

A review of the psychosocial effects of false-positive results on parents and current communication practices in newborn screening

J. Hewlett · S. E. Waisbren

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Summary As more states adopt expanded newborn screening for metabolic disorders, the overall number of false positives increases. False-positive screening results have been associated with increased anxiety and stress in parents of infants who require follow-up testing, even after the infant's good health is confirmed. This article reviews the literature on the negative impact of false-positive newborn screening results on parents, along with a review of current communication practices for follow-up screening. The results of this review suggest that parental stress and anxiety can be reduced with improved education and communication to parents, specifically at the time of follow-up screening. Communication strategies with sample materials are proposed.

Abbreviations

CES-D	Center for Epidemiologic Studies Depression Scales
MAACL	Mean Multiple Affect Adjective Checklist
MS/MS	tandem mass spectrometry
NBS	newborn screening
P-CDI	Parent–Child Dysfunction Interaction
PCP	primary care provider
PSI	Parenting Stress Index

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J. Hewlett (✉)
Division of Genetics, Children's Hospital Boston, Massachusetts, USA
e-mail: jenny.hewlett@chidrens.harvard.edu

S. E. Waisbren
Department of Psychiatry, Division of Psychology, Children's Hospital Boston, and Harvard Medical School, Massachusetts, USA

Introduction

Expanded newborn screening using tandem mass spectrometry (MS/MS) to identify biochemical genetic disorders is an important advance in early disease detection. MS/MS requires no additional blood samples beyond the standard procedure used for 30 years. It has greater sensitivity than past screening methods and allows for presymptomatic detection and identification of metabolic disorders (Levy and Albers 2000). This technology has the potential to reduce infant morbidity and mortality by enabling earlier diagnosis and treatment.

However, a downside of expanded newborn screening with MS/MS is that, as the number of disorders screened for increases, the overall number of out-of-range results also increases, of which the majority are confirmed to be false positives after further testing. For those who receive expanded newborn screening by MS/MS (more than 20 disorders), the prevalence of true screening positives is approximately one out of every 2400 infants screened (0.04%) (Schulze et al 2003). However, approximately one of every 300 infants screened (0.33%) will receive a false-positive designation, which is an out-of-range screening result that, after further follow-up testing, is not shown to indicate a metabolic disorder (Schulze et al 2003). The overall number of false positives in the USA has increased to roughly 8 false positives for every true positive identified and it is estimated that approximately 13 000 false-positive newborn screening results are received each year by parents of newborns (Center for Disease Control and Prevention 2003).

When an out-of-range test result occurs, the state laboratory usually notifies the primary care provider (PCP) listed on the newborn screening card, who is then responsible for contacting the parents of the infant and scheduling confirmatory testing. In New England, the majority of parents are

informed of their child's out-of-range newborn screening results by their paediatrician (54%) and/or the paediatrician's nurse practitioner (28%) (Waisbren et al 2003).

Although there has been recognition of the problem of false-positive results in newborn screening and subsequent long-term stress in parents for over 35 years (Rothenberg and Sills 1968), little if anything has changed to improve this situation. With 37 states currently offering expanded newborn screening (National Newborn Screening and Genetics Resource Center (NNSGRC): National Newborn Screening Status Report; <http://genes-r-us.uthscsa.edu/nbsdisorders.pdf> (accessed 01/03/2006), better communication and education is now more necessary than ever for informing families of patients with out-of-range results when a repeat screen is requested (Kwon and Farrell 2000).

This paper reviews studies on parental response to newborn screening and current newborn screening communication practices in the USA. The aim is to use this evidence to provide recommendations for reducing parental stress in response to newborn screening, especially that due to false-positive identifications.

Methods

A computerized search was conducted for this literature review using MEDLINE (via PubMed) and Journals@OVID. References from existing articles were also perused to locate relevant studies. Key search terms included: *newborn screening*, OR *neonatal screening*, AND *stress*, OR *anxiety*, OR *communication*, OR *education*, OR *false positive*, OR *PKU*, OR *metabolic disorder*.

