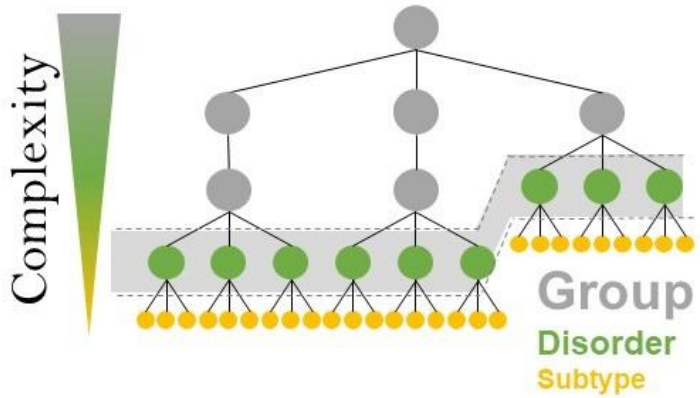
What is the “Aggregation level”?

As the Orphanet nomenclature of Rare Diseases is organized in a multi-hierarchical classification system, a reduced list of ORPHcodes is recommended to be used either in coding setting if the Orphanet classifications are not used, or to allow data sharing and statistical analysis at EU-level if the Orphanet classifications are used in coding setting. This reduced list constitutes the « Aggregation level » and is made by the ORPHAcodes of disorder level, excluding groups and subtypes of the Orphanet classification. The Aggregation level of disorders and subtypes is provided in the Orphanet nomenclature file.



- **What is the Aggregation level of a disorder?**

The Aggregation level of a disorder is itself. It is unique.

Example: **ORPHA:355** Gaucher disease (Disorder) ↳ **ORPHA:355** Gaucher disease (Disorder).

- **What is the Aggregation level of a subtype?**

A subtype is associated to only one disorder. The Aggregation level of a subtype is this disorder. It is unique.

Example: **ORPHA:77259** Gaucher disease type 1 (Subtype) ↳ **ORPHA:355** Gaucher disease (Disorder).

- **What is the Aggregation level of a group?**

A group may encompasse other groups, disorders and subtypes. The Aggregation level of a group is multiple and includes all the disorders included in this group, even if there are not direct descendants. Because it is not relevant for coding and data sharing and to avoid to weigh the Orphanet nomenclature file down uselessly, this list is not provided in the xml files.

- **Exceptional cases: Inactive ORPHAnumbers**

Whenever possible, an inactive ORPHAnumber has an ORPHAnumber of replacement in the Orphanet nomenclature in use. The Aggregation level of the inactive ORPHAnumber is the Aggregation level of the ORPHAnumber of replacement.

Examples:

ORPHA:98836 Bilineal acute leukemia (Disorder) deprecated towards **ORPHA:530995** Mixed phenotype acute leukemia (Disorder)

↳ **ORPHA:530995** Mixed phenotype acute leukemia (Disorder)

ORPHA:157855 HARP syndrome (Subtype) deprecated towards **ORPHA:216866** Classic pantothenate kinase-associated neurodegeneration (Subtype)

↳ **ORPHA:157850** Pantothenate kinase-associated neurodegeneration (Disorder)