Parental Attitudes and Ideas Regarding Newborn Screening for Familial Hypercholesterolemia

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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

Participants

- 8 parents of a child with FH
- 3 parents of a child who was diagnosed with a genetic condition through NBS
- 100% white female
- Age: M = 44.2 yrs. (SD = 7.7)
- Health Literacy*: M = 15.0 (SD = 1.2)

*Health literacy is measured by a composite score ranging from 0-16, with a higher score representing better health literacy

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.

Biggest Factor in the Decision to have Newborn Screened for FH via the NBS

Biggest Factor	Number of Mentions (n=11)
Family	2
Lifestyle Changes	3
Preventative Care	3
Timing	5

All participants (n=11,100%) were interested in having their child undergo universal lipid screening via the NBS

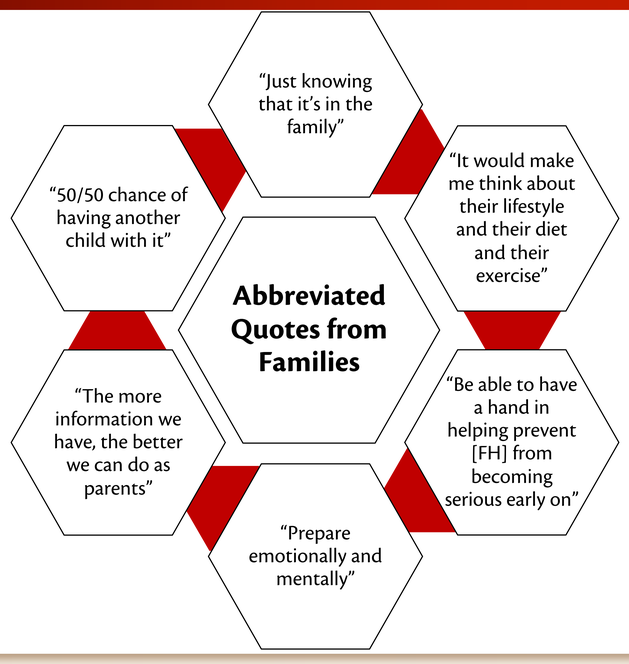
RESULTS

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 pharmacotherapy 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

1. Ned, et al. PLoS Curr 2011.
2. Bowen et al. Public Health Genomics 2012.
3. Allen-Tice et al. J Clin Lipidol 2020.
4. DeSantes et al. J Pediatr 2017.

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