

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

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*Saving lives with
a simple blood spot*

Our Roadmap



Background / Project
Overview



Preliminary Findings



Lessons Learned

Our Roadmap

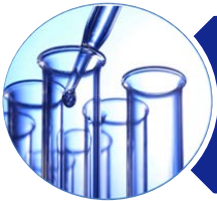


Background / Project
Overview

Washington State Newborn Screening Criteria



Available Screening
Technology



Diagnostic Testing/
Treatment Available



Prevention Potential and
Medical Rationale



Public Health Rationale

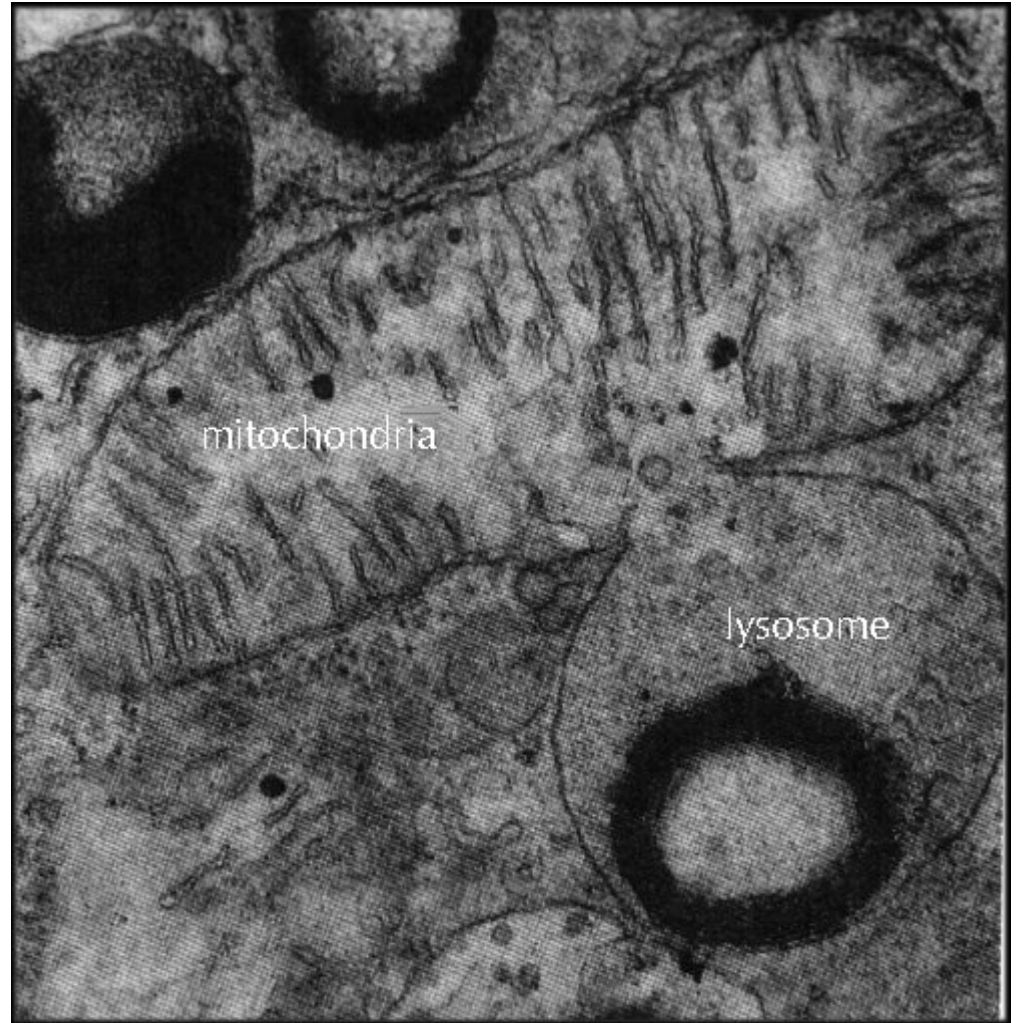


Cost-Benefit/
Cost-Effectiveness



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

Our Roadmap



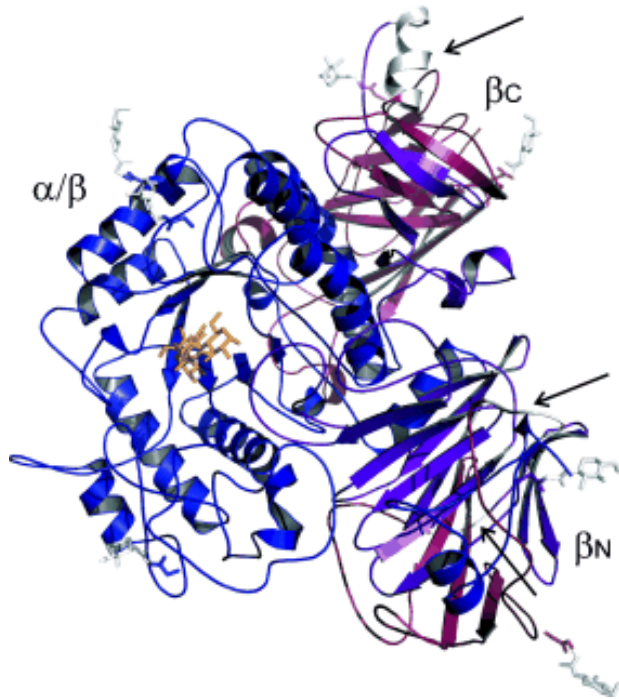
Background / Project
Overview



Preliminary Findings



Preliminary Findings: Pompe Disease



Available Technology



Diagnosis/Treatment



Prevention Potential



