

2018 APHLTM ANNUAL MEETING

and twelfth government environmental laboratory conference

NBS follow-up (it's complicated too)

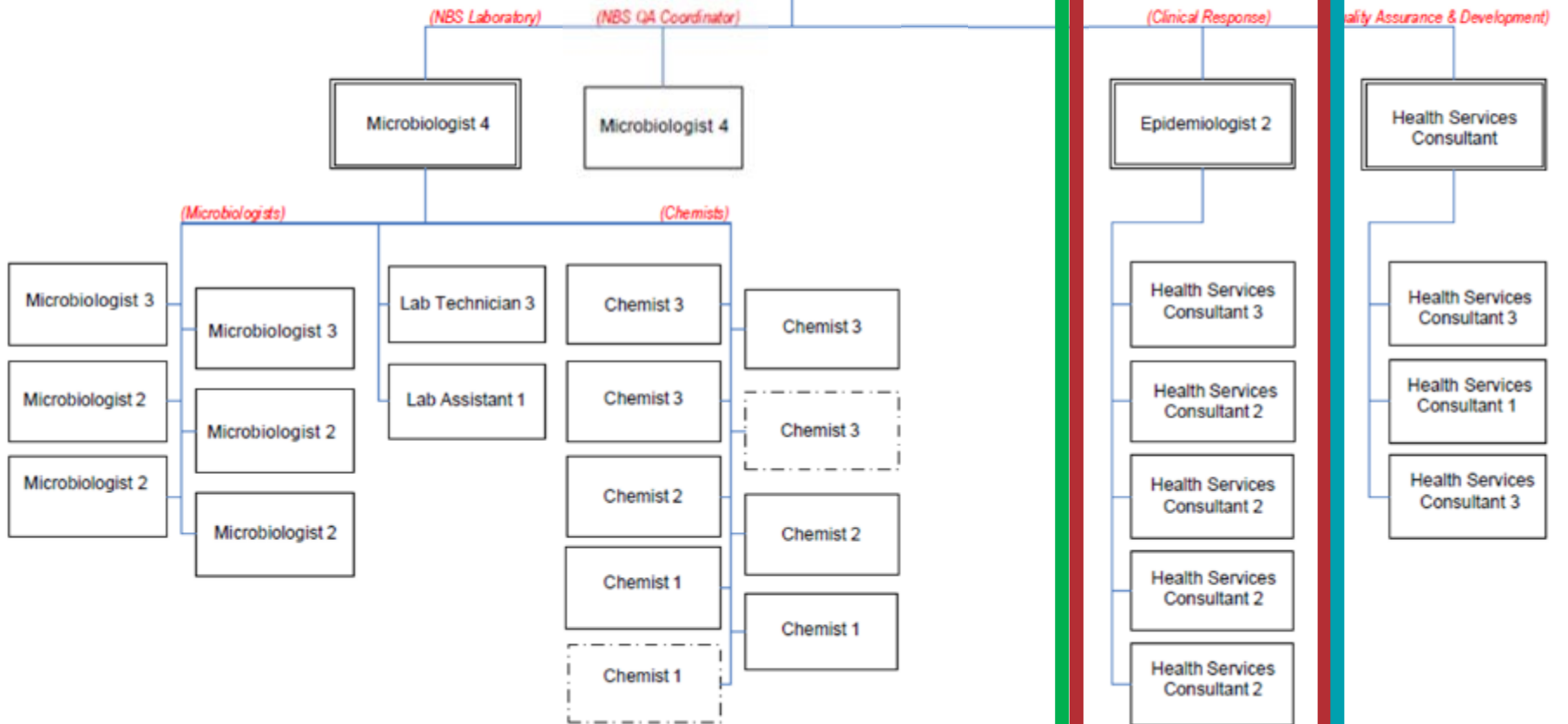
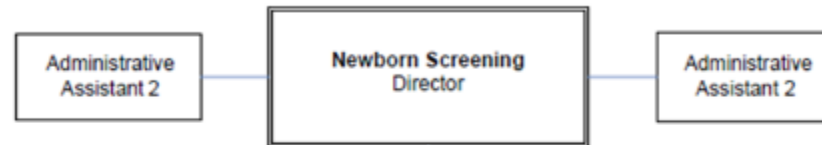
John D. Thompson, PhD, MPH, MPA
Director, Newborn Screening Program
Washington State Public Health Laboratories



Duty – Primary manager of the NBS Clinical Response Section:

- **Oversee and supervise follow-up staff ensuring that**
 - **all abnormal test results are reported to the child's health care provider and**
 - **the appropriate clinical response for each baby with abnormal results occurs**

WA State NBS Org Chart



Follow-up of Abnormal Results

Dedicated 'disorder follow-up' team

- Plan our response based on the case data
 - Borderlines
 - Presumptive Positives
 - Urgent Referrals
 - After confirmed diagnoses, ensure baby is linked into specialty care



