**Protocole curation Freeze 3 :**

* FREEZE n°1

➢ **DATE** : 13 septembre 2022  
➢ **PHENOPACKETS**

■ Parents excluded

■ 6243 with at least 5 HPO

■ 439 solved cases with genes with at least 5 HPO

■File of solved cases: *Solved\_with\_gene\_noparents\_with\_5phenotypes\_aftercuration.xlsx*

* **ALGORITHME** :

■ Resnik  
■ Top 50 phenotypic association

■ product 4 (*product4\_RunSolveRD.xml*) 20 September 2022

* 4210 clinical entities with HPO)
* 3907 ORPHA clinical entities (disorders + subtypes) with at least 1 gene (*en\_product6\_sep2022.xml*) 16 September 2022

■ Groups of disorders excluded

* **FILTRATION** :

■ HPO

● HPO >= 5 cases related to ERN

● HPO > 2 ORPHAcodes

● HPO negated excluded

■ No scores filtration

■ No genes filtration

■ ORPHAcodes

● Groups of disorders excluded (2489)

● ORPHAcodes childs *: product\_ORPHApackets\_childs.xml* (2542) of 21 September 2022

Repartition in nomenclature ORPHA:code  
Group of disorder 2489   
Disorder 6937   
subtype of disorder 1250