Preliminary Analysis of Four Lysosomal Storage Disorders Within the Context of Newborn Screening in Washington State

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Our Roadmap

- Background / Project Overview
- Preliminary Findings
- Lessons Learned
Our Roadmap

Background / Project Overview
Washington State Newborn Screening Criteria

- Available Screening Technology
- Diagnostic Testing/Treatment Available
- Prevention Potential and Medical Rationale
- Public Health Rationale
- Cost-Benefit/Cost-Effectiveness
NBS for LSDs?

• Development of screening assays
• New therapies that are most effective when delivered early in disease progression
• Good target for newborn screening?
The MPH Practicum Process

Assess 4 LSDs against the WA State NBS Criteria

Literature review

Parse/Structure Data

Summary Documents
Preliminary Findings: Pompe Disease

- Available Technology
- Diagnosis/Treatment
- Prevention Potential
- Public Health Rationale
A Closer Look: Pompe Disease

• How do late-onset cases fit in?
• “Patients in Waiting”
• Enzyme replacement therapy can provide benefits once symptomatic
Preliminary Findings: Fabry Disease

Available Technology

Diagnosis/Treatment

Prevention Potential

Public Health Rationale
A Closer Look: Fabry Disease

• Females not simply “carriers”
• Current methodologies will miss some female heterozygotes
• Adequate sensitivity?
Preliminary Findings: MPS I

- Available Technology
- Diagnosis/Treatment
- Prevention Potential
- Public Health Rationale
A Closer Look:
MPS I

• Difficult to determine phenotype in newborn period
• Large disease burden persists with treatment
• Are current treatment outcomes sufficient for a mandatory screen?
Preliminary Findings: Krabbe Disease

- Available Technology
- Diagnosis/Treatment
- Prevention Potential
- Public Health Rationale
A Closer Look: Krabbe Disease

- Prevalence of early-onset much lower than anticipated
- Difficult phenotype predictions
- Research/risk-based protocol rather than mandatory?
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The Product: Summary Documents
Guiding Principles

Decision should be driven by evidence

All screen positive infants should have access to diagnostic/treatment services

Benefits of screening should outweigh harm to families, children, society
Take-Aways & Lessons Learned

• Importance of systematic process to evaluate existing evidence
• Useful tool to direct discussions
• With careful planning, collaborative projects can be mutually beneficial
Come visit our lab!

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