Newborn Screening – It’s Complicated

Newborn Screening and Genetics in Public Health Committee

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NBS – It’s Complicated Panel

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Part 1… It seemed so simple

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Overview

Brief history

• National picture
  • 50 states… 2 territories… DC… Military…
  • Federal pressures

What’s unique about NBS?
In the Beginning…

1963 – Birth of newborn screening Phenylketonuria

- Simple bioassay to detect a single gene disorder
- Simple & quick diagnosis
- Window for treatment
- Effective & relatively inexpensive dietary treatment
- Cost effective
Then there was…

Congenital Hypothyroidism – late 70’s to early 80’s

• Simple assay
• Easy diagnosis
• Window for treatment
• Simple & effective treatment
• Extremely cost effective
  • “Average lifetime costs per person were estimated at $1,014,000 for persons with mental retardation”
  • Economic Costs Associated with Mental Retardation, Cerebral Palsy, Hearing Loss, and Vision Impairment --- United States, 2003. MMWR, 53(03);57-59

• Complications....
  • T4 or TSH? One screen or two?
Evolution of NBS Program

Goal: Identify babies with the condition prior to onset of symptoms

- Generally followed Wilson - Jungner criteria for screening*

1. The condition sought should be an important health problem.
2. There should be an accepted treatment for patients with recognized disease.
3. Facilities for diagnosis and treatment should be available.
4. There should be a recognizable latent or early symptomatic stage.
5. There should be a suitable test or examination.
6. The test should be acceptable to the population.
7. The natural history of the condition, including development from latent to declared disease, should be adequately understood.
8. There should be an agreed policy on whom to treat as patients.
9. The cost of case-finding should be economically balanced in relation to possible expenditure on medical care as a whole.
10. Case-finding should be a continuing process and not a “once and for all” project.

- Newborn Screening is an activity of the State

*Wilson & Jungner. WHO. 1968
National Perspective: States, Territories & Military

- If you’ve seen one Newborn Screening Program, you’ve seen one Newborn Screening Program.
  - Program structure
  - Laws and regulations
  - Policy-making process
  - NBS panel
    - Definitions
    - How do you count conditions?
  - Screening algorithm
    - Methods, analytes, methods, 2\textsuperscript{nd}/3\textsuperscript{rd} tier tests?
    - One screen or two?
  - Follow-up process
  - Fee structure
Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC)

• The Committee advises the Secretary of Health and Human Services on the most appropriate application of universal newborn screening tests, methodologies, policies, guidelines and standards.
  • Recommended Uniform Screening Panel
  • Medical foods
  • National Contingency Plan
  • Timely NBS Goals
  • Cut-offs/Risk Determination
Toward a Uniform Screening Panel and System

- Section I – A Uniform NBS Panel
- Section II – The System
  - Program evaluation
  - Timeliness
  - Cost-effectiveness analysis
  - Information gaps and a research agenda
- Future Needs
And so now we have....

Pompe Disease

• MS/MS
• Enzyme replacement therapy
• Expensive lifetime treatment
• Early infantile onset vs. late onset

Mucopolysaccaridosis Type I

• MS/MS
• Enzyme replacement therapy
• Treatment effectiveness?
X-linked Adrenoleukodystrophy
- MS/MS
- Males are affected. Female carriers?
- Lengthy diagnostic process
- Stem cell transplant

Spinal Muscular Atrophy
- Molecular-based assay
  - Long-term outcomes unknown
  - Very expensive
Federal/Public Pressure

Programs *must* meet ACHDNC recommendations and expectations of adding new disorders while overcoming challenges of keeping false negatives at zero (i.e., without “making mistakes”) as well as timely implementation and timely return of results.
NBS – it’s different from other public health testing

• Generally genetic
  • Concern is for the individual
  • Generally not screening for infectious disease (yet)

• Not many labs do it
  • State public health labs
  • Regional labs
  • Commercial entities
NBS – it’s different from other public health testing

• Steady volume – all babies born
  • Mandated – can opt out
  • Full population screen
  • Not seasonal
  • No outbreaks

• Time sensitive nature
  • STAT testing?
  • Most NBS programs have moved to at least 6 days/week operations
More Considerations

• Funding
  • Fees
  • Medicaid
  • General revenue

• Staffing considerations
  • Retention is critical
    • Increasingly difficult to train new staff
    • Breadth of conditions and knowledge needed
  • Follow-up is key – not just testing
    • NBS Program = Testing + Follow-Up
Staffing Considerations

• Easy to justify hiring lab staff and funding for reagents and consumables
• NBS requires a program approach with a follow-up component so must also justify
  • Educational efforts
  • Quality assurance
  • Support staff
  • Development
  • Follow-up staff
• Effective cooperation within the NBS system is critical
Be aware of what’s going on nationally. It will impact you.

Effective communication and cooperation throughout the NBS system is critical.
Any questions?

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