Decision Making in Newborn Screening

Ethics in newborn screening

Amy R.U.L. Calhoun, MD

Medical Director, Iowa Newborn Screening Program

Decision-making in newborn screening - foundations

- Wilson-Jungner criteria
  - The historical standard
    - Goal: treat people who need treatment without harming people who don't need treatment

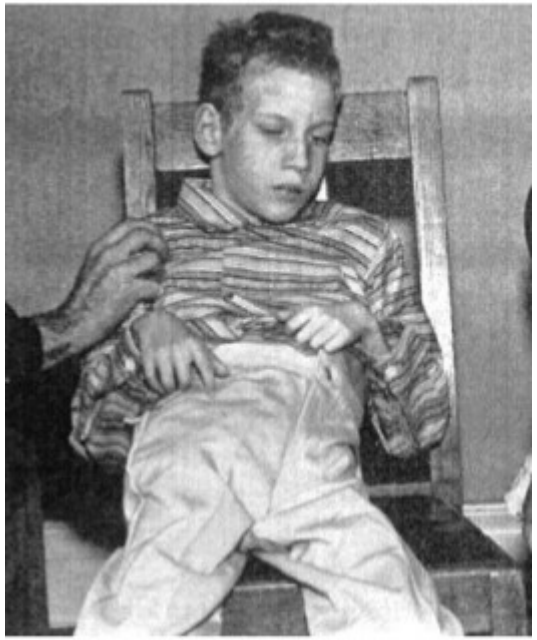
- Classic Wilson and Jungner screening criteria:
  - The condition should be an important health problem
  - There should be an accepted treatment for patients with recognized disease
  - Facilities for diagnosis and treatment should be available
  - There should be a recognizable latent or early symptomatic stage
  - There should be a suitable test or examination
  - The test should be acceptable to the population
  - The natural history of the condition, including development from latent to declared disease should be adequately understood
  - There should be an agreed policy on whom to treat as patients
  - The cost of case-finding (including diagnosis and treatment of patients diagnosed) should be economically balanced in relation to possible expenditure on medical care as a whole
  - Case-finding should be a continuing process and not a "once and for all" project

Foundations – Wilson-Jungner criteria in plain language

- The condition is important
  - Enough people have it; it's serious
- We understand the disease and can TREAT it
  - Treatment is needed BEFORE symptoms start
- We can tell if people actually have the disease
  - The test used to tell if people have the disease is doable
- Money/logistics/policy
  - Don't abandon patients after diagnosis
  - Let's not go bankrupt trying to do this

Decision-making in newborn screening - Foundations

- Example of a “good” target condition
  - The first newborn screening disorder: Phenylketonuria (PKU)
  - Normal at birth
  - Progressive severe intellectual disability
    - Cannot speak
    - Cannot walk
    - Self-injure, severe behavior problems
  - If identified at birth and treated with a special diet, they are normal
    - Attend college
    - Have their own families



Decision-making in newborn screening – a "good" newborn screening condition

- Why is PKU a "good" newborn screening condition?
  - Babies look completely normal at birth
  - Untreated people do VERY poorly
  - Treatment completely prevents problems
  - if we wait for symptoms, babies already have irreversible brain damage
  - Testing is accurate
  - Testing is cheap

“Bad” newborn screening condition?

- Real-life example from my practice in a different state:

- Baby looks completely well
- Newborn screen abnormal
- Many rounds of testing (5 months)
  - Final diagnosis: lethal degenerative disease with NO TREATMENT
- Child still looks entirely well
  - NOW WHAT?

- DID I HELP?

The Ethics – additional considerations

- If we can test for a disorder should we?
- Should we test for disorders with treatment that doesn't work well or is hard to get?
- Will this affect how parents treat their newborn?
  - Will quality of life be the same with this knowledge?
- Will adding the three new tests benefit the public as a whole?
  - What do a high number of false positives do to the validity of the program?

Ethics of treatment

- availability
  - To patients have to travel for treatment? How far?
  - What would the cost be to the family in time and resources?
  - what about families without insurance?
  - What about high-deductible insurance?
- Efficacy
  - Treatment should “work”
    - How sure do we need to be?
      - Most rare disease therapies have not been studied extensively
    - What does “work” mean?
      - Extend life?
      - Improve life?
      - Cure underlying disorder?

Ethics of testing

- Newborn screening is a government program
- Newborn screening is “opt out”
  - Children are automatically screened unless parents specifically decline
- Newborn screening is all or nothing
  - If you are tested, you are tested for everything on the panel
    - We can’t separate out parts of the testing
- “right not to know”
  - Once we have results, we have an obligation to act
- Substituted judgment
  - Parents decide for their child
- The value of finding a treatable condition before it causes problems is typically felt to balance the risks

Iowa's process

- The Iowa Congenital and Inherited Disorders Advisory Committee has a subcommittee – the "Management of the Iowa Newborn Screening Panel Subcommittee" – that is charged with reviewing every condition nominated for addition to the Iowa newborn screening panel.
- The subcommittee reviews current literature, recommendations from and the evidence-based review conducted by the Advisory Committee for Heritable Disorders in Newborns and Children (ACHDNC), the capacity of Iowa's newborn screening program to handle an expansion of the panel and screen for the condition (including enough lab space, need for extra equipment, enough staff, experts in the condition to care for the babies that screen positive for the condition,

CIDAC Management of NBS Panel Subcommittee

- The subcommittee reviews
- current literature
- recommendations from and the evidence-based review conducted by the Advisory Committee for Heritable Disorders in Newborns and Children (ACHDNC),
- the capacity of Iowa's newborn screening program to handle an expansion of the panel and screen for the condition (including enough lab space, need for extra equipment, enough staff with needed expertise, experts in the condition to care for the babies that screen positive for the condition, cost of screening, etc.)

- After the review is conducted, the subcommittee presents its findings to the whole Congenital and Inherited Disorders Advisory Committee
- After discussion, CIDAC members vote on whether to recommend the addition of the condition to Iowa's newborn screening panel.
- CIDAC's decision is sent to the Director of Iowa Department of Public Health
- IDPH staff then present the recommendation to the State Board of Health for final approval

- If the State Board of Health approves, then the Iowa Newborn Screening Program staff establish a pilot program for screening for the condition.
- The pilot gives the newborn screening program staff, IDPH administrators, birthing hospitals, information technology programmers, budget staff and all other stakeholders an opportunity to make sure everything is in place and is operating the way it should before official screening takes place.
- Any abnormal results for the condition discovered through the pilot will be reported to the baby's health care provider for follow-up.

- After a successful pilot, IDPH staff present the information to the State Board of Health again to finalize the newborn screening fee and other details in Iowa law (administrative code).