The Global Challenge of Rare Disease Diagnosis
A Policy Briefing

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Governments are well-positioned to facilitate progress. There is a recognized need for collaboration among countries in order to improve the detection, diagnosis and treatment of rare diseases. It is crucial to support the further research that is needed to ensure accurate diagnosis of rare diseases at an early stage.

We encourage healthcare institutions to develop criteria on the rare conditions to be screened for at birth, and for governments to harmonize the processes by which new conditions are added to local screening panels. There should be a maintained focus on rare diseases as a health policy priority, and countries must continue adopting National Plans for rare diseases.

Shire is leading the ‘Diagnosis Doesn’t Have to be Rare’ initiative which aims to highlight the challenging diagnosis journey some patients with rare diseases often experience, and calls for improvement in the diagnosis pathway.

Rare diseases often hide behind common symptoms of other more common illnesses, making the diagnosis extremely challenging and often leading to misdiagnosis. Despite progress made in this space, there remains a need to better understand the obstacles patients and caregivers within the rare disease community face in obtaining a correct and timely diagnosis.

The pathway to improve diagnosis can only be helped by all stakeholders joining together to find solutions to help ensure accurate diagnosis of rare diseases at an early stage. An accurate diagnosis may be the first step to improving the care for those living with a rare disease, and their families.

A rare disease is a life-threatening or chronically debilitating condition. It is defined as rare in Europe when fewer than 5 in 10,000 people are affected, and in the US when fewer than 200,000 people are affected.

Almost 5% of the world’s population are living with a rare disease.

Approximately 75% of rare diseases affect children and 30% of rare disease patients die before the age of 5.

There are approximately 7,000 rare diseases identified.

80% of rare diseases are genetic and some can be life-threatening.
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The longer it takes to diagnose a rare disease, the more physicians the patient needs to see.

The mean average length of time from symptom onset to accurate diagnosis is approximately 4.8 years. 40% of rare disease patients are misdiagnosed at least once.

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People with a rare disease may experience low quality of life and high levels of disability.

Misdiagnosis and non-diagnosis can pose hurdles to quality of life for thousands of rare disease patients.

This difficult journey to diagnosis and care can increase medical, economic and social burdens.

Burden of rare diseases

**Barriers to diagnosis**

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The misdiagnosis burden
Improving Diagnosis of Rare Diseases – A Call to Action

Diagnosis Doesn’t Have to be Rare

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- It is crucial to support the further research that is needed to ensure accurate diagnosis of rare diseases at an early stage.
- We encourage healthcare institutions to develop criteria on the rare conditions to be screened for at birth, and for governments to harmonize the processes by which new conditions are added to local screening panels.
- There should be a maintained focus on rare diseases as a health policy priority, and countries must continue adopting National Plans for rare diseases.

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Shire aims to partner with the rare disease community throughout 2015 to introduce new initiatives under the “Diagnosis Doesn’t Have to be Rare” campaign umbrella directly aimed at improving the diagnosis journey.

We are committing to helping raise awareness of rare diseases, including the diagnosis challenges, and to implementing initiatives to support an improved diagnosis journey.

We support the efforts of the clinical community in certain countries to evaluate different screening methodologies to support earlier detection of these devastating conditions.

We support local diagnostic testing for rare diseases in certain countries, and offer education for healthcare professionals on genetic testing.

In certain countries we are supporting the establishment of patient disease registries to allow a greater understanding of rare diseases.

What Shire is committing to
About us

Shire puts patients at the heart of the business and is driven by a common goal: to enable people with life-altering conditions to lead better lives. As a leader in the development and marketing of orphan drugs for rare diseases, Shire delivers the science that offers hope to those with rare conditions. We work in partnership with physicians, patients, caregivers, payers and policymakers worldwide to ensure that patients have access to our innovative therapies and the support they need to lead better lives.

For more information and updates on our diagnosis campaign initiatives visit: www.Shire.com/RareDiagnosis

References

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