Key: Communication

Challenging to communicate effectively with clinicians about cutoffs/risk determination

- Analyte values (primary, secondary, ratios)
- Demographics (age at collection, birthweight)
- Test performance (sensitivity, specificity, predictive value)
- Result reports and recommendations
- Educational materials



Specimen Quality



Rely on external partners

- Good quality specimen
- Accurate information
- Quick transit time



Importance of education,
surveillance and reporting



Case Study

Initial hemoglobin results were indicative of transfusion (AA)

Results from second NBS were inconsistent with first results (FA)

Analyzed all four blood spots – obviously blood from two individuals: one baby and one adult

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NEWBORN SCREENING WASHINGTON STATE DEPT. OF HEALTH

P.O. BOX 55729 (1610 NE 150th St)
SHORELINE, WA 98155-0729; Ph (206) 361-2902



MOTHER'S INFORMATION

[Redacted Mother's Information]

CHILD'S INFORMATION

Birth: am pm
Collection:
Name: _____
Medical: _____
Sex: M F
Birthweight: _____ ounces
Transfusion: _____
Race: _____
Ethnicity: _____

SUBMITTER & PROVIDER INFORMATION

OPTIONAL
USE =>

SUBMITTER:

(Check one)

PROVIDER

(Check one):

[Redacted Submitter & Provider Information]

IF TEST IS REFUSED BY PARENT, CHECK HERE
(SIGNATURE IS REQUIRED ON BACK OF FORM)



1565925

LOT # W-001
1565925
pan



Case Study

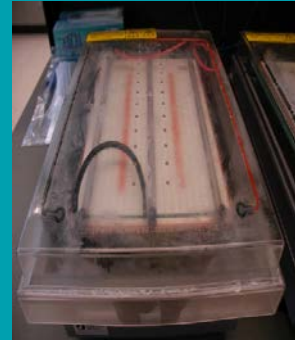
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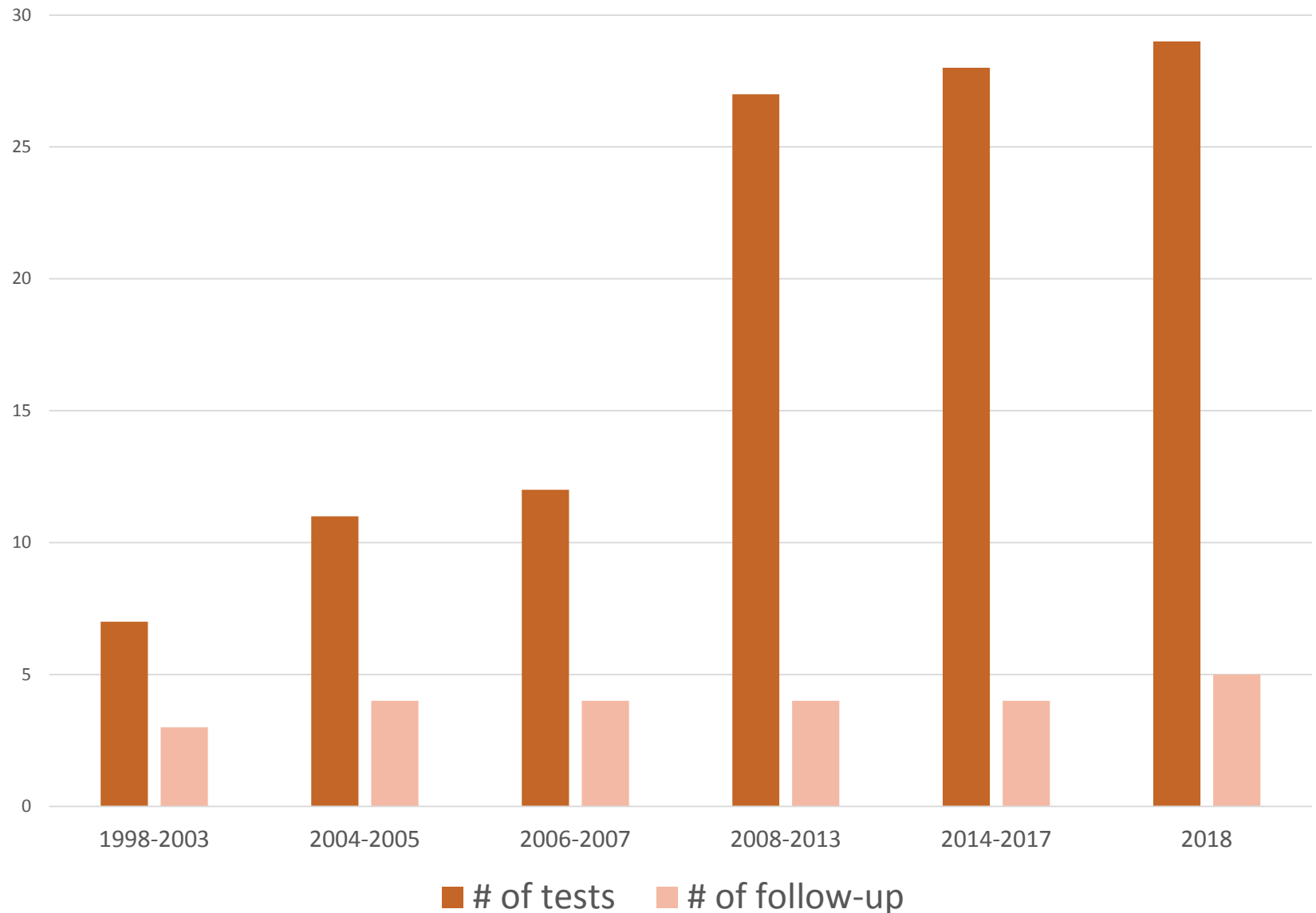
Baby did not bleed well and phlebotomist supplemented with someone else's blood to fill circles

