Parental Attitudes and Ideas Regarding Newborn Screening for Familial Hypercholesterolemia

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**METHODS**

**BACKGROUND**

- Familial hypercholesterolemia (FH) is an inherited autosomal dominant disorder of low-density lipoprotein cholesterol (LDL-C) metabolism resulting in an earlier onset of atherosclerotic cardiovascular disease\(^{1,2}\).
- Current recommendations state universal lipid screening ages 9-11 and 17-21, however screening rates are low (<5%)\(^{3,4}\).
- One suggestion to improve detection is to include FH on the newborn screen (NBS).
- The purpose of this study was to explore parents’ perceptions about the benefits and drawbacks of NBS for FH to improve the health of their children.

**METHODS**

**Data Collection**
- Recruitment took place at the institutional pediatric lipid clinic and the cystic fibrosis clinic
- Interviews conducted by telephone in 30-minute sessions guided by a semi-structured interview script. Participants were given a brief overview of the NBS process and FH.
- All interviews were audio-recorded and professionally transcribed.

**Analysis**
- Thematic analysis performed in several steps using qualitative data analysis software (NVivo 12 Pro).
- Transcripts reviewed to identify key themes
- Open coding was performed line by line by two reviewers. Codes were compared and discrepancies resolved by arbitration
- Final codebook created and used to review the transcripts. Data were categorized and organized into overarching themes.

**RESULTS**

**Supporting Screening**
- Knowledge
- Family
- Incorporation of lifestyle changes
- Access to preventative care
- Timing

**Negatives of Screening**
- Increased stress or anxiety
- Knowledge
- Stigma
- Delayed from diagnosis to initiation of pharmaotherapy for FH

**CONCLUSIONS**

- All 11 participants (100%) indicated that they would be interested in including screening for FH on the NBS.
- Efforts to educate clinicians on the natural history of FH and treatment options available should be increased and guidelines should be readily available.
- The results of this study will be used to help formulate family notification and care protocols for newborns diagnosed with FH.

**ADDITIONAL KEY INFORMATION**

**Resources**

**Acknowledgments**
We would like to thank the following individuals for their assistance in recruitment:
- Ann Dodge, NP
- Darci Pfeil, NP
- Michael J. Rock, MD

Supported by a Research and Development grant from the Department of Pediatrics, University of Wisconsin School of Medicine and Public Health

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