Public Health Rationale



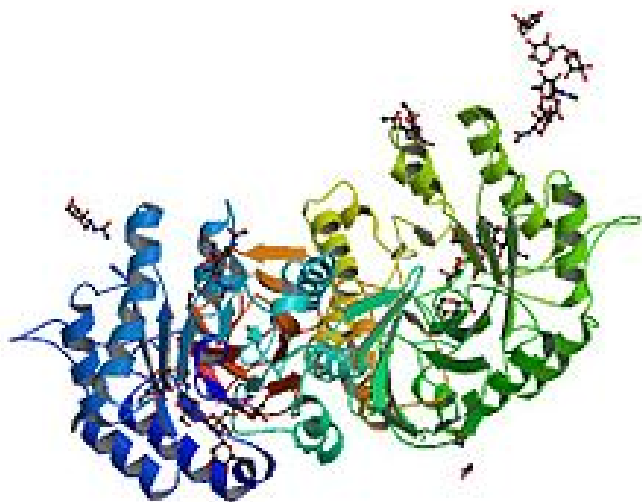
A Closer Look: Pompe Disease



- 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



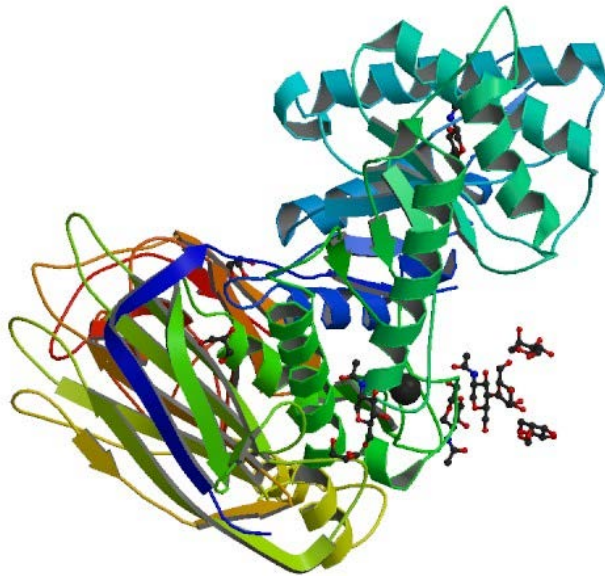
A Closer Look: Fabry Disease



- 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



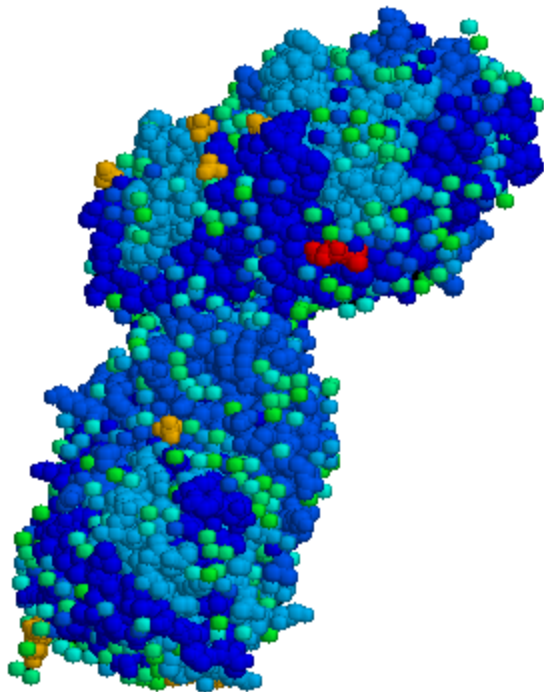
A Closer Look: MPS I



- 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?

Our Roadmap



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Preliminary Findings



Lessons Learned



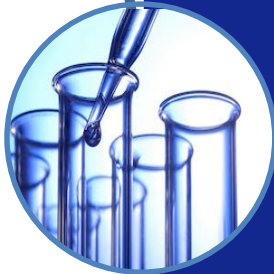
The Product: Summary Documents



Guiding Principles



Decision should be driven by evidence



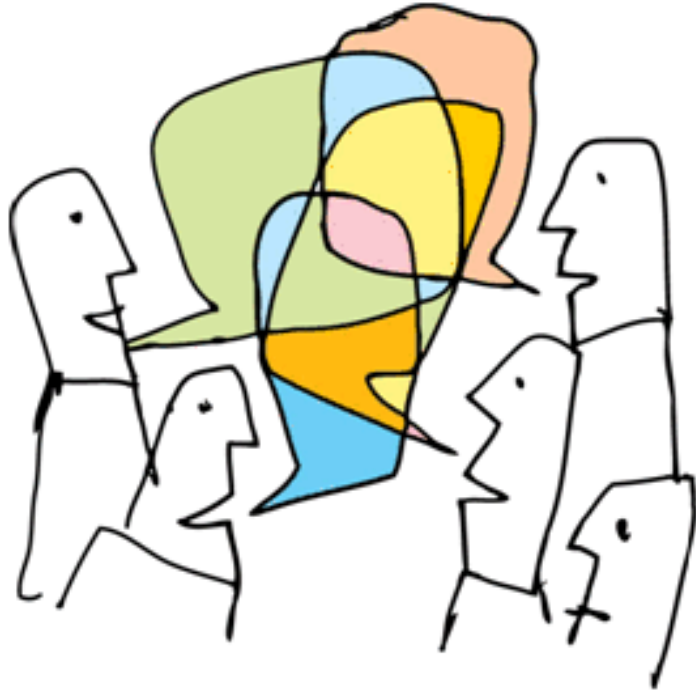
All screen positive infants should have access to diagnostic/treatment services



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



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