Update from Washington State

John D. Thompson
Washington State Department of Health,
Newborn Screening Program

Current Status: Pilot Studies

THE SECRETARY OF HEALTH AND HUMAN SERVICES

WASHINGTON, D.C. 20201

MAR 0 2 2015

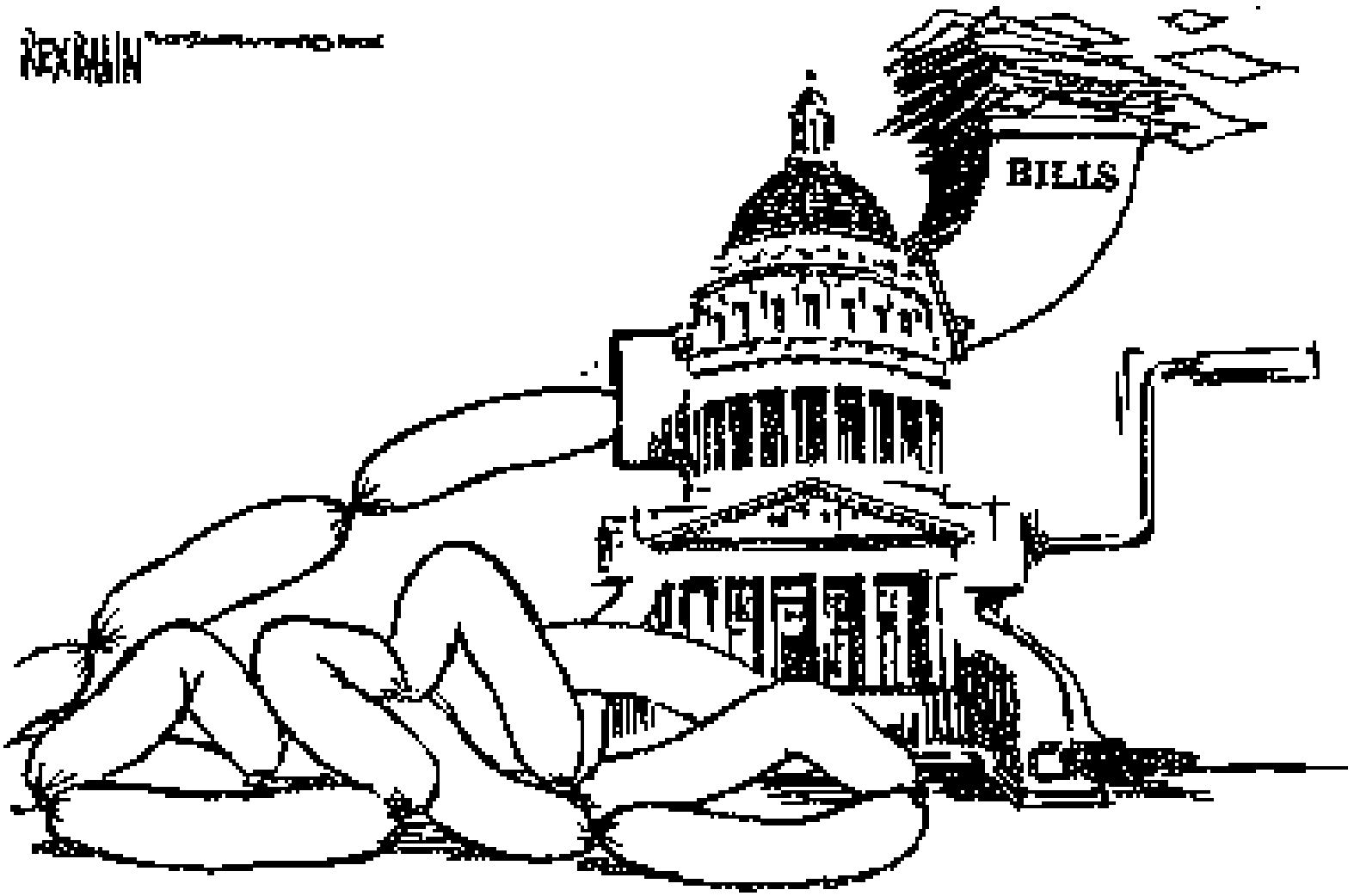
Joseph A. Bocchini, Jr., MD
Committee Chairperson
Discretionary Advisory Committee on Heritable Disorders
in Newborns and Children
Professor and Chairperson
Department of Pediatrics
Louisiana State University
1501 Kings Highway
Shreveport, LA 71130

Dear Dr. Bocchini:

Taking into consideration the information presented in these reports, I accept the DACHDNC recommendation to add Pompe disease to the RUSP. The Affordable Care Act requires that most health plans cover the evidence-informed preventive care and screenings provided for in the comprehensive guidelines supported by Health Resources and Service Administration (HRSA). Because the RUSP is a component of these guidelines, a condition added to the RUSP must be covered. It should be understood that addition of Pompe disease to the RUSP does not constitute a requirement for states to implement screening, only a recommendation. I recognize the complex issues surrounding newborn screening for Pompe disease and encourage Federal agencies to support states as they build capacity and implement state-wide screening.