Nine published studies spanning 23 years on the topic of parental stress after a child received a positive newborn screening result (later confirmed to be a false positive) were reviewed. Articles on newborn screening for metabolic disorders as well as for cystic fibrosis, congenital hypothyroidism and newborn hearing testing were included. The Parenting Stress Index (PSI), used in three studies, is a screening and diagnosis assessment instrument that uses a 5-point Likert scale to measure the magnitude of stress in the parent and the parent-child relationship to identify those at risk for dysfunctional parenting. The Parent-Child Dysfunction Interaction (P-CDI) subscale from the PSI is used to measure rates of parent-child dysfunction (Abidin 1995). One study used the Mean Multiple Affect Adjective Checklist (MAACL) (Sorenson et al 1984) and another used the Center for Epidemiologic Studies Depression Scales (CES-D) (Tluczek et al 2005). Percentage of difference was calculated in four studies (Baroni et al 1997; Gurian et al 2006; Stuart et al 2000; Tluczek 2005) to demonstrate the difference between the reported score for those who received a false-positive newborn screening result and those parents who received

normal screening results. In addition, percentage of increase was calculated from published norms in one study (Sorenson et al 1984).

Results

Table 1 presents nine studies, eight of which report that an abnormal newborn screening result requiring retesting is associated with parental anxiety and/or depression, even when the repeat test is normal. Of the studies including a comparison group (Baroni 1997; Gurian et al 2006; Stuart et al 2000; Tluczek et al 1991, 2005), one found a 23% increase in self-reported maternal stress (Gurian et al 2006), one found a 6% increase in maternal stress (not significant but in the same direction as the other studies) (Stuart et al 2000) and two found an increase in parental depression (146% and 77%) (Tluczek et al 1991, 2005). One study found lower stress scores for parents who received false-positive results than those who received normal results but also reported a 32% lower (23.8 vs 35.2) score on a defensive responding scale, associated with difficulty in responding honestly regarding personal stress (Baroni 1997). The four studies that did not include a control group reported anxiety rates of 14% to 78% among their samples (Bodegard et al 1983; Clemens et al 2000, Fyro and Bodegard 1987; Sorenson et al 1984). In addition to anxiety, parent-child dysfunction has been noted. Mothers whose infants were in the false-positive group had a 41% higher P-CDI score and fathers had a 24% higher P-CDI score than those who received normal results (Gurian et al 2006). Four studies also reported long-term negative effects including alterations in perceptions of their infant's health, an increase in the number of emergency room visits, and hospitalizations for the infant (Bodegard et al 1983; Fyro and Bodegard 1987; Gurian et al 2006; Sorenson et al 1984).

Several studies have established a link between stress and poor parental knowledge of newborn screening. In one study, 36% of parents of infants who received a false-positive result reported concern about the health of their infant even after learning of the normal results of the repeat test. However, this concern was greater in parents reporting that they had not received adequate information about the screening process and its significance for their infant's health (Sorenson et al 1984). In another study, among 166 mothers whose infants received false-positive results for metabolic disorders, 35% knew the correct reason for the repeat screen of their infant. Eleven per cent reported not being given a reason for the repeat screen. Mothers who knew the correct reason for repeat screening reported lower total stress compared to those who did not (69.1 vs 75.3, $0 = 0.005$) (Gurian et al 2006).

Poor parental knowledge may stem from a lack of familiarity with newborn screening among healthcare providers as well. In the USA, all newborn screening programmes

Table 1 Parental stress and psychosocial effects of follow-up screening

Date	Authors	Screening	Sample/Controls	Method/Instrument	Results
2006	Gurian et al	Metabolic disorders	173/67	Phone interviews short form PSI (Abidin 1995)	23% increase in stress levels for mothers 10% increase in stress levels for fathers 11% of mothers' stress levels in clinical range as compared to 0% of mothers in control group
2005	Tluczek et al	Cystic fibrosis	51/35	Interviews CES-D	146% increase in depression levels
2000	Stuart et al	Cystic fibrosis	20/20	PSI (Abidin 1995)	6% increase in stress levels for mothers (but not interpreted as a significant difference between the two groups)
2000	Clemens et al	Hearing	49/–	Phone survey	14% of mothers reported lasting anxiety
1997	Baroni et al	Cystic fibrosis	14/14	PSI (Abidin 1995)	32% decrease in defensiveness score among parents with false-positives 9% decrease in stress among parents with false-positives [Note: Lower defensiveness scores (interpreted as hypervigilance) are reflective of a difficulty in responding honestly regarding personal stress]
1991	Tluczek et al	Cystic fibrosis	104/18	Survey	98% of parents reported anxiety 77% experienced depression
1987	Fyro et al	Cystic fibrosis	32/–	Evaluation of in-person interviews	50% of families reported persistent anxiety
1984	Sorenson et al	PKU	60/–	MAACL (Zuckerman and Rubin 1965)	33–48% higher rates of depression than established mean norm but not above the mean norm cut-off for anxiety
1983	Bodegard et al	Cystic fibrosis	102/–	Evaluation of phone interviews	76% of families reported initial anxiety 18% reported persistent anxiety afterwards

