**Disease Selection based on Genomics England rare disease recruitment**

Diseases removed *before screening*:

Duplicate records removed (n = 0)

Diseases identified from GEL normalised, specific recruitment disease:

(n = 221)

**Identification**

Diseases screened for sufficient case numbers

(n = 221)

Diseases excluded with case number < 10

(n = 53)

Diseases mapped to OMIM disease concepts

(n = 168)

Diseases excluded due to missing OMIM mappings

(n = 40)

**Filtering**

Diseases excluded due to imperfect mappings

(n = 37)

Disease mappings adjudicated by clinical specialist

(n = 128)

Disease mappings screened for sufficient PheCode mappings

(n = 91)

Diseases excluded due to mapped PheCodes < 3 or less than half of HPO codes mapped to PheCodes

(n = 10)

Diseases included in study

(n = 81)

**Included**

*Adapted From:*  Page MJ, McKenzie JE, Bossuyt PM, Boutron I, Hoffmann TC, Mulrow CD, et al. The PRISMA 2020 statement: an updated guideline for reporting systematic reviews. BMJ 2021;372:n71. doi: 10.1136/bmj.n71

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