Provider action sheet for mucopolysaccharidosis type II (MPS II or Hunter syndrome)

If the California Newborn Screening (NBS) Program has identified an infant in your care who has a screen-positive result for mucopolysaccharidosis type II (MPS II or Hunter syndrome):

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 II (MPS II or Hunter syndrome)," 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 multi-disciplinary team of specialists, including genetic counseling services.

Clinical information

MPS II is an X-linked disorder caused by pathogenic variants in the iduronate 2-sulfastase (*IDS*) gene that results in accumulation of glycosaminoglycans (*GAGs*) in cells. This leads to progressive multisystem organ damage.

MPS II clinical presentation is determined by age of onset and primarily affects boys. Severe form of MPS II may show cognitive impairment, coarsening facial features, skeletal changes, cardiac and respiratory issues, that present by 2-4 years of life. Neuronopathic MPS II is most severe while non-neuronopathic MPS II (attenuated), may have milder and later onset symptoms. In some cases, a pseudo-deficiency may cause decreased enzyme activity without disease.

Early and ongoing treatment may slow progression of symptoms and help quality of life. Treatments include enzyme replacement therapy and rarely hematopoietic stem cell transplant. Gene therapy and improved enzyme replacement therapy are in clinical trials.

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

- **HRSA Newborn Screening** (https://newbornscreening.hrsa.gov/conditions/mucopolysaccharidosis-type-ii)
- **GeneReviews** (https://www.ncbi.nlm.nih.gov/books/NBK1274/)

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)
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