Letter from our Program Director to Hospital Management about this dangerous practice



NBS expansion

NBS Expansion (1998-2018) – WA State



Implications of NBS Expansion

- Easy to justify hiring lab staff with new tests
- More difficult for follow-up, support, education

Increasingly difficult to train new staff – level of complexity, nuances

Specialty Care Providers

Regional hubs

- Present challenges (eg. small states, large states)

Identify clinical partners

- CF – pulmonologists, laboratory, nurse managers, dieticians, social workers
- X-ALD
 - neurology (new)
 - endocrinology (old friends)
 - biochemical genetics (old friends)
 - Where is the medical home?

Long-term Follow-up

Newest conditions have LTFU component

- X-ALD
- LSDs
 - early-onset v. late-onset
 - expensive treatment (Pompe)

Public Health/Clinical – who takes care of LTFU?

- Role for advocacy organizations?

Education and Outreach

Not usually budgeted into fee increases

QA efforts – importance of providing a quality specimen

Trend for more out-of-hospital births

Federal funds through NewSTEPs 360

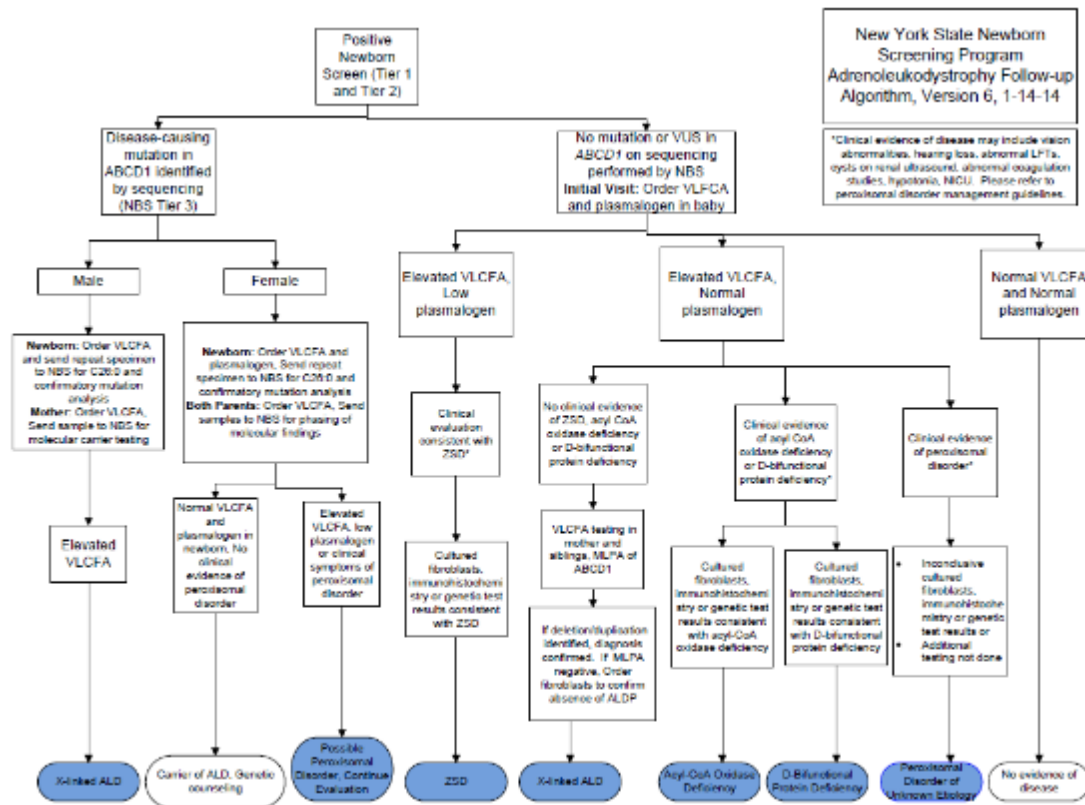


NBS expansion

X-ALD follow-up

MS/MS assay – straight forward

Diagnostic testing (VLCFA analysis) - tricky



X-ALD follow-up

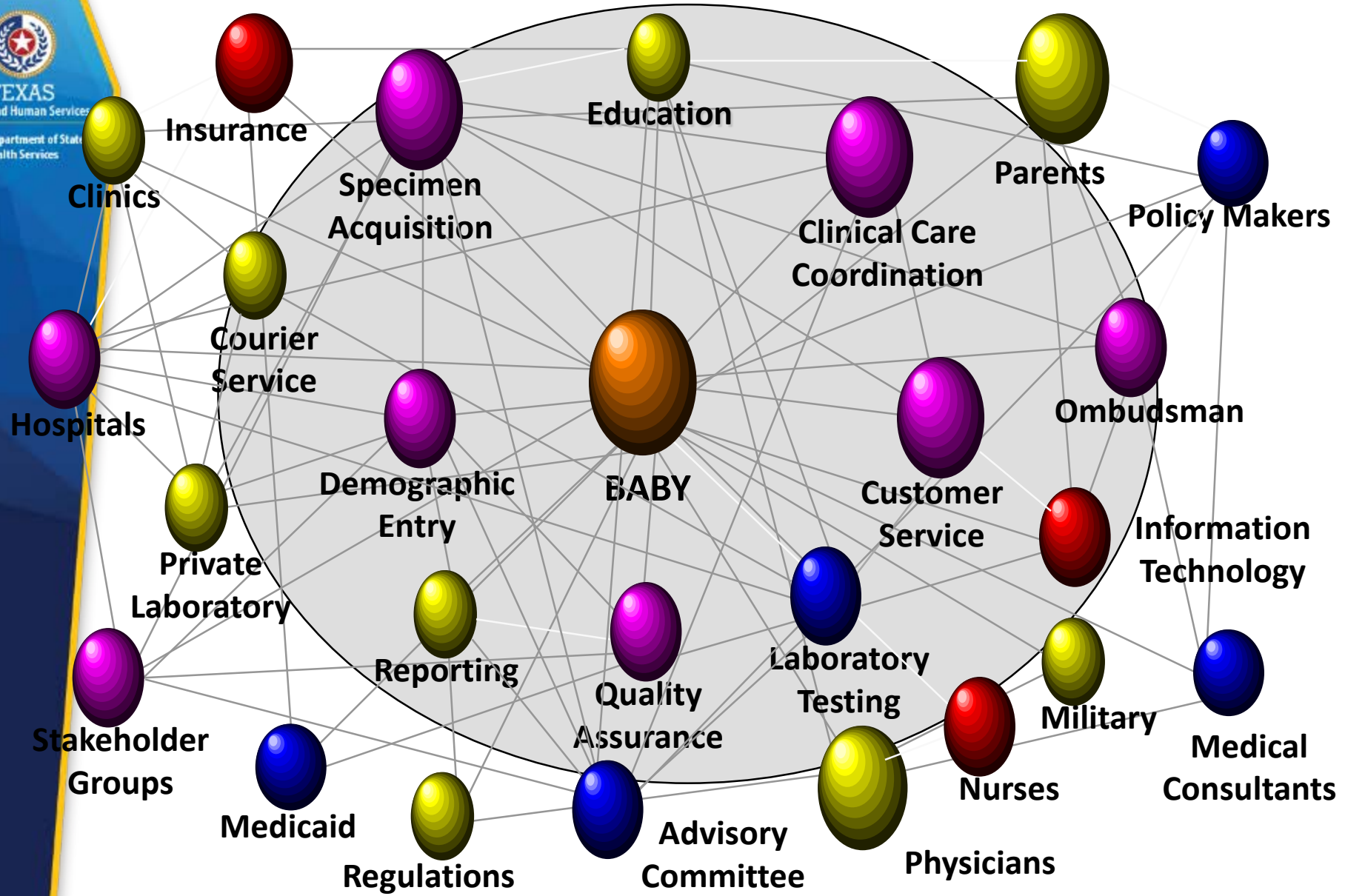
What parents can expect:

- Three (main) subtypes
 - 80% - adrenal insufficiency
 - 35% - cerebral involvement (most severe)
 - 45% - late-onset neurological involvement
- No genotype/phenotype correlation
- Serial testing (yearly to adulthood)
 - Adrenal function
 - Brain MRI
- Long-term follow-up



NBS: it's complicated

Newborn Screening System



**Goal: foster collaboration
and support for NBS**

**Pathway: learning followed
by consistent and proactive
communication**