Year 2, Lesson 3: Ethics of Genetic Testing in Infants and Children

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Learning Objectives
1. Describe four basic ethical controversies surrounding genetic testing of pediatric patients.
2. Define the best interest standard and demonstrate how it can be applied to genetic testing of pediatric patients.
3. Compare and contrast predictive testing for carrier states with predictive testing for actual phenotypic disease.
4. List three characteristics that make a disease a good candidate for newborn screening.
5. Identify ways that expansion of newborn screening programs may lead to ethical controversy.

Goals
For this lesson, the goal is explore the ethics of genetic screening and testing and newborns, as well as more recent advances in the field and implications for newborns and their families. For this session, it may be helpful to invite a practicing pediatric geneticist or prenatal genetics counselor to co-facilitate the live session.

Suggested Reading

Additional Reading
Case Summary
Baby X is born full-term following no prenatal complications. She does well in the newborn nursery and is discharged home with her mother. Her newborn screen returns a week later with positive findings for Krabbe disease, a rare and devastating neurological disorder. Her parents take her to a pediatric neurologist, who explains to them that the infant’s DNA test indicated that she is most likely a carrier for the disease, but further testing is required.

Although there is one genetic mutation strongly associated with early infantile onset of Krabbe, the follow-up testing on Baby X provides only ambiguous results. The neurologist tells the parents that it is possible their child may develop Krabbe at some point in the future, but cannot say for certain. He also cannot predict when it may happen, if there is any way to prevent it or if there will be viable treatment options.

Questions for Discussion
1. What, if any, are the ethical concerns regarding the newborn screening program and genetic testing in general?
2. Should disorders with a variety of clinical presentations, some of which may not have successful treatments, be included in the screening program in order to detect those few cases with the most severe and potentially treatable form?
3. Should parents be able to request genetic testing to determine if their child is a carrier for a condition or will have an adult-onset condition?

Affirmations & Tips
Inviting an additional faculty member with expertise in clinical genetics present to help co-facilitate the lesson will be helpful, such as a pediatric geneticist or prenatal genetics counselor.

The following activity can be an effective white-board exercise. Ask your learners:
1. What makes a good screening test for newborns and why? Identify a volunteer to compile answers into a list, then discuss.
2. If approached, would you give parental permission for genetic sequencing (BabySeq) for your newborn child? Or recommend that a child of a loved one give permission? Why or why not? What are the potential benefits and harms?

Learner-Generated Case Discussion. One of the most effective activities has been to ask learners for cases they have recently encountered in clinical training/practice. Ensuing discussions are typically lively given immediate relevance for learners.