In KSA, a disease is defined as rare when it affects less than 1 per 2000 people; Most rare diseases result from genetic disorders and occur at birth

Definition

~1 per 2000 people affected by a disease then it is defined as rare[1]

Any disease affecting fewer than 200,000 people in the US[2]

General Facts

~30%
of rare-disease patients die before their 5th birthday[1]

~75%
of rare disease of rare diseases could affect children[1]

~6K-8K
rare disease have **been** identified[1]

~80%
of rare diseases have identifiable genetic origins[2]

1: Saudi MoH 2: FDA Office of Orphan Products Development (OOPD)

More than 370K patients are affected with rare diseases in KSA which has the highest prevalence among MENA countries

Prevalence

Global

+350m Patients

More than 350 million patients are affected with rare diseases worldwide[1]

MENA

+3.3m Patients

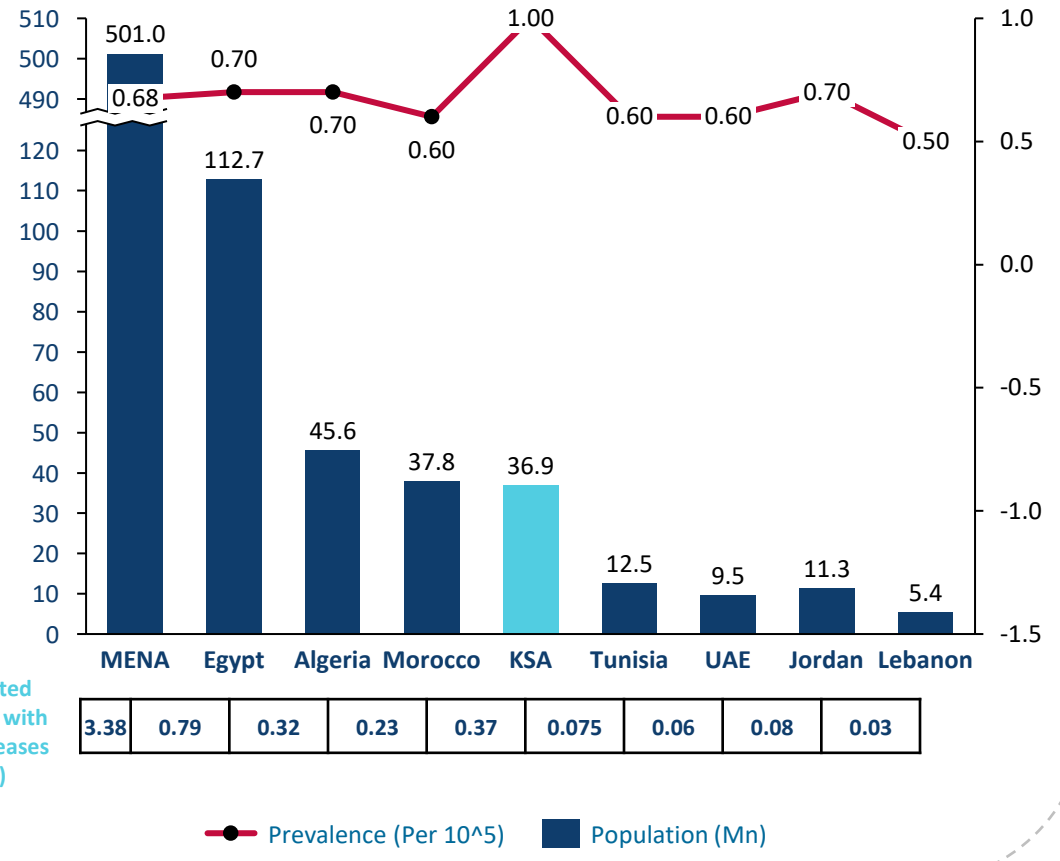
More than 3.3 million patients are affected with rare diseases in MENA region[2]

KSA

+370K Patients

More than 370,000 patients are affected with rare diseases in KSA[2]

Prevalence of rare disease in MENA region – 2023



Estimated Patients with Rare diseases (Mn)	MENA	Egypt	Algeria	Morocco	KSA	Tunisia	UAE	Jordan	Lebanon
	3.38	0.79	0.32	0.23	0.37	0.075	0.06	0.08	0.03

1:Global Genes, 2: IQVIA report on orphan disease in the Middle East and Africa region (2019) while considering 2023 population for all MENA counties in scope

Note: Prevalence is taken from IQVIA report on orphan disease in the Middle East and Africa region (2019) while considering 2023 population for all MENA counties in scope. Population numbers are extracted from Worldometer website

Rare diseases have general characteristics differentiating them from all other diseases

General Statistics

~95% of known rare diseases lack a treatment[1]

~250+ conditions are discovered every year[2]

~1200 rare diseases, have >5 documented cases published in the literature[3]

Social Impact

2/3 of family carers spend more than 2 hours a day on disease-related tasks[4]

3X higher is the proportion of people with rare diseases who report feeling depressed compared to the general population[4]

Foundation

~50%

of RD do not have a disease specific foundation supporting or researching their RD[5]

1: NORD, 2019, 2: EMA 2016 [RACATEUR 2019] 2008;371(9629):2039–41, 3: Nguengang Wakap, S., Lambert, D.M., Olry, A. et al. Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. Eur J Hum Genet 28, 165–173 (2020). 4: Juggling care and daily life: the balancing act of the rare disease community, 5:Global Genes,

