

The Global Challenge of Rare Disease Diagnosis

The benefits of an improved diagnosis
journey for patients



To be as brave as the people we help.

www.Shire.com/RareDiagnosis

LIVING WITH A RARE DISEASE: WHY IS EARLY DIAGNOSIS ESSENTIAL?



About **350m people globally** are affected by rare diseases, many of them life-threatening or chronically debilitating.



Improved understanding of rare diseases is key for accurate and timely diagnosis.



Of the **7,000** rare diseases; **80% are genetic.**



More than **50%** of rare diseases affect children.



30% of patients die before the age of five.

The diagnosis burden



The length of time from symptom onset to an accurate diagnosis is around **4.8 years for a rare disease.**

The **longer** it takes to diagnose a rare disease, **the more physicians** the patient needs to see.

Patients see an average of **7.3 physicians** before a diagnosis is made.

WHY CAN IT TAKE SO LONG TO DIAGNOSE A RARE DISEASE?

Diagnosis challenge

Patients, families and physicians have **limited awareness**.

Symptoms are often hidden behind more common illnesses and initially, may appear to be of only minor concern.

Rare disease symptoms may not always be evident to doctors or nurses who have never encountered the disease.

Impact to patients

Delays in diagnosis can lead to **inappropriate management** as well as **disease progression**.

Misdiagnosis can also lead to additional interventions later deemed to be inappropriate given the underlying disorder.

Patients and families may feel frustrated and scared when diagnosis is delayed

“ Before I was diagnosed, we knew nothing about it as a family. I was having lots of symptoms and I did not know what was wrong with me.* ”

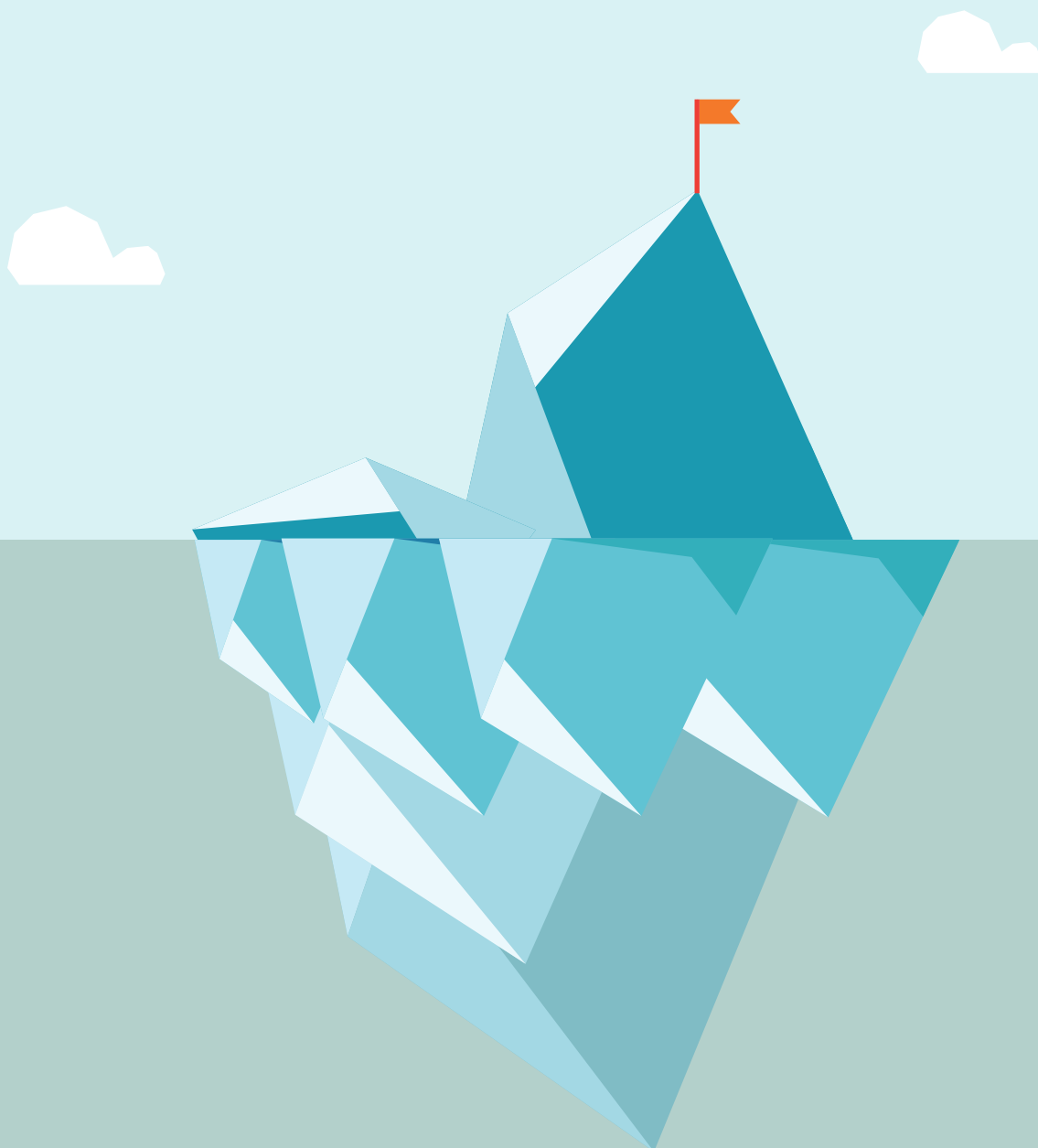
“ We kept going from doctor to doctor and nobody could tell me what was wrong.* ”

“ Getting a correct diagnosis was so difficult, stressful and humiliating at times. There was a lot of ‘passing’ me from doctor to doctor without an overall co-ordinator of care.* ”

*Patient testimonials

HOW CAN DIAGNOSIS BE IMPROVED?

- ☁ An **accurate and timely** diagnosis is the first step to improving care for people with rare diseases and their families.
- ☁ **Knowledge and recognition of rare diseases** are important to empower patients and their carers.
- ☁ Healthcare professionals may suggest the following measures:
 - ☁ **Genetic testing** to confirm disease presence.
 - ☁ **Family history and genetic testing** to ensure an earlier diagnosis.
 - ☁ **Childhood screening** to help identify certain rare diseases early in life.



SHIRE'S COMMITMENT TO SUPPORTING RARE DISEASE DIAGNOSIS

Shire believes that the pathway to improved diagnosis, and in turn to improved care and outcome, can be helped by the rare disease community **working together to find solutions** to help **ensure accurate diagnosis** at an early age.

Shire's '**Diagnosis Doesn't Have to Be Rare**' is:

- ▶ **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



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