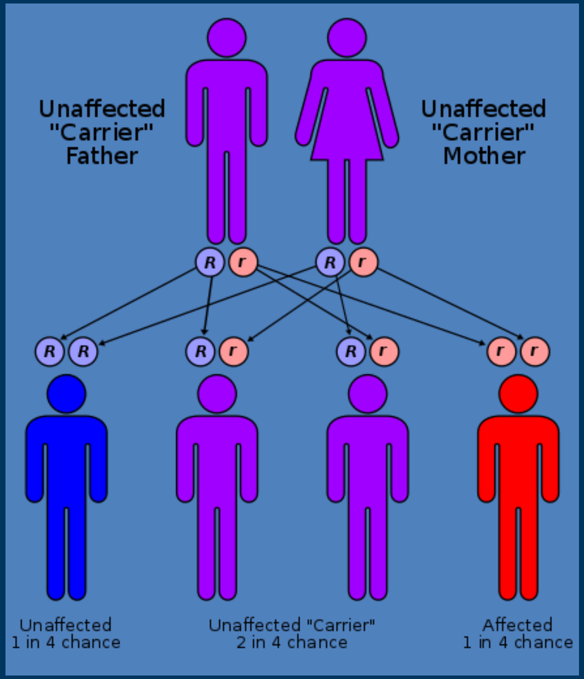

Ethical and social implications of revealing carrier status through Newborn Screening

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Heritable Disorders in Newborns and Children
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To return or not to return….

Mediated by:

Don't Return
Carrier
Results
Through NBS

Return All
Carrier
Results

probability and severity of health impacts

reproductive/family planning

treatability

patterns of inheritance

actual age of onset

Need to be condition specific

Primary Ethical Principles: A Place to Start

Parental Autonomy

Childs "Right to an Open Future"

Best Interests of the Child

Social Implications

- Misunderstandings of carrier status
  - Sickle Cell screening in the 1970's
- Potential for stigmatization and impact on family dynamics
- Impact on self esteem or self image
- Potential for Discrimination
  - Limits of GINA
- Other familial/psychosocial impacts
  - When to reveal?

Potential Health Benefits in Childhood

To return…

- Benefit of early detection
- **Health Benefits**
  - **Screening**
  - **Interventions**

Or not to return…

- Potential harms of misunderstanding status
  - Ex. Sickle Cell in the 70's
- Potential Discrimination or Stigmatization of Carriers
- Unnecessary screening
- Potential Anxiety/Worry

Potential Health Benefits in Adulthood

To return…

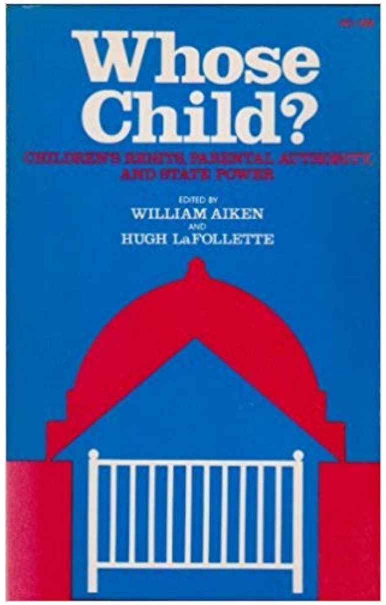
- Increased awareness of risk
- **Health Benefits**
  - **Screening**
  - **Interventions**

Or not to return…

- Potential harms of misunderstanding status
  - Ex. Sickle Cell in the 70's
- Potential Discrimination or Stigmatization of Carriers
- Unnecessary screening
- Potential Anxiety/Worry

Right to an Open Future

- Discussed by Joel Fieberg in 1980
- Dena Davis applied to genetic testing (1997)
- “rights-in-trust” to be “saved for the child until they are an adult”
- Focused on autonomous decision making of the child when they reach adulthood



Other considerations for “open future”

- Most organizational policies (ACMG, AAP) discourage returning carrier statues w/o health benefits to children
- Promotes choice as adults

- Will adults get screened w/o family Hx or group membership?
- “Universal Carrier Screening” panels include CF, Pompe, MPS1, X-ALD , Fragile X, DMD, SMA
- Equity considerations…who has to Expanded Screening or genetic services and counseling?

Expanded Concepts of NBS Benefit: Reproductive Decision Making

To return…

- Reproductive benefits to parents or other family members
- Reproductive benefits to newborns
- Concepts of expanded benefits in NBS (if **only** reproductive benefits)

Or not to return…

- Social/Familial Implications
  - Potential harms of misunderstanding status
  - Potential Discrimination or Stigmatization of Carriers
  - Potential Anxiety/Guilt
- *Moves us away from the ethical justification for NBS?*

Parental Autonomy/Rights

Right not to know

- Can programs force parents to know carrier results?
- Paternalism and Public Health
- Does this fit the ethical and legal justification for mandatory screening?

Right to know

- Autonomy of parents
- Personal Utility vs. Clinical Utility: Who gets to decide?
- ***Would consent solve both of these issues?***

To return or not to return….

