X-ALD Implementation Update

July 16, 2018

Recommended Uniform Screening Panel

Texas Health and Safety Code Sec. 33.011.

To the extent funding is available for the screening, the department shall require newborn screening tests to screen for disorders listed as core and secondary conditions in the Recommended Uniform Screening Panel

RUSP – Recent Additions

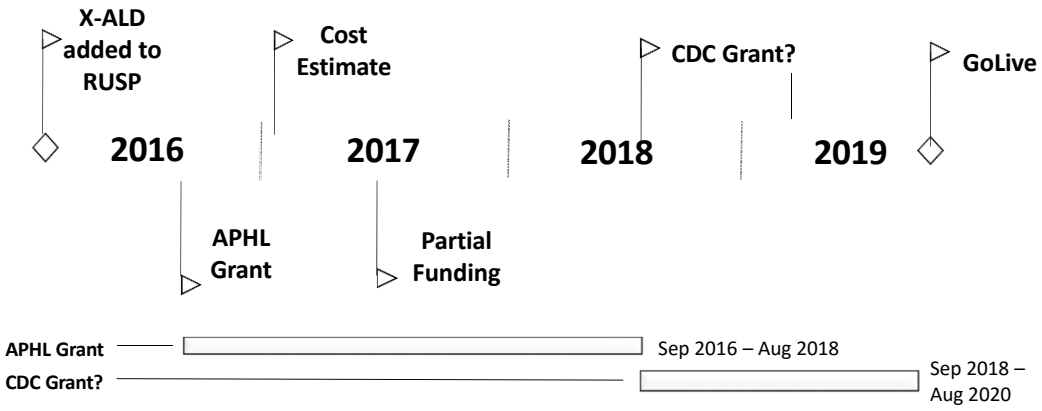
Pompe:
March 2015

Mucopolysaccharidosis Type I (MPS1):
February 2016

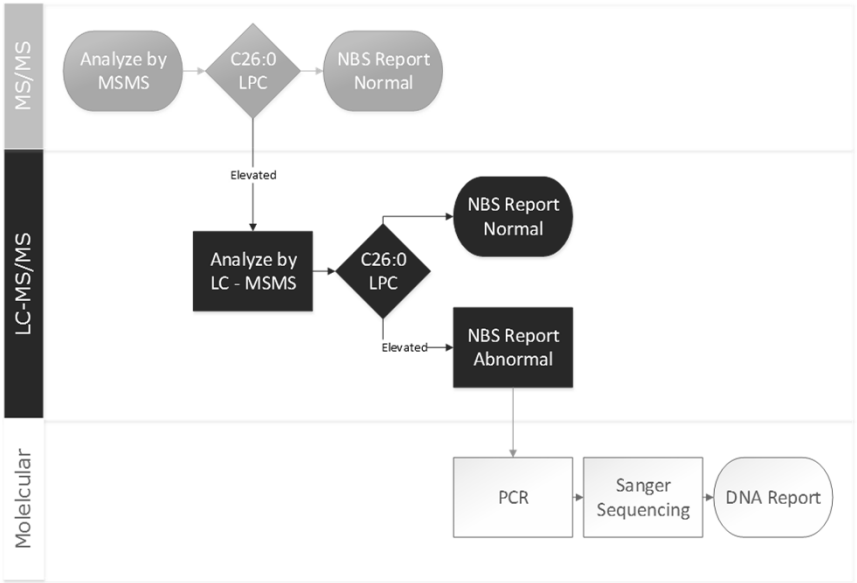
X-linked Adrenoleukodystrophy (X-ALD):
February 2016

Spinal Muscular Atrophy (SMA):
July 2018

Overall Timeline



Proposed Algorithm



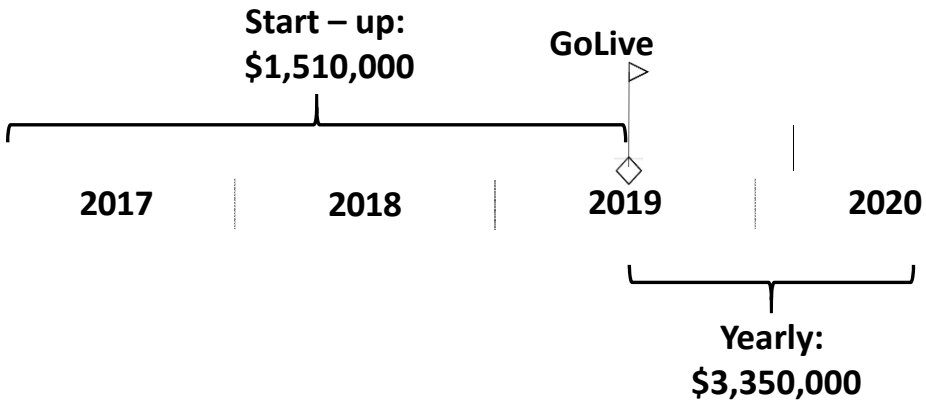
Mutational Analysis of X-ALD

- PCR and Sanger sequencing of all exons
- Testing all NBS specimens with 'Abnormal' X-ALD screening results
  - ~154 specimens per year
- Turnaround time: ~14 working days