except one report providing specific education efforts for PCPs about newborn screening (50 states), through mailings (31 states), provider practice manuals (16 states), Continuing Medical Education (12 states), or information posted on their newborn screening website (15 states) (Kemper et al 2005). Nonetheless, in a recent study of paediatricians in Massachusetts, 14% did not know about expanded newborn screening for metabolic disorders and 42% were less than comfortable talking about newborn screening test results with families (Gennaccaro et al 2005). An assessment of the communication process for cystic fibrosis newborn screening showed almost 14% of parents found to be carriers of cystic fibrosis (owing to false-positive newborn screening results) believed that paediatricians and family physicians needed to be better informed about the implications of a positive screen test so that they could more effectively explain possible consequences to parents. More than half (54.5%) of parents wished more information was provided at the time of the first positive newborn screen test, before retesting and final diagnosis (Ciske et al 2001). More than half of mothers who received a false-positive result for initial hearing screening believed that the purpose of the test and the results were not adequately explained (Clemens et al 2000).

Parents favour being told about the need for retesting in person during an appointment with the physician. Informing parents by phone, leaving a message on an answering machine, and informing parents of the screening results without having the time to discuss the related issues in detail have been associated with more distress in the parent and misinformation about the test results (Tluczek et al 1991). Mothers who received the repeat screening results in person were less stressed (PSI median score of 55) than mothers who received results by other means (PSI median score of 67) ($z = 2.45, p = 0.02$) (Waisbren et al 2003). Physicians seem to be able to reduce parents' stress if they provide information about the process (as well as the results) of newborn screening, estimate the risk to the infant as low, or refer parents for additional information (Sorenson et al 1984; Tluczek et al 1991).

Cognizant of the links between communication, knowledge and stress, the American Academy of Paediatrics (AAP) Task Force on Newborn Screening developed recommendations for what should be covered in information and educational materials about newborn screening for parents (AAP 2000). These include (a) the benefits and potential risks of screening; (b) how parents will receive screening results; (c) the possibility of a false-positive test result; (d) the importance of responding to a positive test result; (e) how to respond to a positive test result; and (f) the screening programme's policy for sample storage and use of stored samples (AAP 2000). The task force also highlighted the importance of developing materials that are written at an appropriate literacy level and that reflect cultural competency (Fant et al 2005).

Despite public health efforts, a national survey of state newborn screening programmes (Fant et al 2005) revealed that none of the state newborn screening materials included all the recommended elements for screening information for parents outlined in the AAP Task Force on NBS and therefore did not meet recommended AAP standards. While 98% of the materials covered the benefits of screening and 87% covered how parents will be informed of results, only 34% covered the importance of responding to a positive result, 28% covered how to respond to a positive result, 19% covered the risks of screening, 13% covered the possibility of a false-positive result, and only 11% covered storage/use of stored samples. The median readability level of the newborn screening materials (calculated using the SMOG Readability Formula utilized by the US Department of Health and Human Services) (McLaughlin 1969) was grade 10, whereas the National Work Group on Literacy recommends that materials be written at or below sixth-grade level (Fant et al 2005).

Newborn-screening programmes routinely provide educational materials to hospitals, midwives, paediatricians and PCPs who are intended to pass them on to parents. However, state newborn screening programmes are unable to say when, or whether, parents actually receive the newborn screening information (Kemper et al 2005; US General Accounting Office 2003).

Discussion

Our review confirms earlier results on stress in parents following newborn screening. 'PKU Anxiety Syndrome' was first coined 38 years ago to describe the common stress reaction in parents who received a false-positive PKU test result for their infants (Rothenberg and Sills 1968). This syndrome was characterized as acute or chronic anxiety in parents due to uncertainties about test results and worrying about their child's health. Anxiety is a normal reaction to a perceived threat to an infant's health; however, this anxiety is sometimes not resolved once a normal follow-up test is obtained and the child's good health is confirmed (Rothenberg and Sills 1968).

