Newborn Screening Practice, Policy and Ethics: Parental Perspectives on the Issues

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Newborn Screening: What Is It?

- Biochemical testing for inherited disorders by analysis of the infant's blood
- Began in mid 1960s with Phenylketonuria
- Every state screens, each creates its own panel
- Informed consent is not the norm
- Today, four million infants screened annually
- Most widely-utilized form of genetic testing in the U.S.

How do States Decide What to Include on Panel?

No uniform system

Traditional criteria

- Screening must be beneficial to infant
- Condition must be relatively prevalent
- Effective treatment must exist
- Technology must be sufficiently accurate to identify disorder
- Substantial harm must be prevented by early identification
- Onset of symptoms must be early
- Facilities for diagnosis and treatment must be available

Recent Shift in Criteria

- ACMG report, and parent advocacy, as catalysts
- "Benefits to Family and Society"
  - Direct benefit to infant no longer necessary?
  - Other factors are now significant policy drivers, e.g.:
    - Cost concerns
    - parent advocacy positions
    - influence over future reproductive decision-making
    - research opportunities
    - interstate competition

Newborn Screening Today

Screening Programs are Growing Rapidly

- From 2004 – 2006, average number of conditions screened for rose from eight to thirty-three
- 90% of US babies tested for 21 of 29 ACMG-recommended condition as of 7/07, compared with just 38% in 2005 (WSJ, 10.30.07)
- One in 250 soon to have TRUE positive
- Much larger number will have false positives.

Policy, Ethics and NBS (a partial list)

- Access to treatment
- Informed consent
- Privacy/confidentiality
- False positives
- Identifying a-symptomatic individuals
- Avenues of policy influence

My Research

- Designed to expand the range of perspectives on NBS at play in public dialogues and policy debates
- Qualitative Interviews, focused on Cystic Fibrosis
- Prenatal, newborn screen, or post-symptoms diagnosis
- Some symptomatic, some not
- Grounded theory analysis, looking at data to surface useful categories
- Research on-going, focused on those with ambiguous diagnoses

How and Where Parents are Informed Matters

- Parents find it comforting to be face to face with a provider, and to be given information to take home
- Telephone and rushed explanations are objectionable

Timing of Diagnosis Matters

- We hear a lot about how important it is, with conditions such as the metabolic disorders, to receive an immediate diagnosis

- For families whose babies were healthy at the time of dx and beyond, there is a different message about timing. Many of these families mourn the loss of a more extended post-partum period in which to get to know their baby without a diagnosis.

Education/Information Before the Test Matters

- Parents who received a positive screen and then diagnosis without any knowledge of the test or the condition suffered inordinately at time of diagnosis
- Some parents also cautious about “too much information” at vulnerable time

Parents' Relationship with information is complex and variable

- Much data from focus groups, and from the Genetic Alliance study, suggests that more information is always wanted by parents, even if it may not improve health outcomes
- What I have heard from parents in my study is a more complicated picture

- Some parents want all available information immediately; they do not want to be "shielded" or "protected" by providers withholding information or meting it out slowly
- Other parents need a “one day at a time” approach right after diagnosis; they resent an avalanche of unsought information

Parents don't always opt – when testing is voluntary – to have their child tested right away

- My data here are small, but suggestive: 6 parents in study with 2nd child – 2 prenatal, 2 in states with automatic NBS, 2 opted NOT to test at birth
- We know that with other genetic tests, there's a substantial difference between predicted use of a test by the target population and its actual uptake. With NBS, because it's mandatory, it's complex to explore this issue.

Service System Must be Strong and Flexible

- Parents need guidance after a positive screen, and after a positive confirmatory test
- Health care professionals must have up-to-date knowledge, and strong inter-personal skills
- Parents do not always want more testing or procedures, particularly in situations with ambiguous findings and healthy babies

Policy Issues Emerging from Interviews

- Parental “voice” must be understood as broader than current advocacy voice
- Qualitative impact of NBS policy must be researched and considered in policy-making process
- Issues of education and consent must be revisited
- Ambiguous test results and variants of unknown significance are increasingly large challenges
- Consider feasibility of moving some tests to pediatric care settings?

Practice Issues (parents' perceptions)

- Educate about NBS
- Find ways to distinguish, in giving positive screening results, between situations where the baby's health is in immediate peril and situations where waiting a few days – say until a scheduled pediatric visit when results can be given in person, etc. – might be plausible
- Ask parents how much information they want at any given time
- Be certain there is a system of care -- INCLUDING GENETIC COUNSLEORS -- in place for parents getting positive screens and diagnoses. Use fees from NBS to build capacity in this area?