Washington State Department of Health

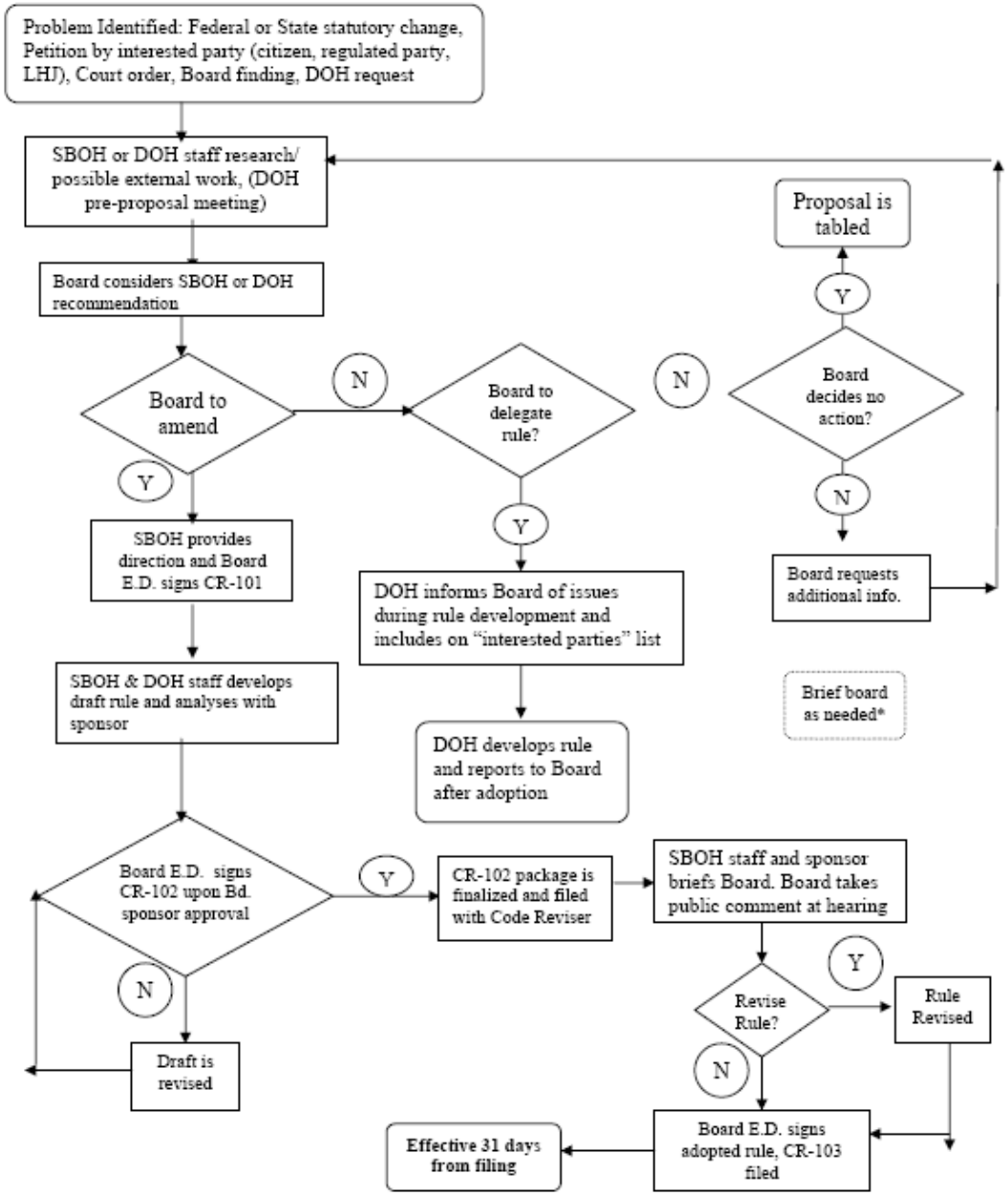
Rule Making Process



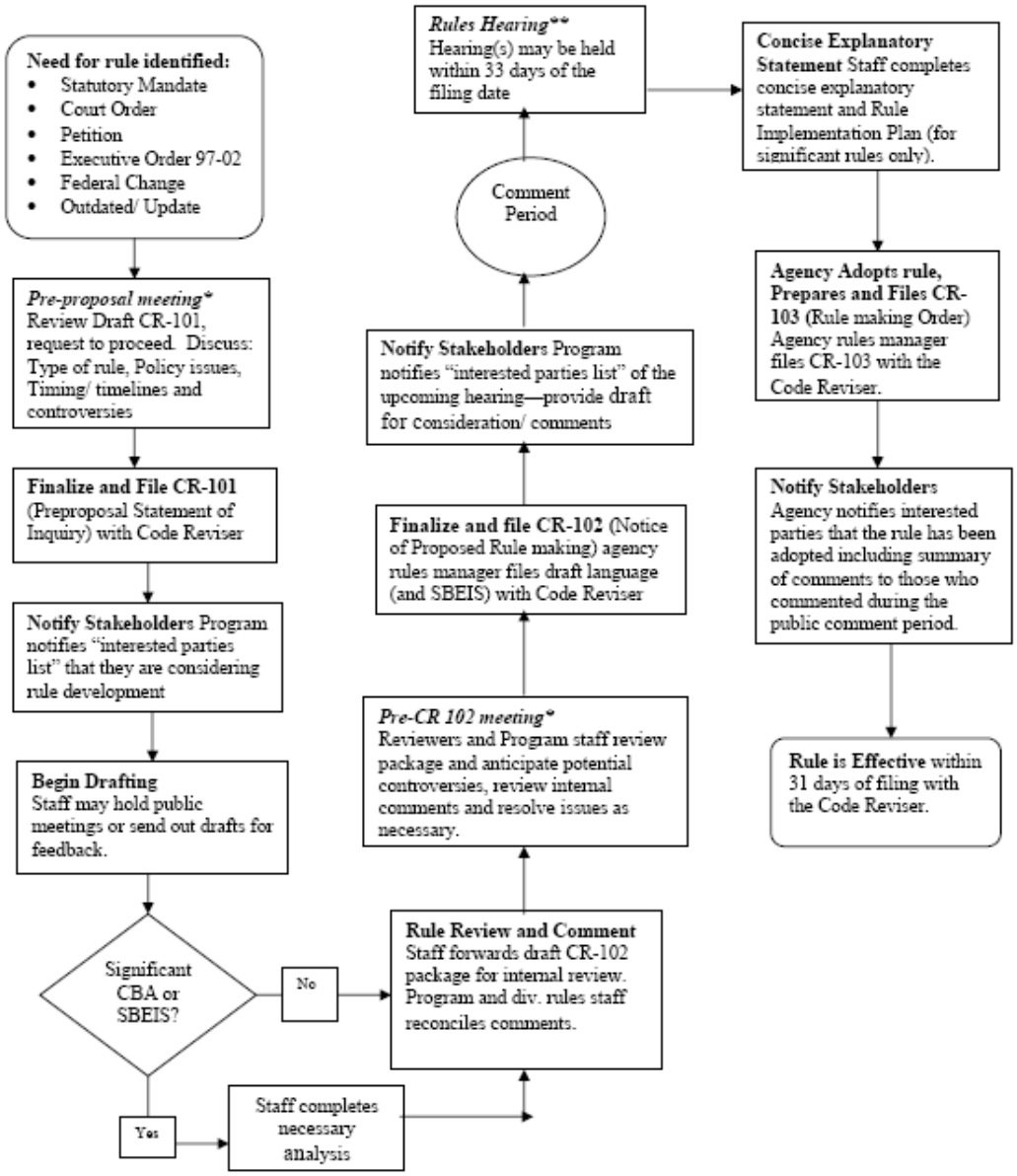
“I have come to the conclusion that the making of laws is like the making of sausages – the less you know about the process the more you respect the result.”

