Provider action sheet for mucopolysaccharidosis type I (MPS I)

If the California Newborn Screening (NBS) Program has identified an infant in your care who has a screen-positive result for mucopolysaccharidosis type I (MPS I).

What steps do I need to take?

1. **Consult** 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 any 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 mucopolysaccharidosis type I (MPS I) 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 multidisciplinary team of specialists, including genetic counseling services.

Clinical information

MPS I is an autosomal recessive disorder caused by pathogenic changes in the Alpha-L-Iduronidase (*Idua*) gene, causing accumulation of glycosaminoglycans (GAGs) in cells. This leads to progressive multisystem organ damage.

MPS I varies in severity, symptoms, and age of onset. The more severe form of MPS I may have coarsening facial features, skeletal changes, respiratory issues, and developmental delays that may present within the first 1-2 years of life. Early progressive MPS I (Hurler) is most severe, while slowly progressive (Hurler–Scheie, Scheie, or attenuated MPS I) is less severe and may present at an older age. In some cases, a pseudo-deficiency may cause decreased enzyme activity without disease.

Early and ongoing treatment can prevent or delay symptoms of the condition. Treatments include enzyme replacement therapy (ERT) and/or hematopoietic stem cell transplant (HSCT). Gene therapy and improved enzyme replacement therapy are in clinical trials.

Please visit these sites. Search for the site name and "MPS I".

- **HRSA Newborn Screening** (https://newbornscreening.hrsa.gov/conditions/mucopolysaccharidosis-type-i)
- **Gene Review** (https://www.ncbi.nlm.nih.gov/books/NBK1162)

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)