

Rare Diseases: Understanding the Unrecognized Challenge

A Comprehensive Guide for General Practitioners
and Pediatricians

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- 🎯 Objective: Raising awareness about rare diseases
- 👥 Target Audience: GPs and Pediatricians
- ! Key Message: Individually rare, collectively common



Defining Rare Diseases

A disease is considered **rare** when it affects a small portion of the population, though definitions vary by region.



United States: Fewer than 200,000 people



European Union: No more than 1 in 2,000 people

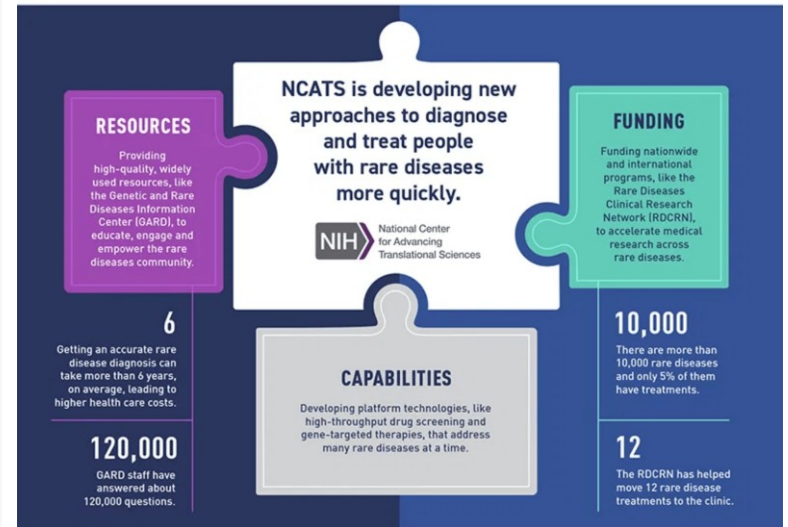


WHO: Fewer than 65 per 100,000 people

"Individually rare, but collectively common"

(Affecting 3.5% - 5.9% of global population)

RARE DISEASES: Individually Rare, Collectively Common



How Many Rare Diseases Exist?

7,000+

Identified Rare Diseases

80%

Genetic in Origin


70%


Manifest in Childhood

3%

Children Affected



 Despite individual rarity, collectively they affect a **significant portion** of the population.

 A large number of patients remain **undiagnosed** or misdiagnosed for years.

Why Are Rare Diseases Difficult to Diagnose?



Low Prevalence

Physicians have limited clinical exposure to these conditions.



Non-Specific Symptoms

Presentations often mimic common diseases, masking the rare cause.



Clinical Heterogeneity

The same disease can manifest differently in different patients.



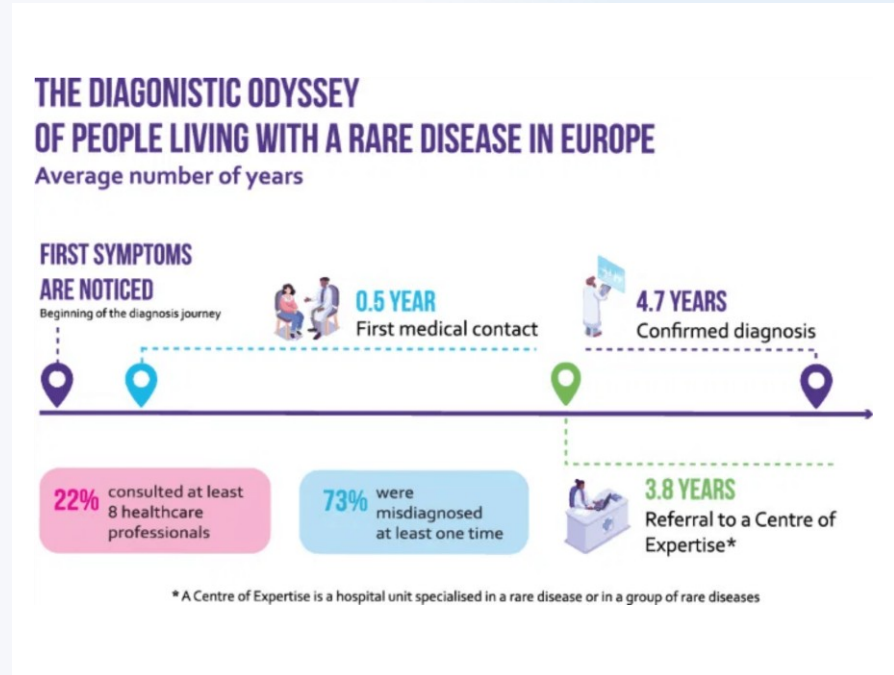
Lack of Criteria

Many rare diseases lack established diagnostic guidelines.



Diagnostic Delay

Patients visit an average of 7.3 physicians before diagnosis.