- Frank W. Tracy quoting an unnamed member of the Illinois House of Representatives (about 1878)

State Board of Health Rule Making Process

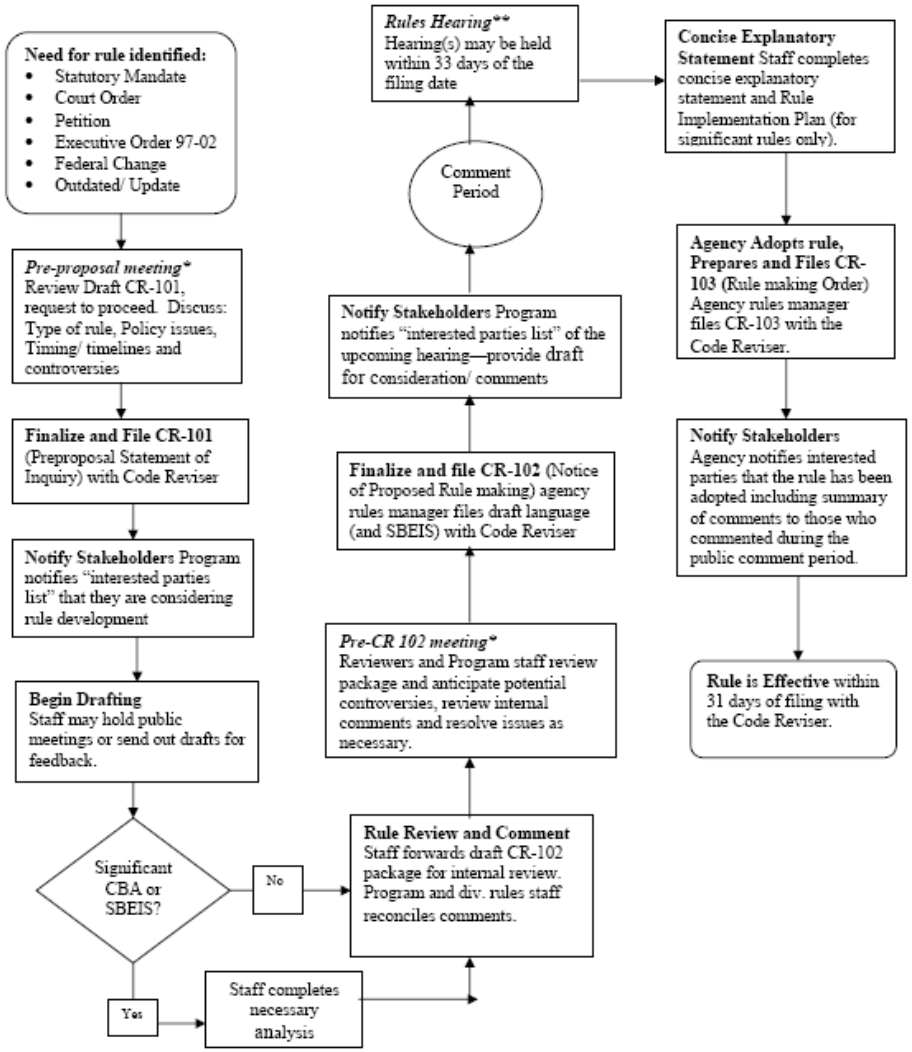


Department of Health Rules Process Flow Chart



* Division rules contacts determine whether these meetings are necessary.
** A public hearing is not necessary for rules adopted under expedited adoption.

Department of Health Rules Process Flow Chart



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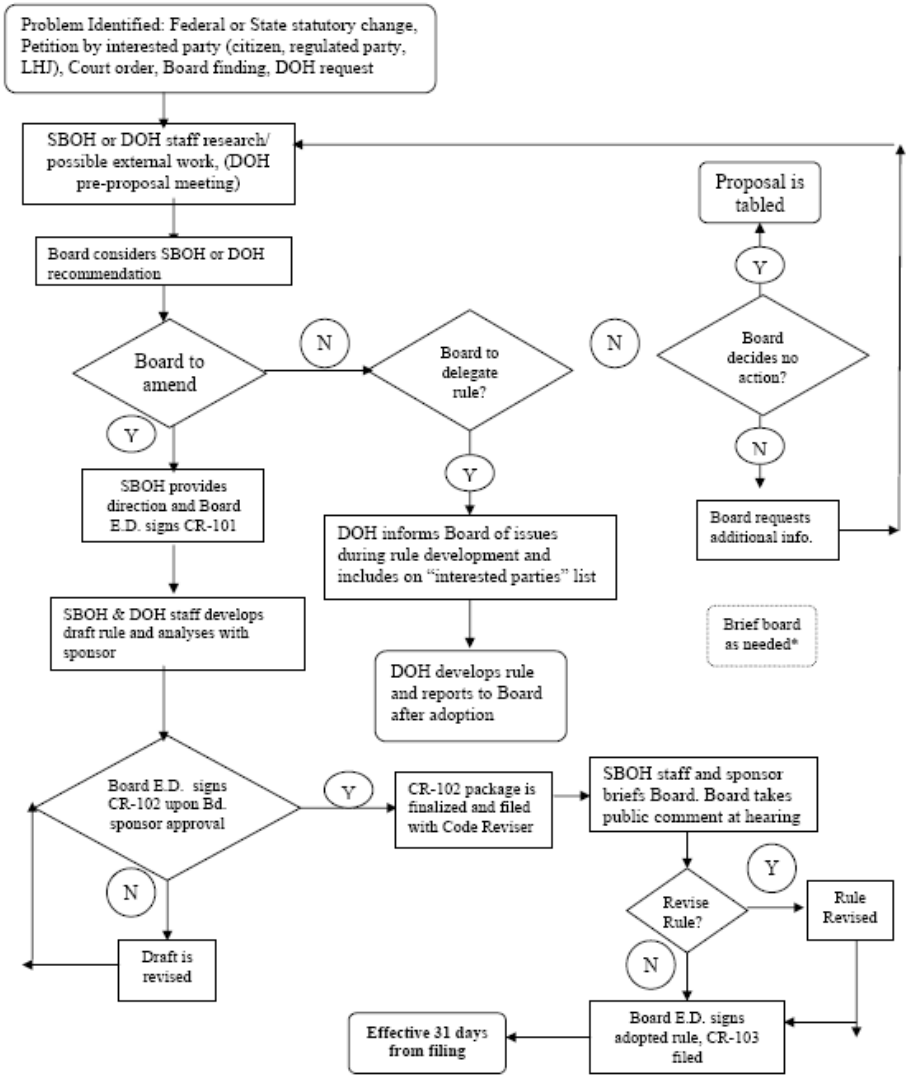


