# Rare diseases

There are an estimated 6,000–8,000 rare diseases in the world today affecting more than 300 million people(1). Because only around 500 of these diseases have approved treatments, this is a hugely under-served area with great unmet medical needs. At Sobi, we specialise in rare diseases because it is where we can make the greatest difference to people’s lives.

The rare disease space is unlike any other in medicine. In Europe, a rare disease is defined as one affecting fewer than one person per 2,000. In the US, the Orphan Drug Act of 1983 defines a rare disease as a condition affecting fewer than 200,000 people. Because physicians may never have seen the condition before, many cases can go undiagnosed for years. Around 75 per cent(2) of identified rare diseases affect children, and many have a devastating effect on life expectancy and quality of life. An estimated 35 per cent of children with a rare disease will not live to see their fifth birthday, and rare diseases are thought to be responsible for around 35 per cent of deaths during the first year of life. Around 80 per cent of rare diseases are inherited rather than acquired: they involve a defect in the genes that tell our bodies how to work. As a result, the body may fail to produce an essential enzyme or protein, for example, or its own immune defences may attack its own systems. Because they are most commonly genetic disorders, rare diseases tend to be lifelong. Treatment is often focused on resolving the problem caused by the defective gene, alleviating the symptoms and allowing the person to live a more normal life.

(1) Global genes, www.globalgenes.com
(2) Eurordis, www.eurordis.org