

Orphanet classification of rare diseases



General methodology

Orphanet has created a clinical classification system for rare diseases present in its database. This system consists of classifications organised according to the medical and/or surgical speciality that manages specific aspects of each rare disease within a healthcare system. When relevant, a distinction is made between childhood and adult disorders. The diseases have been classified within each speciality according to clinical criteria or etiological criteria when diagnostically or therapeutically relevant.

The Orphanet classification system employs two non-exclusive sources:

Documented sources: reference texts, scientific literature

Expert opinions from working groups focusing on specific diseases

The Orphanet classification system is an evolutionary process:

It is updated monthly to incorporate findings from the scientific literature

Revisions by entire section are regularly effectuated using the same methodology (newly documented sources, expert gatherings)