

Information on Drug Access in the Brazilian and Latin American Market.

## The reality of the journey of those living with rare diseases in Brazil.

In February, more precisely on the 28th, World Rare Disease Day is celebrated. The event was created in 2008 by the European Organization for Rare Diseases (Eurordis) to raise awareness of governments, health professionals, and the population about the existence and care of such diseases. The purpose is to spread knowledge, seek patient support, and encourage research to improve treatment.

When it comes to a rare disease, access to diagnosis is a challenge faced in Brazil and worldwide. Many patients end up in the Brazilian Universal Healthcare Program (SUS) service network for years, consulting different specialists, receiving wrong diagnoses, and undergoing random treatments, before obtaining the correct diagnosis.

According to the World Health Organization, rare diseases characterizes by a broad diversity of signs and symptoms, which vary from disease to disease and from person to person, even if affected by the same condition. Most degenerative, chronic, progressive, and disabling diseases still have no cure and can even lead to death.

Diagnosing rare diseases is challenging; according to studies\*, it can take around seven years to diagnose a rare disease, and 7 to 8 medical visits and up to 3 different diagnoses requires in this period. This scenario has a negative impact on patients, their families, and the health system.

Brazil adopted the "Brazilian Policy of Comprehensive Care for People with Rare Diseases", created by Ordinance No. 199/2014, which since 2014, has been implemented to organize the health care network for prevention, diagnosis, treatment, and rehabilitation. The Brazilian Universal Healthcare Program (SUS) provides care for the prevention, diagnosis, treatment, and rehabilitation of people with rare diseases, in addition to treating symptoms.

According to an article on the Brazilian Ministry of Health website, the history of rare diseases relates to creation of the newborn screening test, also known as Teste do Pezinho (Heel Prick Test or Guthrie Test), adopted by the SUS in 1992. The test was created in the late 1950s by research biologist Doctor Robert Guthrie, who was studying ways to test infants for phenylketonuria (PKU) before the onset of symptoms to prevent the development of intellectual disability (ID) caused by the disease.

**A significant development occurred in 2021 when the Federal Government increased 50 the number of diseases that can be detected by the Heel Prick Test offered by SUS. More than 80% of newborns in Brazil undergo the Heel Prick Test. The Brazilian Universal Healthcare Program (SUS) performs 2.4 million annual tests in over 28,000 locations, including maternity hospitals and Primary Healthcare Units.**

Thinking of other ways to expand access to the diagnosis of rare diseases, the group of companies Multicare and Intriads made donations to help build the Center for Comprehensive Care and Training in Rare Diseases, which will be inaugurated on February 28, in Porto Alegre, by Instituto Genética para Todos and Casa Hunter, civil associations that develop projects relating to rare genetic diseases. Casa dos Raros is an unprecedented initiative in Latin America, created from the need to expand access to rapid and accurate diagnosis, ensure treatment and promote research.

The project is audacious, with specialized assessments and guidance for diagnosis and treatment with the most advanced technology, relying on the expertise of a highly qualified multidisciplinary team, which will also connect with professionals and patients in remote locations through telehealth.

**Multicare advises its commercial partners in preparing their access and price plans, sharing the knowledge accumulated over its 38 years of history, maximizing opportunities, and ensuring that the best price is obtained.**

**Talk to our Commercial and Access teams and ensure the best strategy for your company in the Brazilian market.**



### SOURCES

\*<https://www.roche.com.br/pt/por-dentro-da-roche/as-doencas-raras-no-brasil.html#:~:text=O%20diagn%C3%B3stico%20de%20doen%C3%A7as%20raras,e%20o%20sistema%20de%20sa%C3%BAde>

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