

The complex promise of newborn screening

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Outline

- Newborn screening
 - Mass intervention
 - In India
- Two challenges
 - Practical: realizing the benefits
 - Values-based: defining the benefits
- The need for better decision-making frameworks
 - The case of public health ethics

Newborn screening (NBS)

- Screening newborns to identify rare - primarily genetic - conditions
 - Began 1960s: phenylketonuria (PKU)
 - Growing recently: CF, IMDs, susceptibility
 - Technology (MSMS; DNA analysis)
 - Parent & professional advocacy
 - Commercial opportunity

Mass intervention

- Provided to populations - not upon request
- Provided to asymptomatic citizens - not patients
- Benefits and risks assessed in populations
 - “One size fits all” approach
 - But benefits and risks affect individuals

(Angus Dawson)

NBS in India

- Growing in ad hoc fashion
 - NGO: Spastic Society of Tamil Nadu operates NBS clinics for prevention
 - Government: Indian Council of Medical Research pilot projects in 5 cities, each screening 500,000 for free
 - Chandigarh funds mass genetic screening (PND and NBS) with reduced fee or free screening
 - Business: Dr Lal PathLabs, Delhi; NeoGen, Bangalore

NBS in India?

- Arguments against
 - Insufficient health system capacity
 - Low priority for infant health
- Arguments for
 - Epidemiologic transition
 - High burden of some genetic disease (e.g., SCD) and consanguinity
 - Demand of Indian middle classes - benefits of genetic medicine not reserved for West

Challenges of NBS: Practice

- NBS a total program: pre- and post-screening
 - To realize benefits, full system must be well designed and integrated
- Harms of NBS
 - Misunderstanding results (e.g., false +ves; carrier status) and impoverishment risk
 - Identification of misattributed paternity
 - Culture- & gender-specific impacts

Challenges of NBS: Principle

- What are the “ends” of NBS?
- Traditionally: clinical benefit for screened infant
- Increasingly: Wider benefits for family
 - Reproductive risk management
 - Knowledge: life planning; avoidance of “diagnostic odyssey”; research and experimental treatment

NBS: How do we decide?

- To date
 - Clinical calculus, values implicit
 - Debates over clinical benefits and harms; call for informed consent
- Need for values-explicit frameworks
 - Look to emerging frameworks identifying principles to justify public health interventions

Sum

- NBS offers complex promise
 - Challenges of practice
 - Challenges of principle
- Indian case throws this into sharp relief
 - Can illuminate issues in the international conversation about NBS

Thank you

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