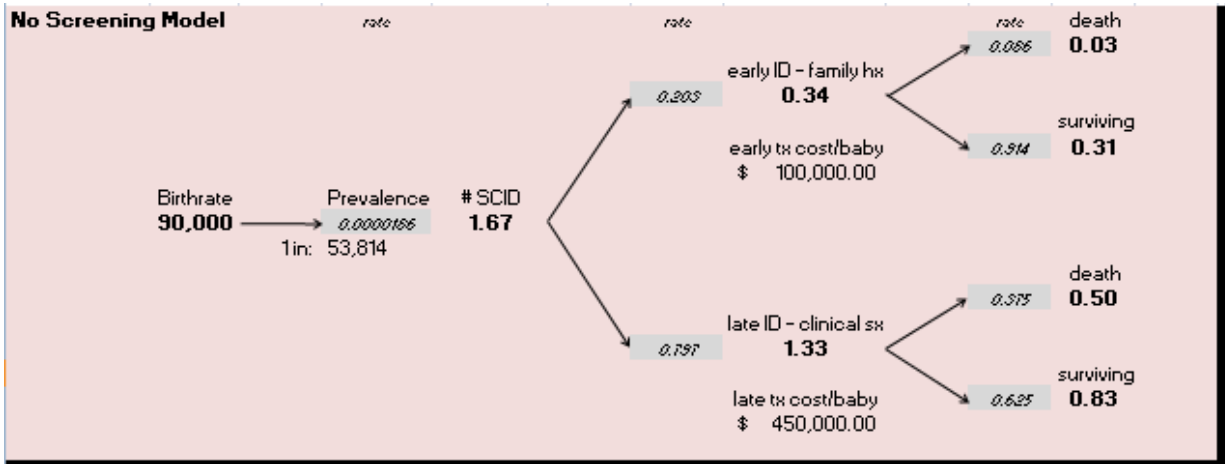
Washington Criteria for NBS

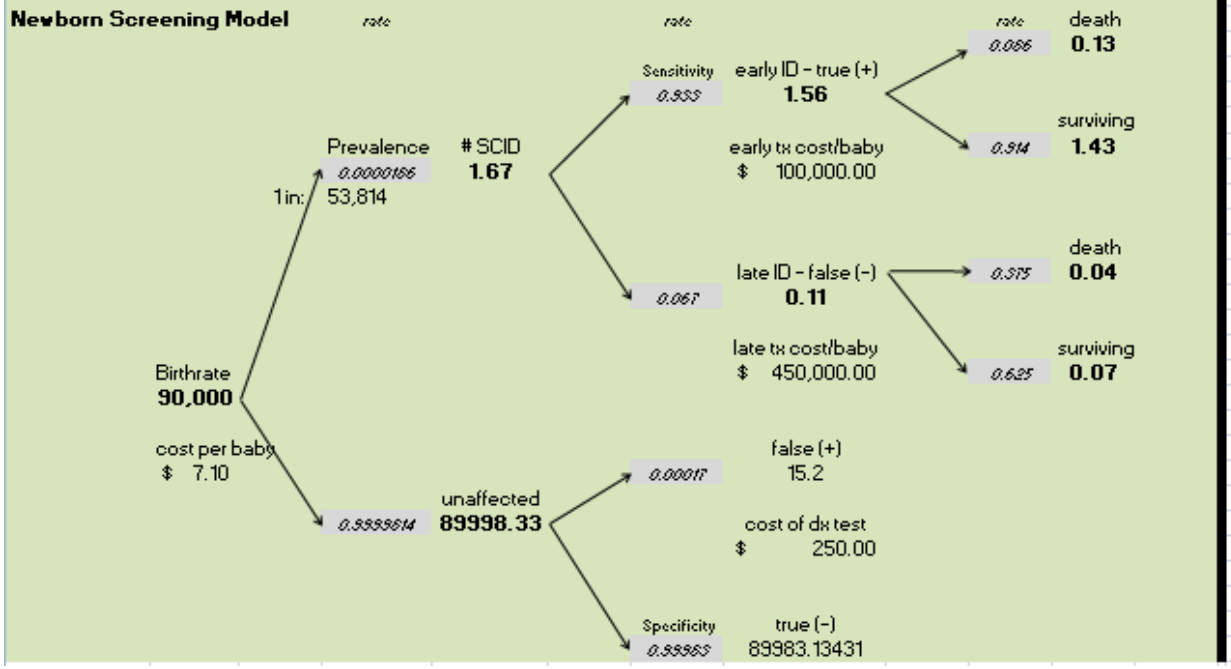
- Available Screening Technology
- Diagnostic Testing and Treatment Available
- Prevention Potential and Medical Rationale
- Public Health Rationale
- Cost-Benefit/Cost-Effectiveness

Cost-Benefit Overview

- Decision Tree
  - compares status quo v. screening model
- Data from
  - primary literature
  - reports from NBS programs
  - expert opinion
- Sensitivity analysis – vary assumptions
  - lower and higher estimates for parameters

SCID Benefit-Cost Analysis





