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Treatment decision making for adolescents with familial hypercholesterolemia: Role of family history and past experiences

Thomas I. Mackie, PhD, MPH, MA*,

Institute for Clinical Research and Health Policy Studies, Tufts Medical Center, Boston, MA, USA

Lisa L. Tse, BA,

Institute for Clinical Research and Health Policy Studies, Tufts Medical Center, Boston, MA, USA

Sarah D. de Ferranti, MD, MPH,

Department of Cardiology, Boston Children's Hospital Harvard Medical School, Boston, MA, USA

Heather R. Ryan, BA, BS, RN, and

Department of Cardiology, Boston Children's Hospital, Boston, MA, USA

Laurel K. Leslie, MD, MPH

Institute for Clinical Research and Health Policy Studies, Tufts Medical Center, Tufts Clinical and Translational Science Institute, Tufts University School of Medicine, Boston, MA, USA

Abstract

BACKGROUND: Adolescents and young adults (AYAs) with familial hypercholesterolemia (FH) are at high risk for underdiagnosis and inadequate treatment. Yet, little is known about the factors that influence the medical decision making of AYAs with FH and their families.

OBJECTIVE: This study explores how family medical history, family narratives of medical experiences, and AYA medical experiences together function as “experiential evidence” and influence screening and treatment decisions.

METHODS: Twenty-four parents and AYAs affected by FH from a pediatric preventive cardiology practice responded to a survey and a semistructured qualitative interview. Transcribed interviews were analyzed using a modified grounded theory approach. Study design, instruments, and interpretation of results were informed by a 20-member stakeholder panel.

RESULTS: AYAs and parents reported extensive personal and family experiences with cholesterol and cardiovascular conditions and treatments, sometimes distinct from FH, which were used as evidence to inform their own perceptions of FH risk and treatment. This experiential evidence impacted perceptions of: (1) hereditary risk for FH diagnoses, (2) risk for future cardiovascular disease, (3) risks associated with treatments, and (4) capacity to comply with recommended treatments. Although experiences of family members initially informed screening

*Corresponding author. Tufts Medical Center, Institute for Clinical Research and Health Policy Studies, 800 Washington Street, MC 345, Boston, MA 02111, USA. tmackie@tuftsmedicalcenter.org.

and treatment decisions, the subsequent personal experiences of AYAs led to new experiential evidence that informed future decisions.

CONCLUSIONS: Family cardiovascular history related to and distinct from FH influenced screening and treatment decisions of AYAs and parents affected by FH. Additional clinical assessment of personal and family medical experiences may enhance understanding of the decision-making processes among AYAs and ultimately improve adherence to screening and treatment recommendations.

Keywords

Cholesterol; Familial hypercholesterolemia; Adolescent; Experiential evidence; Decision making

Introduction

An estimated 1 in every 200 to 500 children and adolescents are affected by familial hypercholesterolemia (FH), a genetic cholesterol disorder caused by a mutation of low-density lipoprotein (LDL) receptor gene.^{1,2} FH sharply increases the risk of future cardiovascular events (eg, stroke and heart attack); classical studies suggest 25% of affected, untreated women and 50% of men experience an event by age 50 y.³ Because of its autosomal dominant pattern of inheritance, expert guidelines recommend screening of children and adolescents and young adults (AYAs) with strong family or personal histories of cardiovascular risk (eg, early event or death, obesity, smoking, hypertension, and diabetes).^{4–8} If screening identifies high LDL cholesterol, the recommended treatment emphasizes lifestyle modifications (diet and exercise) and, if cholesterol levels do not decline sufficiently, lifetime use of HMG-CoA reductase inhibitors (henceforth “statins”).

