Provider action sheet for Pompe disease

If the California Newborn Screening (NBS) Program has identified an infant in your care who has a screen-positive result for Pompe disease:

What steps do I need to take?

1. **Consult** immediately with a California Children's Services Metabolic Special Care Center (SCC) specialist. They will ask for your assessment of the infant's current health status and if the infant has signs of the disorder. Further evaluation and confirmatory tests will be arranged by the SCC.
2. **Contact** the family to explain the positive newborn screening test result. For information on how best to communicate, consult the **Health Resources & Services Administration Newborn Communication Guide** (https://bit.ly/HRSAGuide or search online for "HRSA" "heritable" "communication")
3. **Review** the accompanying, "Family action sheet for Pompe disease", with the family and ensure they understand.
4. **Inform** the family that this result requires more testing and evaluation. Emphasize that you, with NBS Program Area Service Center (ASC) staff and the SCC specialist, will guide them through the next steps of confirmatory testing and follow-up services.
5. **Advise** parents to (1) follow the plan for confirmatory testing right away, (2) keep their infant's appointments with the SCC specialist, and (3) start treatment if indicated. The family should receive services from a team of specialists, including genetic counseling services.

Clinical information

Pompe disease is an autosomal recessive disorder caused by a deficiency of the lysosomal enzyme acid-α-glucosidase (GAA), resulting in accumulation of lysosomal glycogen, primarily in cardiac and skeletal muscles. The clinical presentation is divided primarily into two forms determined by severity of the enzyme deficiency:

1. Infantile Onset Pompe Disease (IOPD) presents primarily with symptoms of generalized hypotonia, failure to thrive, and hypertrophic cardiomyopathy. Without treatment by enzyme replacement therapy (ERT), IOPD is commonly fatal.

2. Late-Onset Pompe Disease (LOPD) presents primarily as proximal muscle weakness and respiratory insufficiency. Cardiac involvement is uncommon. ERT may stabilize respiratory and motor symptoms.

In some cases, a pseudo-deficiency may cause decreased enzyme activity without disease.

Early and lifelong enzyme replacement therapy (ERT) is associated with improved quality of life and increased survival.

Please visit these sites. Search for the site name and "Pompe" if needed.

- **HRSA** (https://newbornscreening.hrsa.gov/conditions/pompe-disease)
- **GeneReviews** https://www.ncbi.nlm.nih.gov/books/NBK1261/

Questions?

For follow-up questions, please call your NBS Program Area Service Center.

For program questions, please email NBS Program staff at NBS@cdph.ca.gov or visit the NBS Program website (www.cdph.ca.gov/NBS).

California Department of Public Health
Newborn Screening Program (www.cdph.ca.gov/NBS)