The PSI scores shown in Table 1 reflect the occurrence of unnecessary concern among parents who received a false-positive newborn screening test result. One study shows that the number of hospitalizations was almost double for childhood symptoms unrelated to metabolic disorders among infants who received false-positive results for newborn screening (Gurian et al 2006). Furthermore, exposure to maternal stress during infancy has been reported to increase that infant's sensitivity to subsequent stress exposure (Essex et al 2002). In a follow-up study of infants screened for congenital hypothyroidism, half of all children examined showed disturbed behaviour 4 years after their parents received

false-positive newborn screening results (Fyro and Bodegard 1987).

The initial shock of receiving an abnormal screening result and the ensuing anxiety may be due in part to the timing of the repeat screening (Fyro and Bodegard 1987; Tluczek et al 2005). In the first few months after a baby's birth, parents are often experiencing sleep deprivation, are adjusting to having a new family member, and may be dealing with insecurities about their parenting abilities (Tluczek et al 2005). Negative effects from false-positive screening may also stem from inadequate information and understanding about both newborn screening and follow-up testing of infants with initial out-of-range screening results. After receiving out-of-range results (especially if provided with ambiguous or incomplete information), parents often try to find more information from the Internet, relatives or friends, all sources of information with potential inaccuracies (AAP 2000; Tluczek et al 1991, 2005). Improved communication and education regarding follow-up testing at this critical point may reduce the negative consequences that have been cited as reasons to limit expanded newborn screening (Thomason et al 1998).

Recommended communication strategy

In addition to recommendations stemming from a review of newborn screening studies and communication practices, reports from scholars, professional associations and policy makers have suggested broad strategies for improving communication and education for follow-up to newborn screening (AAP 2000; Green et al 2004; Hiller and Landenburger 1997; Kim et al 2003). The following recommendations are proposed based on the evidence outlined in the literature review above and input from these reports.

(1) Involve newborn screening stakeholders in newborn screening programmes within their state

- Include parents, PCPs and paediatricians on newborn screening programme advisory boards and committees and in the planning of newborn screening educational programmes.
- Pre-test newborn screening key messaging and educational materials with parents and PCPs through focus groups and/or on-line surveys.
- Re-evaluate educational materials with stakeholders on an on-going basis.

(2) Provide materials that will better prepare paediatricians to effectively communicate with parents in regards to follow-up screening

(a) A one-page memo for physicians highlighting the research on parental stress and false positives (Table 1) and the need

for improved communication, and recommending the use of targeted materials (see below) when informing parents of the need for repeat screening.

This memo should touch on the importance of telling parents about the need for retesting in person and supplying additional reliable sources of information to parents if necessary (Sorenson et al 1984; Tluczek et al 1991).

(b) A follow-up informational brochure for physicians to distribute to parents. Current examples include:

- 'Newborn Screening and Services. A guide to test results, programs, and follow-up for your baby', a brochure published by the Rhode Island Department of Health (2005)
- 'These Tests Could Save Your Baby's Life: Newborn Screening Tests', a pamphlet published by The American College of Obstetricians and Gynecologists (2006)
- 'Follow-up to Newborn Screening: A Guide for Parents', a pamphlet developed at Children's Hospital Boston (2005a).

These educational materials recognize the importance of addressing follow-up screening and were all tested through focus groups and/or pilot tested with stakeholders. The readability levels of these materials are still a little higher than recommended (ranging from grades 9 to 11); however, they cover all but one or two of the recommendations for newborn screening materials set out by the American Academy of Pediatrics (AAP 2000).

Ideally, the parent brochure should be written at a lower readability level and translated into additional languages as necessary to meet local demographic requirements.

(c) A Frequently Asked Question (FAQ) Sheet outlining questions parents ask about newborn screening follow-up intended to assist pediatricians with messaging. (Children's Hospital Boston 2005b)

It is recommended that materials be distributed to paediatricians through mail, e-mail, postings on relevant websites, professional associations, and/or continuing medical education (Gennaccaro et al 2005).

(3) Conduct evaluative research to determine what communication methods are most effective

Such research should include:

- Focus groups and pilot testing of consumer and professional materials
- Specific research about what strategies are effective during the informing process and for counselling parents about newborn screening results
- Training PCPs to use recommended methods and materials

- Follow-up studies of parental stress and repeat screening to track potential improvements

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