A rare disease is a life-threatening, seriously debilitating, or serious and chronic condition affecting a relatively small number of patients – in the EU this is defined as less than five persons affected in 10,000.

Orphan drugs are intended for the diagnosis, prevention or treatment of these rare diseases, of which there are more than 6,000 classified as rare, most of them of genetic origin and often chronic and life-threatening. In the EU (as well as other regions) there is a range of incentives to encourage the development of orphan medicines. The interests of the pharma industry in developing and marketing medicinal products for rare diseases has previously been small, especially under normal market conditions. Since the introduction of the so-called Orphan Regulation (EC 141/2000) in Europe, with the range of incentives offered, the number of orphan drugs is increasing in these markets. Last year (2014) saw the highest number of orphan designated medicines by the EMA for marketing authorisation in a year. Of the 82 medicines recommended in 2014, 17 are intended for the treatment of a rare disease. The past year also saw the first recommendation worldwide of a therapy based on stem cells, Holoclar, for the treatment of limbal stem cell deficiency, a rare eye condition that can result in blindness.

The collecting of data to demonstrate safety and efficacy of an orphan drug – and proving that benefits outweigh the risks of the medicine – is challenging for applicants and regulators alike