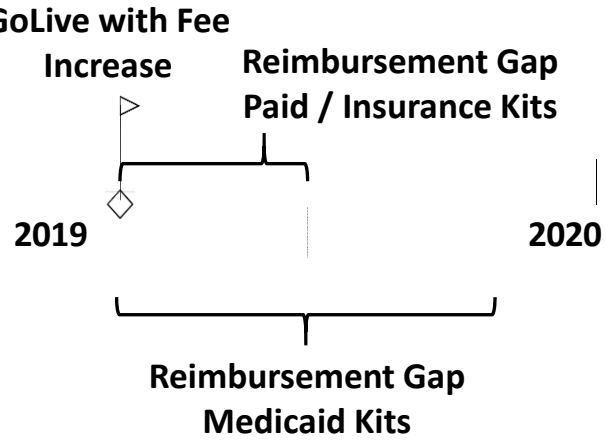
X-ALD Cost Estimate

1. Building Retrofit (MSMS and Molecular)
2. Lab FTEs
3. Reagents, supplies, equipment
4. Validation of methodologies / equipment
5. Information System (LIMS) modifications
6. Educational materials
7. Training
8. Office equipment, computers, supplies

X-ALD Cost Estimate



X-ALD Cost Estimate



X-ALD Funding

Start-up Costs
+ Testing Costs (gap)
$3,510,000

- Legislative Appropriation ($1,200,000)
($2,310,000)

APHL New Disorders Grant

- 2 Year grant (~$120,000)
- Goals - Develop:
  - Ability to perform second-tier X-ALD sequencing
  - Follow-up algorithms for abnormal X-ALD screens
  - Clinical referral network
  - Develop educational materials for families, providers, and the general public

APHL Grant Accomplishments

- Molecular Testing
  - Received technical training
  - Purchased instruments, reagents, and supplies
  - Completed Building retrofit
  - Developed Validation Plan
- Clinical Care Coordination
  - Hosted consultants meetings
  - Developed educational brochure
  - Drafted follow-up flows
  - Scheduled staff training

X-ALD Funding

Start-up Costs
+ Testing Costs (gap)
———————————————————
$3,510,000

- Legislative Appropriation
- APHL Grant
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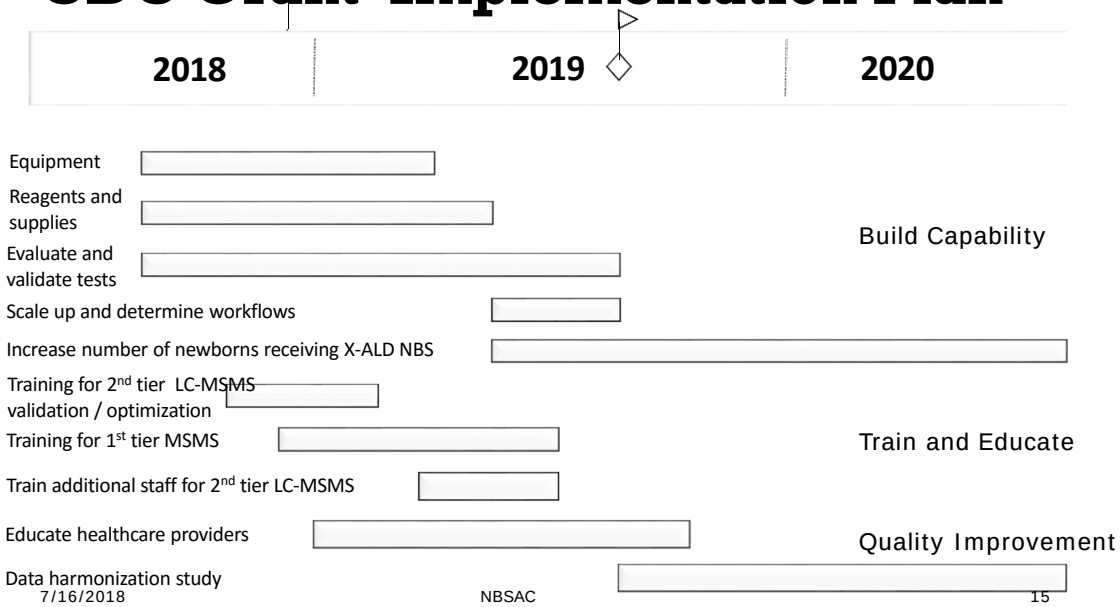
($2,190,000)

CDC New Condition Implementation Grant

- 2 Year grant (~$600,000)
- Goals - Supplement appropriation to:
  - Acquire reagents, supplies, and equipment (validation and after go Live)
  - Optimize / validate / scale up test methodologies and protocols
  - Train staff
  - Educate healthcare providers
  - Participate in inter-laboratory data harmonization

CDC Grant Implementation Plan



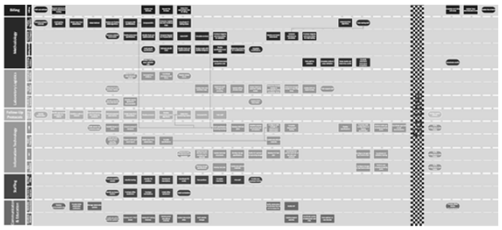
X-ALD Funding

Start-up Costs
\+ Testing Costs (gap)
———————————————————
$3,510,000

\- Legislative Appropriation
\- APHL Grant
\- CDC Grant
———————————————————
($1,590,000)

X-ALD Implementation - Next Steps

- Additional funding
- Project planning
  - Staffing
  - Finalize test algorithms
  - Finalize follow-up protocols
  - Information systems
  - Laboratory logistics
  - Communications
  - Educational materials



Thank you

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