THE SIGNAL TO NOISE RATIO:
A SAUDI MINISTRY OF HEALTH RARE DISEASES MOONSHOT PLEDGE

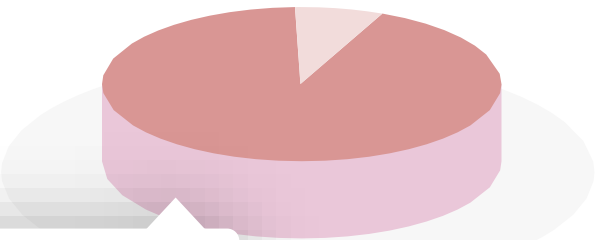
WHAT ARE RARE DISEASES?

Rare diseases (RDs), a heterogeneous group of health conditions, affect a small number of subjects compared to other common diseases in the general population, and represent a major public health issue at the global level. There are about 6,000 to 8,000 types of rare diseases.

About %4 of the total population

More than **300 Million** people worldwide

In Saudi Arabia, a disease is defined as rare when it affects less than **1** per **2000** people



80% of them are genetic

75% affect **children**

Die less than **5 years** **30%**

BURDEN OF RARE DISEASES

7.2 $ Trillion

8.6 $ Trillion

Economic burden for RDs across the world

The economic burden for rare diseases across the world could range from **7.2$ trillion** to **8.6$ trillion** per year. However, when there are no available treatments for these diseases, the costs will increase even more.

RARE DISEASES DIAGNOSIS

Accurate diagnosis can take about **10 - 15** years. This delayed diagnosis is due to the heterogeneity of symptoms of each disease, irregular presentation in the body and to the overlapping symptoms of many rare diseases.

The majority of misdiagnosed or undiagnosed patients suffering from a rare disease receives only supportive care.

SEVERAL UNMET NEEDS & CHALLENGES REMAIN ACROSS THE MANAGEMENT SPECTRUM OF RDs

Misdiagnosis or delayed diagnosis

For example, the hereditary transthyretin-mediated amyloidosis, or **hATTR, an exceedingly rare genetic disease**, affects approximately **50,000 people** worldwide with more than 130 different genetic variations. As a result of its **complex genetic footprint**, small patient base, the **average hATTR case takes 10 to 15 years to diagnose**.

Lack of Diagnostic tests and lab biomarkers

Traditional diagnostic techniques rely heavily on heuristic approaches. A large number of rare disease patients remain undiagnosed for years and many even die without an accurate diagnosis. In recent years, gene panels, microarrays, and exome sequencing have helped to identify the molecular cause of such rare and undiagnosed diseases. These technologies have allowed **diagnoses for a sizable proportion (25– 35%).** However, **a large proportion of these patients remain undiagnosed**.

Challenges Facing Physicians and Investigators

Because of **lack of awareness among healthcare professionals** many patients with rare diseases **do not receive appropriate treatment**. Challenges facing investigators include scarcity of funding and recruiting cohorts require multicentre collaborations.

Regulatory and Access to Breakthrough therapies

Countries in the MENA region usually have **low orphan drug prices due to ERP** (external reference pricing). The cost of high development, small patient pool, and **low prices discourage MNCs from launching orphan drugs in the MENA region.**

Lack of Treatment Options, and Clinical guidelines

Scarcity of experienced physicians and medical centres specialized in the treatment of rare diseases and **scarce local and regional clinical guidelines enabling accurate treatment pathway are considered a major limitation**.

Pharmaceutical companies are facing multiple limitations in rare disease research due to the high cost of drug research and development (R&D), the limited number of patients and the small orphan drug market.

RARE DISEASES TREATMENT

- An early diagnosis will improve a patient's chances of receiving treatment and enrolment in clinical trials.
- Effective treatments are lacking for most rare diseases, especially when diagnosis is delayed, mainly because of the inability to identify a molecular cause.
- Extensive research is being done, such as Next Generation Sequencing (NGS), which has made it possible to look for therapies for diseases that were once fatal. Finding a molecular cause can occasionally point to a therapy option.

WHY IS RAISING AWARENESS SO IMPORTANT

1

There are many hard working patient advocacy groups working at local and national levels with the goals of raising awareness, educating and supporting patients and families affected by rare diseases.

2

Their hard work can help bring solutions to people living with a rare disease.

ARITFICIAL INTELLIGENCE AND DIGITAL HEALTH SOLUTIONS

The ability of AI technologies to integrate and analyse data from various sources (e.g., multi-omics, patient registries, and so on) can be used to overcome challenges faced in RDs' (e.g., low diagnostic rates, reduced number of patients, geographical dispersion, and so on). Eventually, RDs' AI-mediated knowledge could significantly enhance therapy development by identifying disease biomarkers and increasing patient recruitment for clinical trials.

Digital Health Solution-Through collecting data via digital channels, pharmaceutical companies and HCPs can build a better understanding of the daily life for patients with rare diseases. They can develop a broad picture of symptoms, triggers, and identify patterns in patients with unrecognized conditions leading to innovations in treatment, as well as assisting individual patients with the management of their own health and wellbeing.

Digital health, including telehealth and other virtual care interactions/consults, reduces the previous challenging geographic restrictions, reducing the need to make long trips to visit a physician knowledgeable of a particular condition.

THE ROAD TOWARDS A SAUDI RARE DISEASES CENTER OF EXCELLENCE

The **Centre of Excellence** will combine and coordinate a wide range of services to provide multidisciplinary expertise and specialized care for patients suffering from a given rare disease; It will also act as a hub for knowledge research, treatment and services in a disease state.