AYAs with FH are at high risk for both underdiagnosis and inadequate treatment. For example, Nordestgaard et al⁹ provide an estimate that less than 1% of individuals with FH are diagnosed with the condition in the United States. In addition, it is not known what proportion of AYA care is consistent with the most recent recommendations included in the National Heart, Lung, and Blood Institute’s Expert Panel’s 2011 report recommending universal screening for hyperlipidemia and the use of statins in children and AYAs.⁷ Moreover, little is known about the factors that influence AYAs with FH and their families to undergo or forego screening and treatment. AYAs are at an age developmentally where decision making moves from primarily parent-driven to patient-driven. This presents specific clinical challenges for screening, such as when AYAs attend clinic visits unaccompanied by an adult and are unaware of family history,¹⁰ and for treatment adherence.

To date, there has been little research conducted on how the process of shared decision making between clinicians, AYAs with FH, and their parents might influence screening and treatment perceptions and decisions. Shared decision making is defined as a “collaborative process that allows patient and their providers to make health care decisions together, taking into account the best scientific evidence available and the patient’s values and preferences.”¹¹ In this article, we posit that patients and families use a variety of “evidence” in their decision making, drawing on the family’s medical history and shared narrative of family medical experiences, as well as the index patient’s personal medical experiences.

Together, this “experiential evidence” influences screening and treatment perceptions. Ideally, a shared decision-making process incorporates this experiential evidence with up-to-date research and clinical evidence to create a shared treatment plan. We examine this process within the context of decision making among AYAs and parents of AYAs regarding screening and treatment for FH.

Material and methods

Recruitment

Participants were recruited from a pediatric preventive cardiology practice affiliated with a free-standing pediatric hospital in Massachusetts. The practice primarily serves privately insured patients, with 12.8% of families receiving public insurance (Medicaid and/or Medicare). Approximately 8% of the patient population self-identified as African American. Electronic records were used to identify AYAs aged 17 to 21 y with a history of LDL >190 mg/dL before statin treatment or an LDL >160 before statin treatment and a family history of early cardiovascular disease in male first- or second-degree relatives aged <55 years or female first- or second-degree relatives if aged < 65 y. For both AYAs and parents, additional inclusion criteria included (1) no cognitive or communication disorders that would limit participation in interviews and (2) working knowledge of spoken and written English. AYAs (aged 17–21 y) diagnosed with FH and parents of AYAs with FH were sent letters describing the study with the opportunity to ‘opt-out’ if they did not wish to be recruited into the study. Twenty-four participants (12 AYAs and 12 parents) were recruited and interviewed over the course of 13 mo (07/13–09/14); participants were not required to consent as a dyad. We concluded sampling when we achieved saturation in our thematic analyses within the sample of AYAs and parents, respectively (see Data Analysis below). The study was approved by the Institutional Review Board of Boston Children’s Hospital.

Interview and survey procedures

A brief, quantitative survey followed by a face-to-face, semistructured qualitative interview was conducted with participants, both parent and AYA. Interviews lasted approximately 60 min and were held at 2 outpatient clinic sites. Six interviewers received initial training and ongoing review from a trained qualitative researcher (T.M.) to ensure comparable interview guide facilitation to acquire AYA and parent perspectives on several clinical conditions linked to cardiovascular disease during adulthood (this article focuses only on those with FH). Participants received monetary compensation for their travel and time. Each interview was audio-taped and transcribed verbatim.

Measures

The quantitative survey addressed 3 primary domains, specifically (1) demographic characteristics of the respondent, (2) respondent-reported hereditary risk based on AYA’s biological family, and (3) AYA health care decision making and service use. For the interview guide, we used hypothetical vignettes and open-ended questions to query the perceptions of (1) high cholesterol, (2) screening, (3) lifestyle modification and statins as treatment modalities, and (4) treatment preferences. The interviewer asked open-ended questions to pursue themes of central interest to this study, including the role of family

medical history, family's past experiences, and the index AYA's prior screening and treatment history for FH. Additional details about the measures used for the quantitative survey and the semistructured interview guide are available in the Supplementary Materials for this article.

