

Review of New Disease Terms:

1- look up each term in DO

- Is there a DO term, matching on Disease Name ?
 - NOTE: the ID range, this is a term defined early in the DO

- look at definition
 - Does the definition follow our current format

- review source of definition
 - NOTE : is there a more recent paper ?
 - add new PubMed xrefs to the DO definition

- Does the new term match a synonym of a DO term ?

- look at xrefs
 - Check OMIM, Orphanet, Genetics Home Reference
 - What new information can you gather from Orphanet, GARD, Genetics Home Reference ?
 - Genetics Home Reference:
 - check the Other Names, to determine if a previous name was used for a disease name or synonym in the DO

- > what is the supporting evidence for this DO term
 - Does it need to be revised ?
 - Are there multiple OMIMs
 - > look to determine if a Phenotypic series has been added to OMIM, update the DO

2 - New terms:

- Check spelling variations
- Is this term a synonym of a DO term, an Obsolete term?

- Review published/authoritative definitions for these terms.
 - NOTE : is there a more recent paper ?
 - add new PubMed xrefs to the DO definition

NOTE: → Determine the usage, synonyms of these terms
→ search PubMed, PubMed Central, then Google

→ look up the cited papers: note the year of publication, the title of the publication

To write a definition:

→ Go to the authoritative sites, OMIM, GHR, Orphanet, etc,

-- Identify pertinent phrases from these sites, record the text, to devise a DO definition