

DIAGNOSIS  
DOESN'T  
HAVE TO BE

**RARE**

# The Global Challenge of Rare Disease Diagnosis

The role of healthcare  
professionals in  
improving diagnosis



**Shire**

To be as brave as the people we help.

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# RARE DISEASES – THE DIAGNOSIS BURDEN



The often long and complex journey to an accurate diagnosis puts huge additional burden on both healthcare professionals and patients. The longer the diagnosis takes, the more difficult the process is for patients.<sup>2</sup>



An estimated **350 million people** globally – almost **5%** of the world's population<sup>1</sup> – are affected by rare diseases.



A delay in correct diagnosis may delay appropriate disease management, unnecessarily worsening the disease state,<sup>6</sup> and can also lead to additional interventions later deemed to be inappropriate given the underlying disorder.<sup>2</sup>

An accurate diagnosis is the first step to improving the care for those living with a rare disease and their families<sup>2,5</sup>



Of the circa **7,000** rare diseases identified to date,<sup>1,2</sup> **80%** are genetic,<sup>3</sup> more than **50%** affect children<sup>1</sup> and **30%** of patients die **before the age of five**.<sup>4</sup>



Misdiagnosis and non-diagnosis pose hurdles for thousands of patients with rare diseases and their families,<sup>2</sup> and may increase medical, economic and social burdens.<sup>2,5</sup>



The mean length of time from symptom onset to an accurate rare disease diagnosis is around **4.8 years**.<sup>2</sup>



Patients see an average of **7.3** physicians before a diagnosis is made.<sup>2</sup>

# THE DIAGNOSIS CHALLENGE FOR HEALTHCARE PROFESSIONALS



## Rarity

There is a limited ability for healthcare professionals to know all the signs and symptoms of rare diseases.<sup>5,7</sup>

Many diseases are so rare that there are physicians that will never see a single case during their whole career.<sup>2,8</sup>

The time it takes to reach a diagnosis depends upon the disease in question and the complexity of diagnostic needs.<sup>5</sup>

## Rare disease symptoms

Rare diseases are often misdiagnosed which can also lead to additional interventions later deemed to be inappropriate given the underlying disorder.<sup>6</sup>

Rare diseases may present with multiple symptoms, but physicians may not be familiar with the rare disease and this can lead to a lack of referral to the appropriate specialist.<sup>9</sup>

## Resources

Available information on rare diseases is inadequate. The appropriate training and awareness is not available to enable more effective diagnosis of these diseases.<sup>10</sup>

# WAYS TO IMPROVE DIAGNOSIS OF RARE DISEASES



**Key to early diagnosis is the specialist knowledge of rare diseases.<sup>8</sup>**

- ☁ Greater collaboration among physicians and access to specialists with expertise in rare diseases, may help to expedite the lengthy process to a correct diagnosis.<sup>8</sup>
- ☁ More awareness and support is needed for physicians to become experts on a specific rare disorder.<sup>8</sup>

**Linking potential symptoms together rather than treating just one individual symptom may help to reduce the time to diagnosis.<sup>8</sup>**



- ☁ Not underestimating the benefits of medical and family history information can help with identifying a rare disease and alleviate the current diagnostic delay.<sup>11</sup>

**In addition, specific diagnostic methods may be taken into account:**



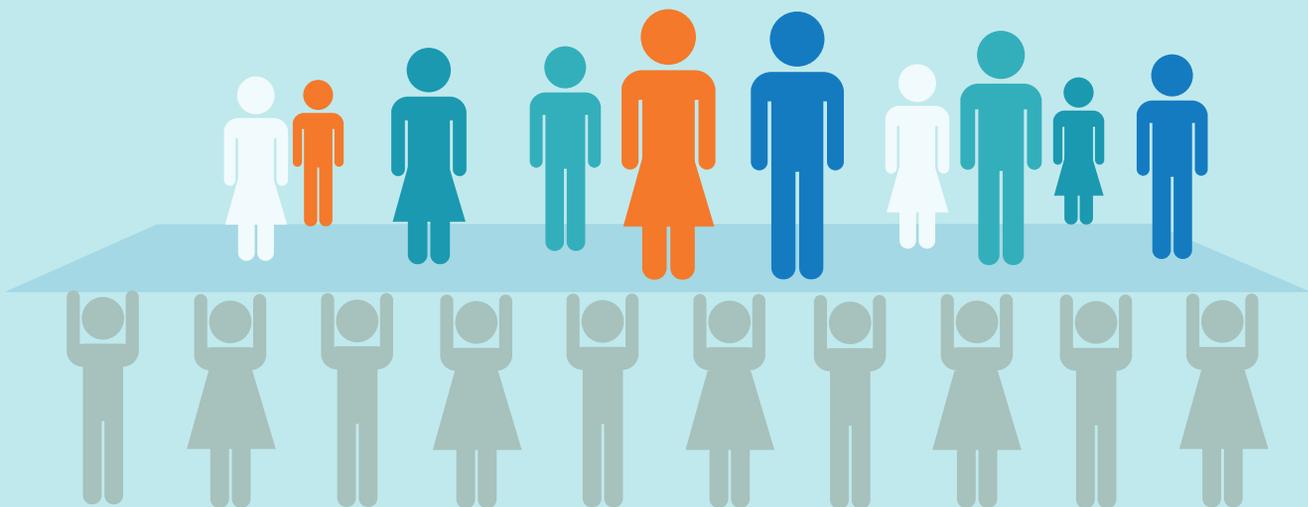
- ☁ For certain diseases, genetic testing can provide confirmation of disease.<sup>12</sup>
- ☁ Childhood screening may help to identify certain rare diseases early in life.<sup>8</sup>
- ☁ Thorough family history evaluation or pedigree analysis can ensure an earlier diagnosis and more appropriate care.<sup>11</sup>



# SHIRE'S COMMITMENT TO SUPPORTING THE RARE DISEASE COMMUNITY

In the framework of its 'Diagnosis Doesn't Have to Be Rare' initiative, Shire aims to support all stakeholders in improving the rare disease diagnosis journey. Our activities to improve rare disease diagnosis include:

- ▶ **Helping to raise awareness of rare diseases**, including the diagnosis challenges, and implementing initiatives to support an improved diagnosis journey.
- ▶ **Supporting local diagnostic testing** for rare diseases in certain countries and **providing education** for healthcare professionals on genetic testing.
- ▶ Supporting the efforts of the clinical community to **evaluate different screening methodologies** to support early detection of rare conditions.
- ▶ Supporting the **establishment of patient disease registries** to allow a better understanding of rare diseases.



## About Shire

At Shire, we enable people with life-altering conditions to lead better lives. We focus on developing and delivering innovative medicines for patients with rare diseases and other specialty conditions.

**For more information and updates on our  
'Diagnosis Doesn't Have To Be Rare' initiative visit:  
[www.shire.com/patients/improving-diagnosis](http://www.shire.com/patients/improving-diagnosis)**

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