Stakeholder engagement

Consistent with priorities of the Patient-Centered Outcomes Research Institute, which provided funding for this study, stakeholder engagement was considered central to the research process. A 20-person stakeholder panel was purposefully structured to include a diverse sample of 9 patient and parent stakeholders affiliated with 2 urban medical centers in the Northeast, representing 3 clinical subgroups: (1) FH, (2) cardiovascular risk due to another chronic illness (eg, obesity) and, (3) no known cardiovascular risks. The panel also included 11 additional members, purposefully structured to include (1) pediatric and adult clinicians and researchers with expertise in pediatric obesity and cardiovascular health, (2) individuals who assisted in the development and dissemination of relevant guidelines on high cholesterol screening and treatment, and (3) a health communications expert.

Before funding, stakeholder engagement occurred through one-on-one conversations to assist in developing priorities for the proposed research. Once the study was initiated, stakeholder engagement continued as needed through capacity building sessions, bi-monthly Web-based conference calls, surveys, and cognitive interviews. Bi-monthly meetings occurred for approximately an hour and a half to garner input on study design, development of study tools, and interpretation of findings. For this article, stakeholder engagement led us to (1) use individual interviews with AYAs and parents rather than focus groups to reduce concerns around confidentiality and sensitivity, (2) change the language and ordering of questions in the semistructured interview guide to improve comprehension, and (3) generate additional interpretations of the research findings, as described in the results section of this article.

Data analysis

Survey data were analyzed using descriptive statistics in Stata.¹² Qualitative data from semistructured interviews were analyzed using a modified grounded theory approach, referred to as "Coding Consensus, Co-occurrence, and Comparison," in which analyses are derived from the data and then illustrated by characteristic examples. Accordingly, we used intensive group discussion and "consensus" as our agreement goal rather than quantitative measures of inter-rater agreement.^{13,14}

Transcripts of 6 initial interviews were independently coded by an interdisciplinary team of investigators, including an AYA physician, pediatric preventive cardiologist, 3 clinical researchers, and a health services researcher. Over the course of several meetings, investigators coded excerpts of the transcripts, ranging from a phrase to several paragraphs, based on a priori or emergent themes. A final set of code definitions were then discussed, resolved, and delineated. All remaining transcripts were independently coded by at least 2 investigators, with discrepancies discussed and resolved. Coded data were subsequently entered into DeDoose,¹⁵ a mixed methods software program. Data were organized into

thematic categories, and links were generated between categories to build a theoretic model. This model was developed with assistance from the stakeholder panel and analytics conducted in DeDoose.

Results

Study sample

The sample consisted of 12 AYAs with FH and 12 parents of AYAs with FH. Demographic characteristics are provided in Table 1. Although all respondents were interviewed individually, 9 participants composed of 4 dyads (ie, parent and AYA arrive from the same family, with 2 parents of 1 AYA participating). All parents (11 of 11 parents with nonmissing data) and 10 of 12 AYAs reported having a first- or second-degree relative (ie, sibling, parent, grandparent, aunt, or uncle) with a known high cholesterol diagnosis. In addition, 7 AYAs and 8 parent respondents reported that a relative had previously experienced at least one heart attack.

Conceptual framework

Consonant with studies of other disorders,¹² our exploratory investigation found that family medical history (ie, risk of FH diagnoses or associated cardiovascular disease within the family), family medical experiences, and personal medical experiences influenced risk perceptions and decisions related to cholesterol screening and treatment among AYAs with FH and their parents. As FH tends to be a “silent disease” during adolescence, family medical history and medical experiences primarily influenced the decision to screen and pursue specific treatments (indicated in Fig. 1 with a “+”) or not to pursue treatments (indicated with a “-”) during childhood by informing perceptions of: (1) hereditary risk of FH diagnoses, (2) risk of cardiovascular disease, (3) risks associated with treatment options, and (4) perceived capacity to comply with recommended treatments. Notably, AYAs and their parents drew on familial medical evidence of both cholesterol-related and unrelated disorders in assessing the risks associated with different treatment options for FH. In concert with the clinical team, who bring the available research and clinical evidence to the decision-making process (as noted in Fig. 1), families and AYAs will decide to be screened and if positive, select among treatment options. In turn, the subsequent experiences of the index AYAs provided additional personal medical evidence that fed back to inform AYA and parent perceptions of the medical history and experiences drawn from other family members. We characterize this relationship as a feedback loop. Specifically, familial medical evidence influenced perceptions of health risk and, by extension, the health-related decisions of AYAs with FH. Over time, our data suggest that the AYA’s experience became part of the family’s narrative, thus forming a full feedback loop not only for the index patient but for the nuclear family as well.