Consequences of Delayed Diagnosis

Patient Impact

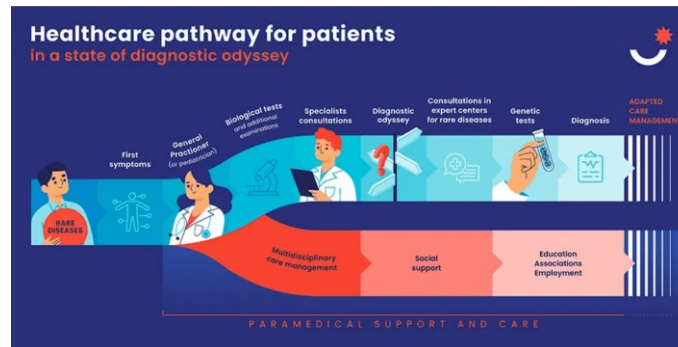
- Prolonged suffering and uncertainty
- Psychological burden on patients & families
- Unnecessary investigations & treatments
- Delayed access to appropriate care

Clinical Impact

- Disease progression during delay
- Increased morbidity and complications
- Worse long-term clinical outcomes
- Higher healthcare costs

Social Impact

- Delayed family planning decisions
- Educational and employment disruptions
- Social stigma and isolation
- Financial strain on families



Difficulties in Managing Rare Diseases



Limited Treatment Options

- Few approved therapies for most rare diseases
- Lack of clinical practice guidelines
- Limited evidence-based management protocols



Multidisciplinary Complexity

- Requires coordination across multiple specialties
- Complex medical and psychosocial needs
- Need for integrated care pathways



Specialist Scarcity

- Few experts available for consultation
- Geographic concentration of specialists
- Limited access to specialized care centers



Medication Challenges

- High cost of orphan drugs
- Limited availability and access
- Potential off-label use with unknown efficacy

Why Primary Care Physicians Matter



First Point of Contact

GPs and pediatricians are the gatekeepers, playing a critical role in early recognition and referral.



Significant Volume

Rare diseases account for approximately **1.6%** of outpatient visits—a substantial collective burden.



Recognition Challenges

Patients are often mislabeled with functional or psychiatric diagnoses due to non-specific symptoms.



Opportunity for Impact

Early suspicion and appropriate referral can drastically improve long-term patient outcomes.



How to Recognize Rare Diseases

! Clinical Red Flags

- 🚩 Unusual combination of symptoms not explained by common diseases
- 🚩 Multiple organ system involvement (e.g., neuro + dermatology)
- 🚩 Rapid disease progression or developmental regression
- 🚩 Failure to respond to standard evidence-based treatments
- 🚩 Family history of similar symptoms or unexplained deaths



Recognition Strategies

- 🔍 Maintain high index of suspicion
- 🕒 Take detailed family history
- 📄 Consult rare disease databases

Tools and Resources for Diagnosis



Collaborative Approach

- ✓ Multidisciplinary team consultation
- ✓ Specialist referral networks
- ✓ Rare disease centers of expertise
- ✓ Telemedicine consultations

Diagnostic Resources

 Orphanet

 OMIM Database

 GeneReviews

 Genetic Sequencing

Educational Resources

- Rare disease organizations (e.g., NORD, EURORDIS)
- Professional medical societies & guidelines
- Continuing Medical Education (CME) programs
- Clinical decision support tools

Rare Diseases in Children: Special Considerations



Pediatric Prevalence

- **~50-70%** of rare diseases manifest in childhood
- Significant burden on pediatric healthcare services
- High mortality rate in early childhood for severe cases



Diagnostic Challenges

- Overlapping symptoms with common developmental disorders
- Limited ability of young children to communicate symptoms
- Rapid disease progression requiring urgent diagnosis



Long-term Implications

- Need for lifelong management and multidisciplinary care
- Impact on physical growth, cognitive development, and education
- Complex transition from pediatric to adult care systems



Family-Centered Care

- Critical need for genetic counseling for family planning
- Psychosocial support for parents and siblings
- Connecting families with support groups and resources

What You Can Do



Increase Awareness

01

Educate yourself about rare diseases and their impact.

Stay updated with the latest diagnostic approaches.

Participate in continuing medical education (CME).



Improve Recognition

02

Maintain a high index of clinical suspicion.

Use diagnostic resources and decision support tools.

Consult specialists early when symptoms are unclear.



Support Patients

03

Provide compassionate care during the diagnostic journey.

Connect patients with support groups and resources.

Coordinate multidisciplinary care effectively.



Contribute to Knowledge

04

Participate in rare disease registries.

Report cases and outcomes to improve data.

Support and engage in research initiatives.

Rare Diseases: A Collective Challenge



Individually rare, but
Collectively Common



Majority are genetic
80% Genetic Origin



Average diagnostic delay
7.3 Physician Visits

The Critical Role of Primary Care

Early recognition, high index of suspicion, and appropriate referral can significantly alter the disease trajectory.

"Your patients may have unrecognized rare diseases. Early recognition saves time, suffering, and resources."