

CASE DEFINITION FOR POMPE DISEASE (PD)

Report any patient (**new or previously diagnosed**) less than 18 years of age meeting the following criteria:

1. **Genetic criteria:** Pathogenic mutations affecting both *GAA* genes (encodes the acid alpha-glucosidase protein) as determined by sequence analysis or deletion/duplication analysis

AND/OR

2. **Biochemical criteria:** Measurement of acid alpha-glucosidase (*GAA*) enzyme activity performed on one or more of:
 - Dried blood spot *GAA* enzyme activity assay
 - Whole blood *GAA* enzyme activity assay
 - Skin biopsy (fibroblast culture) *GAA* enzyme activity assay
 - Muscle biopsy *GAA* enzyme activity assay

Exclusion criteria

Clinical evidence of proximal muscle weakness without genetic or biochemical confirmation of disease