

1. Purpose

To examine the factors that affect parental attitudes toward voluntary newborn screening for genetic disorders.



Photo courtesy of the Museum of Disability History.

Blood samples for genetic screening are collected from a heel prick before the newborn is discharged from the hospital.

2. Background

Little information is available on parental attitudes toward newborn screening:

- A recent review found only 28 articles that examined newborn screening, compared with 78 on antenatal screening.
- Fewer studies have examined the attitudes of parents in the general population; almost all examined attitudes and reactions of parents of affected children or children with false positive results.

3. Methods

- We searched PubMed using the key words "newborn screening" in combination with the following terms: attitudes, preferences, or opinions. We identified 48 articles, 9 of which reported data on parental attitudes toward newborn screening. (A list of articles can be obtained from the first author.)
- The study populations, timing of assessment, and type of measures varied across studies, and sample sizes were generally small. Thus, we did not attempt to generalize study results, but merely summarized their findings.

4. Attitudes and Affects of Newborn Screening for Specific Conditions

Cystic Fibrosis

- Most parents favored newborn screening regardless of their child's test result.
- Those who favored screening believed early diagnosis would lead to better care and outcomes.
- Additional reasons for support among parents of affected children were as follows:
 - Prevention of misdiagnosis, particularly those that blamed parents
 - Prevention of delayed diagnosis
- Delays in final diagnosis after screening test eroded support for screening.
- Reproductive planning did not often provide reason for favoring screening.

Duchene Muscular Dystrophy

- Parents showed mixed support for screening.
- There was almost universal agreement that screening should be voluntary.
- Reasons for support were as follows:
 - Information for practical and emotional preparation
 - Unwillingness to believe that nothing could be done
 - Earlier physiotherapy
 - Reproductive planning
 - Avoidance of diagnostic delays;
 - Avoidance of misunderstood symptoms
- Reasons against support were as follows:
 - Information provided was insufficient
 - Regret for early diagnosis

Fragile X

- Families of affected child strongly supported voluntary testing for carriers, newborns, and at earliest concern.
- Most thought best time for testing was before a woman became pregnant.
- Outcomes parents considered somewhat or very likely were as follows:
 - Strained relations with family members
 - Discrimination by insurance companies
 - Increased parental stress
 - Earlier and additional service availability
 - Informed reproductive planning
- Increased understanding of child's problems

Phenylketonuria

- Parents overwhelmingly supported screening.
- Some were concerned about future use of blood samples for research.
- Most supported mandatory testing.
- Reasons for supporting voluntary testing were as follows:
 - Religion
 - Parents as decision makers
- All agreed that more information was needed on screening.

Multiple Metabolic Disorders

- Half of parents of affected children rated knowledge of screening as inadequate.

5. Findings

General Attitudes on and Knowledge of Newborn Screening

- Generally, parents were not knowledgeable about newborn screening and were often unaware that the testing had been performed.
- A review of studies on decision making regarding antenatal genetic testing found that parents who chose to have testing and those who did not considered the same factors but gave them different weight.
- A study of parents of various ages and backgrounds found that
 - Most wanted access to genetic testing and believed that parents should be the final decision makers.
 - Most believed it was important to share genetic information.
 - Most parents, regardless of race, considered confidentiality important, but black parents were more willing to accept disclosure in some cases than white parents.

6. Discussion

- The literature on parental attitudes toward newborn screening is scant and limited by small sample sizes and inconsistent methodologies (but does provide some preliminary suggestions).
- Parents generally have favorable attitudes toward screening, even for untreatable conditions.
- Parents may have unrealistic expectations of the ability of early diagnosis to improve outcomes, even when told a condition is untreatable.
- Good information is critical.
 - Parents need to receive information about the screening when they have time to digest it.
 - A well-designed procedure for conveying test results reduces unwarranted stress and anxiety.