NORTH CAROLINA NEWBORN SCREENING PROGRAM

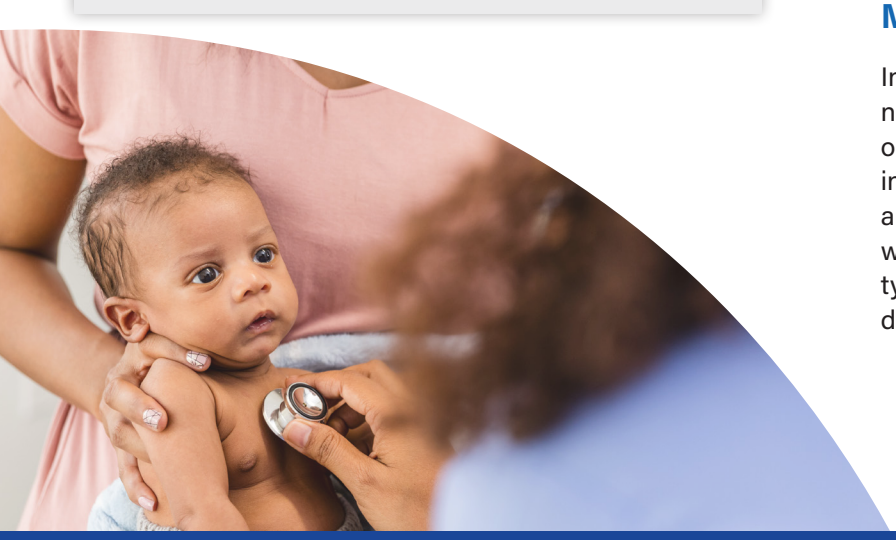
Mucopolysaccharidosis II (MPS II) Fact Sheet for Providers

What is MPS II?

Mucopolysaccharidosis II (MPS II), also known as Hunter syndrome, belongs to a group of inherited lysosomal storage disorders known as mucopolysaccharidoses (MPS). MPS II is caused by the absence or deficiency of a specific enzyme, iduronate-2-sulfatase, required to break down glycosaminoglycans (GAGs). Because of this deficiency, GAGs are stored in cells throughout the body, resulting in progressive cellular damage with multisystem involvement.

How do I handle an abnormal NBS for MPS II?

Newborn screening (NBS) for MPS II uses a two-tier approach measuring iduronate-2-sulfatase enzyme activity first, followed by MPS II marker (GAG) analysis if the enzyme level is below the cutoff. A positive NBS is reported when an infant has BOTH deficient enzyme activity and an abnormal MPS II marker value. If you are notified that one of your patients received a positive MPS II NBS result, immediately contact the Muenzer MPS Center with UNC Pediatric Genetics and Metabolism (919-228-2432) for next steps and prompt referral. Infants with a positive NBS for MPS II require clinical evaluation and confirmatory testing, as early diagnosis is critical for timely treatment initiation to optimize long-term outcomes.



What are the signs and symptoms of MPS II?

Infants with MPS II typically show no symptoms at birth, but they can develop symptoms starting at 1 to 2 years of age. MPS II impacts multiple systems including neurological, respiratory, cardiac, musculoskeletal, auditory, and ophthalmologic functions. MPS II has a continuous spectrum of disease severity ranging from severe to attenuated forms.

MPS II severe form

Without treatment, infants with the severe form of MPS II typically show developmental delays by the end of the second year of life, with a plateau during the third to fifth years of life followed by a progressive cognitive decline. Language development may be very limited because of hearing loss and recurrent ear infections. Physical features can include hernias, skeletal abnormalities, distinctive facial features (coarse facies, prominent forehead, broad upturned nose), enlarged tongue, chronic rhinitis, cardiac valve disease, joint stiffness, and hepatosplenomegaly. Without treatment, children with severe MPS II typically die in their teenage years due to progressive neurological involvement and complications from obstructive upper airway disease and cardiac disease.

MPS II attenuated form

Individuals with attenuated MPS II typically have normal intelligence. Physical features may include obstructive upper airway disease, musculoskeletal involvement with decreased joint range of motion, and valvular heart disease. Lifespan in individuals with the attenuated form is variable, with death typically in adulthood from obstructive upper airway disease and cardiac disease.

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How is MPS II diagnosed?

The diagnosis of MPS II is based on elevated urine GAGs and deficient iduronate-2-sulfatase enzyme activity, with confirmation by DNA analysis. The Muenzer MPS Center will ensure all appropriate testing has been completed and will interpret all labs for diagnostic purposes.

How is MPS II treated?

The current recommended treatment for MPS II is weekly intravenous enzyme replacement therapy (ERT) using idursulfase, which replaces the absent or deficient enzyme. ERT can prevent or slow progression of some physical symptoms but does not affect the neurologic disease, as the recombinant human enzyme does not cross the blood-brain barrier. Although hematopoietic stem cell transplantation (HSCT) is recommended for Hurler syndrome (severe MPS I), it is not routinely recommended for **MPS II** due to limited clinical benefit. Additionally, other promising treatment options are currently under investigation in clinical trials.

Treatment options are currently available in North Carolina, and the UNC Muenzer MPS Center providers will discuss all therapeutic approaches with families during their follow-up visits.

Where do I go for more information?

Use your phone's camera to scan the QR codes below

- UNC Pediatric Genetics and Metabolism
- UNC Health Information and Referrals
- ACMG MPS II ACT Sheet
- Medline Plus

Where do I send parents for information?

Use your phone's camera to scan the QR codes below.

- Muenzer MPS Research and Treatment Center
- Baby's First Test
- National MPS Society
- Project Alive
- Newborn Screening Information Center

NC DEPARTMENT OF HEALTH AND HUMAN SERVICES

State of North Carolina Department of Health and Human Services Division of Public Health
www.ncdhhs.gov
https://slph.dph.ncdhhs.gov/newborn/default.asp
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This fact sheet was supported by Health Resources and Services Administration (HRSA), Department of Health and Human Services (HHS) as part of a financial assistance award totaling $344,348 with 100 percent funded by HRSA/HHS. The contents are those of the author(s) and do not necessarily represent the official views of, nor an endorsement, by HRSA/HHS or the U.S. Government.