to explore the practical, ethical, and societal questions the study raises.[12]

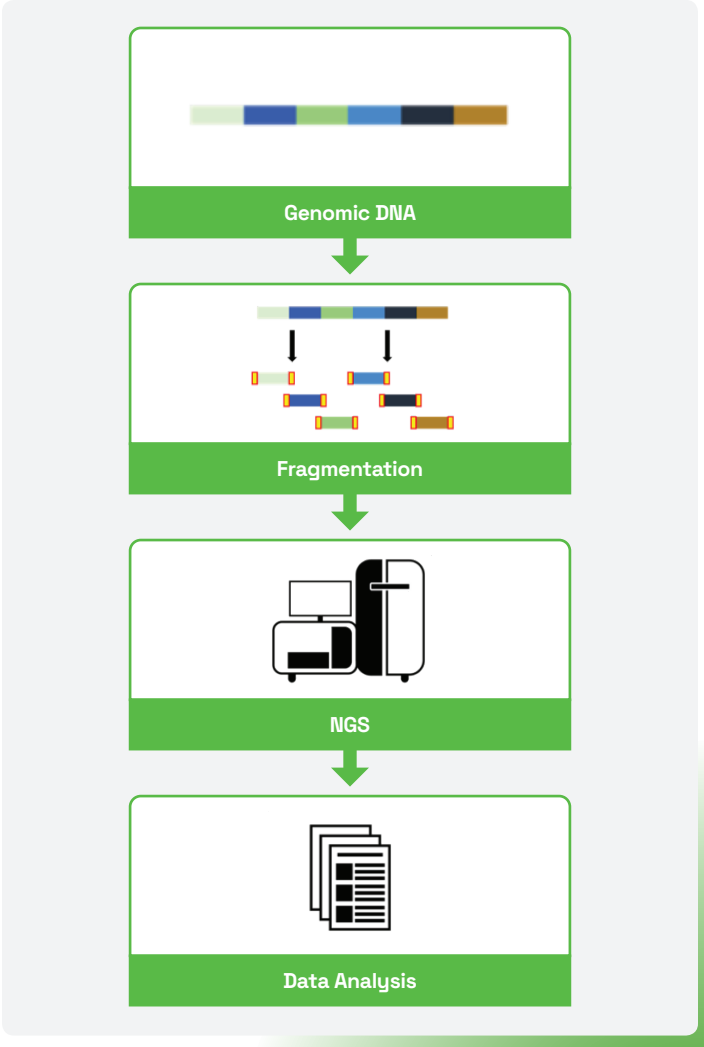

Figure 1: The whole genome sequencing process

Source: Bio Basic Asia Pacific

Sickle cell disease (SCD)

- Children with SCD have a significantly reduced life expectancy, often 20-30 years less than the healthy population.[13]
- Precision medicine, through advanced 'omics' research, is improving our understanding of SCD genomic architecture and identifying new biomarkers, while paving the way for tailored treatments.[14]
- For one group of patients, the gene-editing technique (CRISPR 2.0) is a potentially curative or highly disease-modifying option for abating the complications of the disease.[15]
- Understanding the different types of gene therapy in use, the differences in their endpoints, and their potential risks and benefits will be key to optimizing the long-term use of this therapy.[16]
- The US and the UK approved the world's first commercial gene-editing treatment for sickle cell disease in late 2023, 11 years after scientists first developed the DNA-snipping technology called CRISPR and building on 50 years of research on fetal hemoglobin.

See Insight Report No. 3 for more information on precision health, including the potential applications of innovations and technologies for the humanitarian and development sector.

Impact

- An accurate genetic diagnosis improves understanding of their prognosis and enables more personalized treatment and tailored management and surveillance.[17]
- A precise diagnosis also helps the child and their family to access better services such as patient and caregiver support groups, education, health, and other social care.
- In the coming years, therapies tailored to the genetic profiles of rare paediatric diseases are expected to become more effective, accessible, and affordable, driven by drug repurposing and innovation.[18]
- A global survey of 1,430 researchers on the future of genetic therapies for rare genetic diseases reveals that most expected gene therapies to be the standard of care for rare genetic diseases before 2036 and potentially offer cures thereafter.[19]
- CRISPR-Cas9 is highlighted as the gene editing technology most likely to effectively correct or replace faulty genes within the next 15 years.[20]
- Regarding the approved CRISPR treatment for sickle cell disease, it is too early to assume the treatment is permanent and without side effects. Ongoing monitoring is needed to fully understand its effectiveness. At this stage, the treatment costs over USD 2 million[21] and is out of reach for those who need it most.

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Insights Briefs

Innovation Nodes Insights Briefs serve as resource for practitioners and decision makers to quickly get up-to-speed on new and unknown areas of potential innovation for children.

Publication produced by the Innovation Nodes aim to facilitate the exchange of knowledge and stimulate discussion. The findings, interpretations and conclusions expressed are those of the researchers and authors, and do not necessarily reflect UNICEF policies or approaches.

For more information on Innovation Nodes and collaboration opportunities:

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Notes

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