NEWBORN SCREENING FOR
POMPE DISEASE AND MPS-I

Washington State Department of Health
Newborn Screening for Pompe Disease and MPS-I

Washington State Board of Health
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Newborn Screening
Lysosomal Storage Disorders

- Lysosomes = cellular recycling center
  - Fats
  - Proteins
  - Long-chained sugar molecules
  - Rare inherited conditions
  - Defective Lysosomal Storage Disorders (LSDs)
  - 50 enzymes cause toxic build-up of metabolites
Recommended Uniform Screening Panel (RUSP)

- From Federal NBS advisory board

- Evidence review for candidate conditions
  - Pompe (2008) - no
  - Niemann-Pick disease (2008) - no
  - Fabry disease (2008) - no
  - Krabbe disease (2010) - no
  - Pompe disease (2013) - yes
  - Mucopolysaccharidosis type 1 (2015) - yes
University of Washington Pilot Study

- 2007 – Fabry disease screening
- 2009 – multiplex (Fabry, Pompe, MPS-I)
- 2012 – multiplex (9 LSDs)
- Current – feasibility studies for 15 conditions
- Tandem mass spectrometry (MS/MS)
  - Separate method from current MS/MS screening
Cost of Testing

- **Instruments**
  - Tandem mass spectrometers
    - Potential to multiplex with X-ALD
  - Digital microfluidics

- **Consumables**
  - Reagents, pipette tips, etc.

- **Staff time**
  - 1 FTE - front-line chemist
  - part FTE - lead worker
  - part FTE - follow-up specialist
Technical Advisory Committee Timing

- Window for technical advisory committee is small to prevent 1-year delay

- Decision package for NBS fee increase
  - Due in May 2017
  - Needs Agency’s approval and Governor’s support
  - Needs Legislature’s approval (2018)

- Potential to start screening as soon as July 2018
Questions

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