

Rare disease

A rare disease, also referred to as an orphan disease, is any disease that affects a small percentage of the population. Rare diseases are commonly defined as life-threatening or chronically debilitating diseases which are of such low prevalence (fewer than 1 in 2,000 people) that special combined efforts are needed to address them. Diseases that are statistically rare, but not also life-threatening, chronically debilitating, or inadequately treated, are excluded from this definition. A disease may be considered rare in one part of the world, or in a particular group of people, but still be common in another.

Most rare diseases are genetic so that most people show symptoms from childhood (although some rare diseases only become apparent later in life).