No Screening	deaths	0.53
	surviving	1.14
	early tx costs	$ 33,872.17
	late tx costs	$ 600,172.53
	total tx costs	$ 634,044.70

Screening	deaths	0.18
	surviving	1.50
	early tx costs	$ 156,038.51
	late tx costs	$ 50,424.02
	total tx costs	$ 206,462.53

SHIFT

Benefits	deaths averted	0.35
	value of a life	$7,700,000.00
	value of lives saved	$ 2,712,005.92
	less tx costs	$ 427,582.17
	TOTAL benefits	$ 3,139,588.10

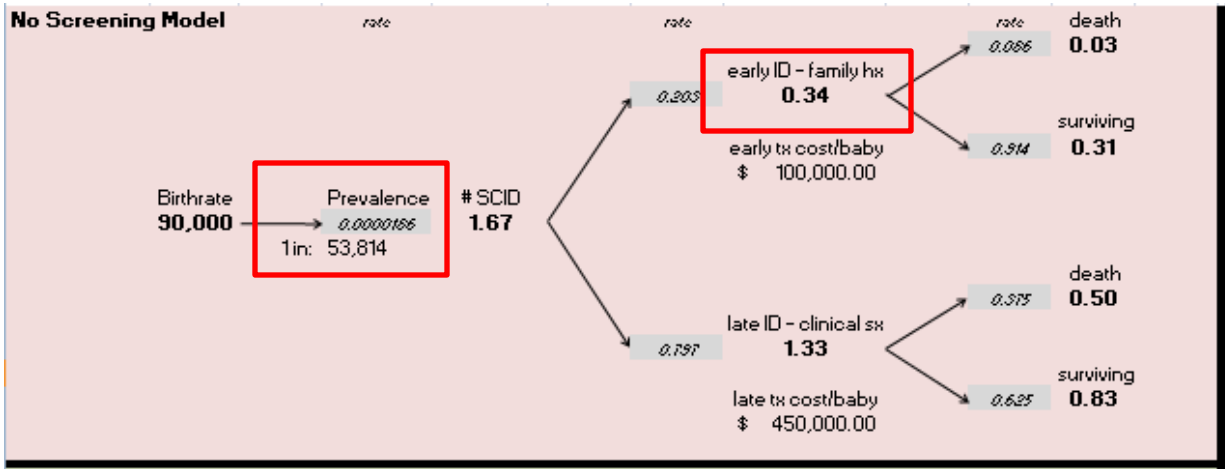
Costs	costs of screening	$ 639,000.00
	costs of false(+)	$ 3,798.31
	TOTAL costs	$ 642,798.31