~5 years is the average length of time from symptom onset to accurate diagnosis; SAR 6 Bn annually is the estimated treatment cost for RD

Diagnosis

4-6 years it takes for rare patients to receive an accurate diagnosis[1,2]

~3 misdiagnosis on average to diagnose rare diseases[1]

40% of rare disease patients are misdiagnosed at-least once[3]

7.3 physicians (avg.) is seen before initiation of correct diagnosis[4]

Economic Impact

~SAR 6 Bn annually
estimated lifetime treatment cost for RD in KSA[5]

~$517K
economic impact of a delayed diagnosis in avoidable costs per patient[6]

1: Marwaha S, Knowles JW, Ashley EA. A guide for the diagnosis of rare and undiagnosed disease: beyond the exome, 2:: Global Genes,M 3: Schieppati A, Henter JI, Daina E, Aperia A. Why rare diseases are an important medical and social issue. Lancet. 2008;371(9629):2039–41, 4: Engel PA., Bagal S., Broback M., Coice N. Physician and Patient Perceptions Regarding Physician Training in Rare Diseases, 5: MoH-KFMC study (not including cost of medical staff) , 6: Global Genes

In terms of policies, SFDA has published a guidance for orphan drug designation to identify eligible drugs and offer incentives

Eligibility Criteria

1. **One indication of orphan condition:**
   - ✓ Unregistered drug in SFDA
   - ✓ Registered drug in SFDA with a new indication, a new dosage form or other major variation applications
2. **Seriously debilitating diseases or life-threatening conditions or diseases**
3. **Prevalence of rare disease or condition or lack of financial viability**
4. **Under development for this orphan condition**
5. **Comparison with other methods for diagnosing, preventing, or treating the condition**

Incentives

- **Pre-submission meetings**
- **Scientific & Regulatory advice & consultation**
- **Priority Review**
- **Market Exclusivity**
- **Pricing**

In addition, MoH is establishing a Rare disease COE with the aim to improve the coordination and quality of care for patients

Rare disease Center of Excellence

Center of Excellence (CoE)

Brings together a multidisciplinary team to provide comprehensive care for patients with rare diseases while facilitating the development of networks and partnerships among stakeholders, including patients, families, healthcare providers, and policymakers, to promote collaboration and knowledge-sharing

Key pillars of COE

Clinical Research	• Attracting CTs to MOH hospitals as part of pharma value propositions • Upscaling MOH hospitals infrastructure & processes to accommodate clinical trials
HTA & economic evaluations	• Valuation of the EQ-5D-Y to generate local set of utilities for Children & adolescents • Putting MCDA into action • Working on a CE threshold for rare diseases
Digital Care Pathways	• Digital care pathways in rare diseases can help to ensure that patients receive timely and appropriate care by providing a standardized framework for diagnosis, treatment, and management. • These pathways may include tools such as decision support algorithms, remote monitoring technologies, and patient portals that allow for enhanced communication and collaboration between patients and healthcare providers. • By leveraging digital technologies, care pathways can also facilitate data collection and analysis, which may help to improve our understanding of rare diseases and inform the development of new treatments and therapies.
Disease registry	• A data repository for top RDs diagnosed at the MOH level with collaboration plans to include other key hospitals across the national level • Integrating remote management of patients & care givers through e-PROs, tele-consultation & remote monitoring devices
Capacity building	• Scaling up different selected RD sites to standardize and unify patient journey from diagnosis through treatment • Introduction of multiple multi-patient centric

Objectives

- ✓ Improving diagnosis and treatment options for patients with rare diseases
- ✓ Promoting research and innovation to advance knowledge and understanding of rare diseases
- ✓ Fostering collaboration and sharing of expertise among healthcare professionals
- ✓ Providing support and resources for patients, families, and caregivers affected by rare diseases
- ✓ Advocating for policies and initiatives that improve access to care and treatment for rare diseases on a broader scale

Among the major research initiatives, RDIA has recently led the development of the SGP 2.0; Identify and diagnose rare diseases is among key objectives

Strategic Objectives

الجينوم السعودي
SAUDI GENOME

- Reduce burden of preventable diseases
- Enable precision medicine
- Identify and diagnose rare diseases
- Integrate pharmacogenomics
- Build a Genomics database
- Foster Innovation and Collaboration

Potential Aspirations

- Position KSA as a leader in Genomics
- Improve public health landscape
- Boost IP co-development in collaboration with pharma companies
- Contribute to KSA GDP and jobs creation

On the other hand, various challenges are facing a rare disease patient journey in KSA and the region

Low number of patients

- Requiring National or regional datasets for patients' registration, identification and tracking

Lack of awareness

- Lead to significant diagnostic delays, misdiagnosis, and inadequate treatment options.
- Contributes to a sense of isolation and stigma among patients and families

Complex pathways

- Complex pathways, involving multiple genetic, environmental, and lifestyle factors, making them difficult to understand and diagnose

Regional variations in genetic testing

- Critical for effective diagnosis, treatment, and management of genetic diseases and can help inform targeted public health interventions.

Limited treatment options

- Significant challenge as these conditions are chronic, progressive, and life-threatening
- Small patient population, limited ROI, hence limited investment in R&DB for rare diseases