Ethical and social implications of revealing carrier status through Newborn Screening

Aaron Goldenberg, PhD MPH
Department of Bioethics
Center for Genetic Research
Ethics and Law

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To return or not to return....

Mediated by:

Don't Return Carrier Results Through NBS

probability and severity of health impacts
reproductive/family planning
treatability
patterns of inheritance
actual age of onset

Need to be condition specific

Return All Carrier Results
### 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*

Return All Carrier Status

Need for condition specific policies?
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.”
Need for more ELSI research in NBS pilot studies. Upcoming NBSTRN paper!
Thank You!

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