

**IOWA**

State Hygienic Laboratory

# Screening for Mucopolysaccharidoses I and II

April 16, 2026



# Screening Methodology

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- No FDA approved methods incorporate MPSI and MPSII
- State Hygienic Lab screens for MPSI and II using a lab developed test
- Utilized Liquid Chromatography – Tandem Mass Spectrometry
- Multiplex method - Pompe Disease, MPSI, MPSII, Krabbe Disease

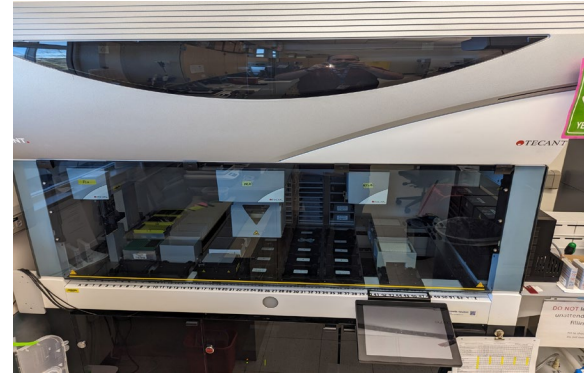
# Screening Methodology

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- Tests the activity levels of enzymes of interest:
  - MPSI: Alpha-L-Iduronidase(IDUA)
  - MPSII: Iduronate-2-Sulfatase(I2S)
- High False Positive rate due to the presence of Pseudodeficiency alleles
- Second tier testing through Mayo Clinic – reduces false positives
- Reporting is done based on the outcome of second tier testing
  - First tier results trigger reflex to reference lab for second tier
  - Positive second tier results are reported to providers

# Method Details

- LC -Tandem Mass Spectrometry
- 16 – 20 hours of incubation time
- Typically 18 hours
- Total processing time is around 19 hours
- Instrument run time is 5 hours per plate
- Total time to completion is 23 hours
- Testing is initiated and completed on night shift

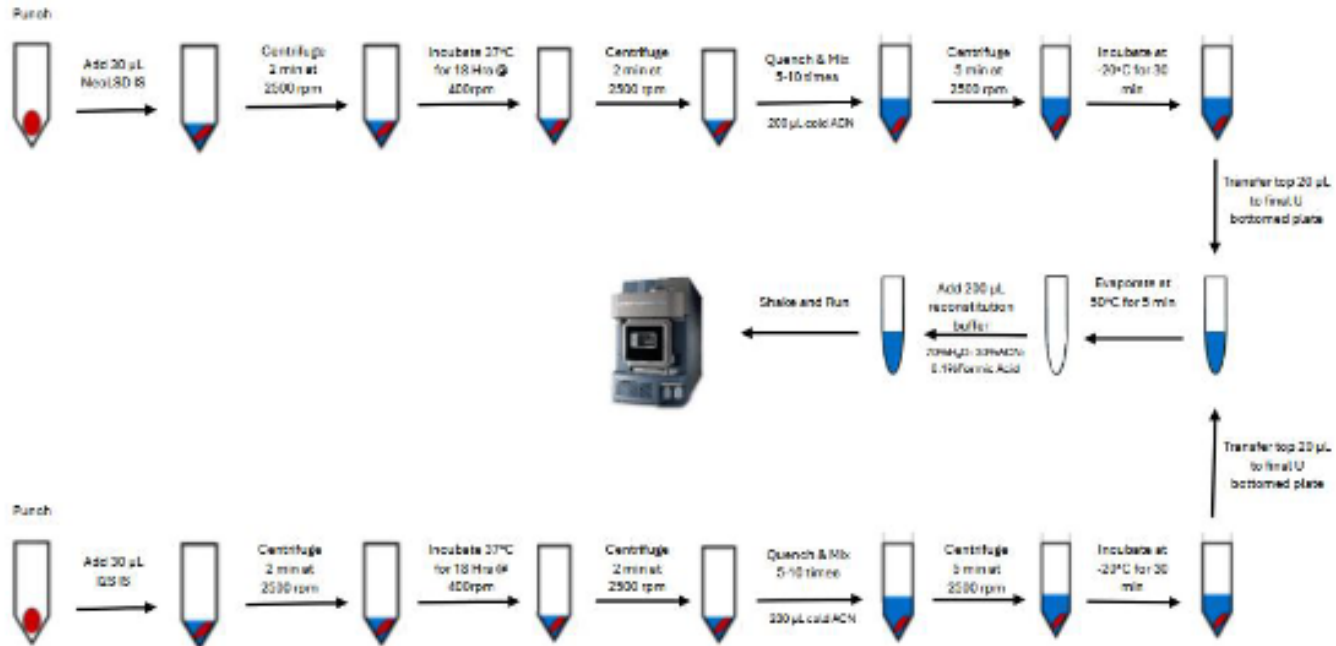


Tecan Fluent Liquid Handler



Waters TQ-S micro TMS instrument

# Procedure



# Test Performance

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- First tier – high false positive rate
- Second tier test for Glycosaminoglycan(GAG) level
  - Performed by reference lab – Mayo Clinic Labs
  - Not a good candidate for in-house development
- Turnaround Time
  - Mayo Clinic Labs(MCL) testing performed Mondays and Thursdays
  - Turnaround time can be 5-6 days from completion of first tier testing
  - MCL is in Rochester, MN and courier will pick up in Ankeny and deliver to MCL same day

# Test Performance

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- Currently screening ~4000 specimens per month for MPSI and MPSII
- 3-5 specimens reflexed per month for second tier testing for MPSI and MPSII combined (Jan-Apr 2026)
- More than 130,000 MPSI screens over 3 years
- More than 29,000 MPSII screens since 8/1/25
- 6 Presumptive Positive second tier screens
- MPSII – all abnormal referred for second tier if EA is low (X-linked condition)

# Testing Capacity

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- Test method:
  - In use for Pompe and validated for MPS1/MPSII
- Instrumentation:
  - Currently have 2 Waters TQ-S micro instruments dedicated for this screening
  - Advanced Robotic Liquid Handler
- Staffing:
  - Adequately staffed for this method

# Testing Capacity

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- Second tier testing:
  - Procedure in place for send outs
  - Reference lab is close
- Workflow Changes:
  - No difference for MPSI
  - Punch and process additional MPSII plate

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# Questions?

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