Possible Middle Roads:

Don't Return
Carrier
Status
Through NBS

- ***Only screen targeted groups***
- ***Implement consent process for carrier status***
- ***Put carrier status in medical record to be revealed later***

Need for condition specific policies?

Return All
Carrier
Status

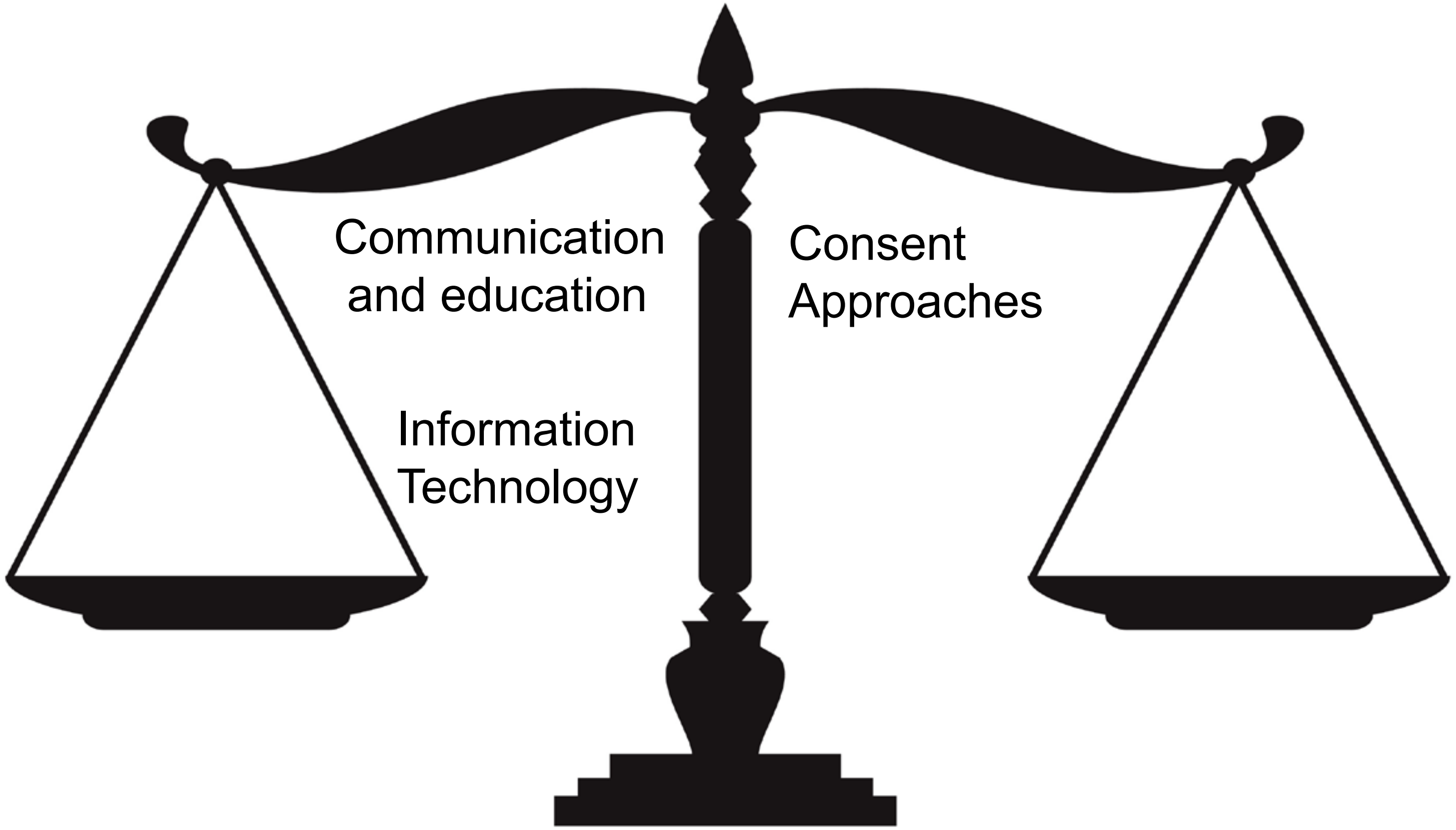
A related programmatic question…

- To detect or not to detect!
- Is it ethical to filter out carrier status? Is it possible?

Conflicts with Professional Ethics?

- “We need to be very clear about like the definition of an actionable result…we would need some guidelines about ‘What are actionable results…So to understand that just because we can do the test, doesn’t mean we’re prepared to deal with the results, and maybe we shouldn’t, as Public Health systems.”

- “Ethically, I think most programs feel that they need to report what they find, and as a labratorian, you report what you find. To window something out means to me that you may be missing something that might be a very key piece of information for a family. And how do you live with that.”



Communication and education

Information Technology

Consent Approaches

Need for more ELSI research in NBS pilot studies. Upcoming NBSTRN paper!

Thank You!

- HRSA Child and Maternal Health Bureau (R40MC268050102)
- Michele Puryear, Jeff Brosco, and Mike Watson
- The NBSTRN Bioethics and Legal Workgroup
- Beth Tarini and Amy Gaviglio