	Benefit/Cost ratio	4.884250678

Key Parameters

- Prevalence of Pompe Disease
  - IOPD
    - Classic form
    - Non-classic form
  - LOPD
- Screening test performance
  - Sensitivity
  - Specificity
  - Positive Predictive Value (PPV)
- % of affected babies with (+) family history

Benefit-Cost Analysis



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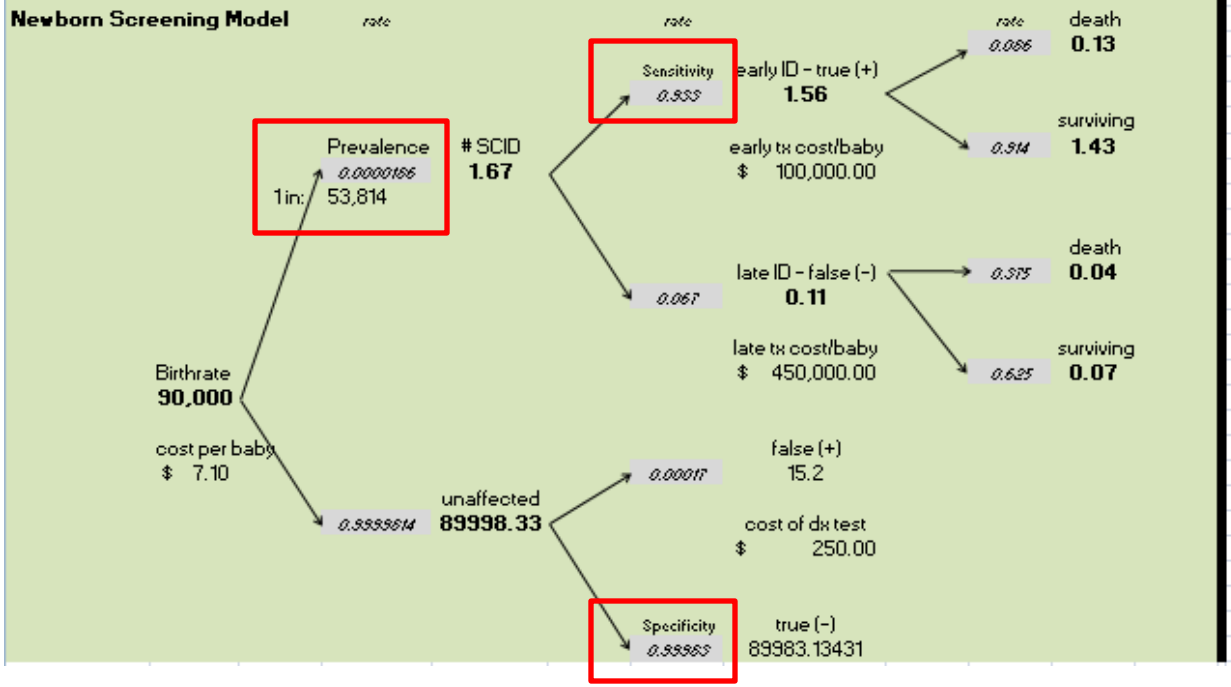

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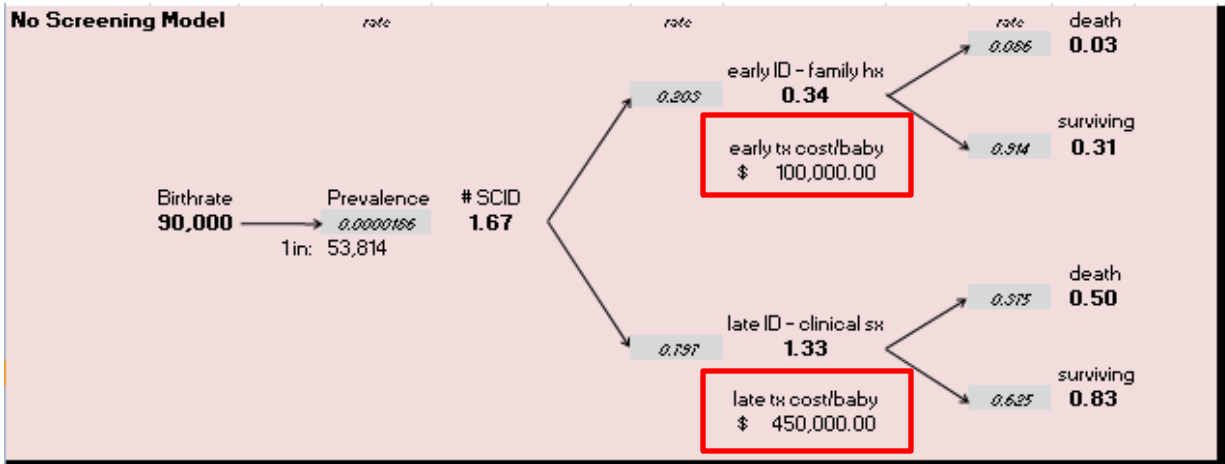
Costs

