NBS follow-up (it’s complicated too)

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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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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

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

The bar chart shows the number of tests and follow-ups over different time periods:

- **1998-2003**: Low number of tests and follow-ups.
- **2004-2005**: Increase in the number of tests and follow-ups.
- **2006-2007**: Further increase in both tests and follow-ups.
- **2008-2013**: A significant rise in the number of tests and a slight increase in follow-ups.
- **2014-2017**: Continued high number of tests with a steady number of follow-ups.
- **2018**: The highest number of tests and follow-ups recorded.

The chart indicates a steady increase in both tests and follow-ups from 1998 to 2018, with a particularly pronounced increase from 2008-2013 to 2014-2017.
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

- Insurance
- Clinics
- Hospitals
- Courier Service
- Private Laboratory
- Stakeholder Groups
- Medicaid
- Regulations
- Demographic Entry
- Baby
- Specimen Acquisition
- Reporting
- Quality Assurance
- Clinical Care Coordination
- Customer Service
- Laboratory Testing
- Nurses
- Physicians
- Advisory Committee
- Ombudsman
- Information Technology
- Military
- Medical Consultants
- Policy Makers
- Education
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Goal: foster collaboration and support for NBS

Pathway: learning followed by consistent and proactive communication