- Cost of screening
- Cost of diagnostic testing
- Cost of treatment – late diagnosed
- Cost of treatment – early diagnosed
- Cost of false (+)
- Cost of monitoring for LOPD

Benefit-Cost Analysis



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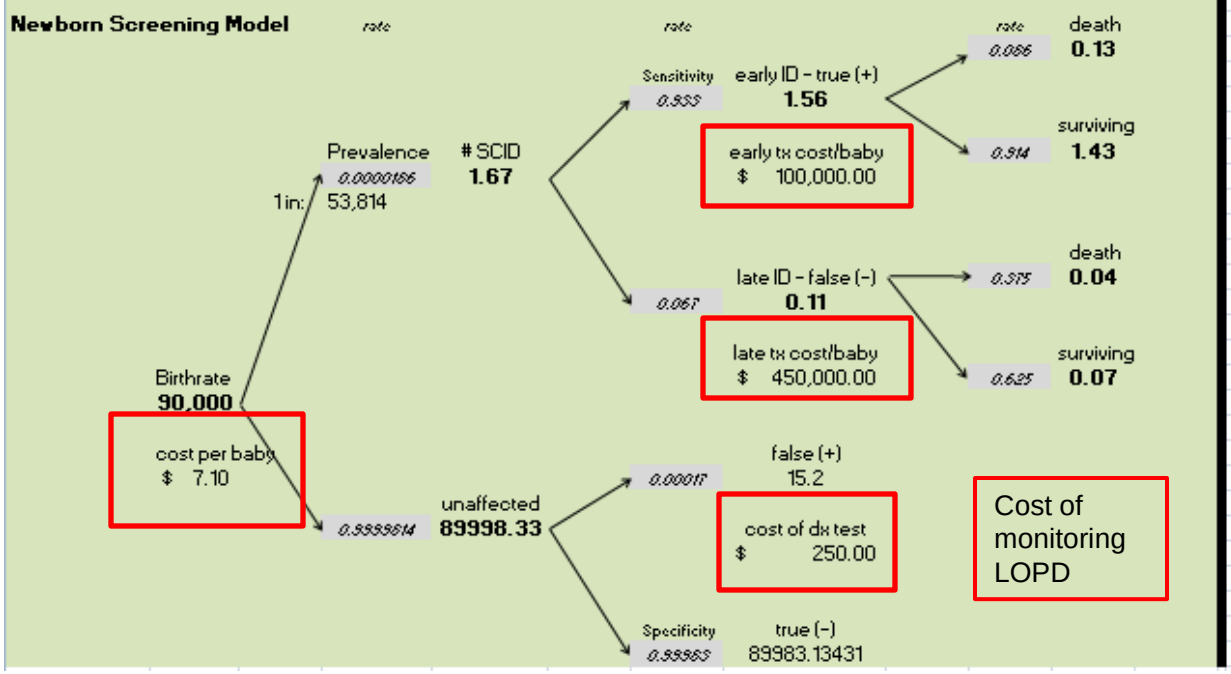

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Final Cost Analysis

- Benefits
  - Deaths averted
  - Value of lives saved
  - Less treatment costs
- Costs
  - Cost of screening
  - Cost of false (+)
- Benefit/Cost Ratio
- Incremental Cost-Effectiveness Ratio

LSD pilot v. live NBS

- Blinded study – one punch to get it right
  - if positive → DNA
- Chance to repeat test
  - positive initial results
  - failed runs

- 1st screen only: typically 18-48h
- 1st screen: 18-48h
- 2nd screen: 7-14d

- Testing performed when DBS is 30+ days old
- Testing performed in real time

LSD pilot v. live NBS

- Volume
  - 3-6 plates/day
- Volume
  - 5-14 plates/day

- Instruments
  - 1 MS/MS
- Instruments
  - need redundancy
  - need space

- Staff
  - one part-time tech
  - one to crunch numbers
- Staff
  - needs dependent on # of LSDs mandated


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