Findings

Perceived hereditary risk

All 12 AYAs with FH and 12 parents of AYAs with FH articulated the etiology of FH to be hereditary and spoke of an intergenerational family history, frequently mentioning the

specific biological relatives who shared the condition. For example, one mother stated: “[My daughter] had high cholesterol ... And her father and her father’s family had high cholesterol so it was not a big surprise.” AYAs also emphasized the genetic etiology of the high cholesterol, “I know for a fact that my whole family has high cholesterol.”

Parents and AYAs frequently compiled patterns of high cholesterol, heart attacks, and/or strokes into a narrative of shared family history. Hereditary risk might be linked to a single individual or multiple family members (see Table 2, Theme 1.2, parental quotes). Parents frequently cited the extent of a family history, such as one parent’s numeric representation of “2 of 3” family members being affected. Notably, concern of hereditary risk persisted among parents even in cases where a child may be assessed as having “normal” levels of cholesterol. This ongoing risk was articulated by one parent as a “time bomb in there that isn’t showing now but that doesn’t mean that it’s not going to show up sometime in the future and again simply because of the DNA and genetics.” At times, family medical history of another, unrelated disorder also served to heighten a sense of risk, providing a “horns” effect (ie, the negative direction of the “halo” effect): “You know, I have risk in my family of breast cancer because my mom had ovarian cancer and her mom and all. So you’re never like, you wouldn’t rate it as somebody who didn’t have that family history. So even though he’s doing great, he’s still going to have that risk.”

Perceived risk of associated adverse outcome (ie, cardiovascular disease)

Although these shared medical histories and medical experiences were persistently influential, the nature of the available evidence could either cause additional concern around the potential health impact or minimize these concerns. Notably, diagnosis and treatment for high cholesterol gained additional import after experiencing the impact of a cholesterol-associated event on a close relative (see Table 2, Theme 2.0, for illustrative quotes). As one parent said: “My daughter. I don’t think she really understood what [high cholesterol] really meant until she came here and talked with doctor. And then I think shortly after that her dad had a heart attack, and he had to have a stent put in and that was kind of confirmation that it was not something that she could just ignore even though she felt very healthy.” One AYA described how his father’s death serves to heighten his adherence to treatment, as well as his anxiety about possible outcomes: “Yeah, well, my dad passed away of a heart attack. He was on medicine, overall a healthy guy. so the worry [of high cholesterol] comes from that. You know, what if that happens to me if I don’t keep it under control. So it’s always a stress and anxiety trigger.” Contrastingly, another AYA expressed less concern because of his father’s experience with the condition: “It doesn’t really worry me that my cholesterol would be very high. I think the first lipid profile I had was in the three hundreds, which I guess is bad. I think my dad had one way worse back when he was like thirty.” Parents also highlighted that recent treatment advances have altered perceptions of the associated adverse outcomes for younger generations (see Table 2, Theme 2.0, second parental quote).

Influence of shared family narratives across the screening and treatment trajectory

Shared family medical histories and experiences derived intergenerationally and intragenerationally served as (1) cues to screening and (2) criteria for assessment of treatment effectiveness based on cholesterol-related and unrelated medical conditions.

Reflecting the salient role of family's experiences on perceived risks and associated outcomes of FH (see Table 2, Theme 2.0), these data also demonstrate how the family's experiences, rather than AYA's own experiences, initially influenced decisions to screen and selection of treatments options for the AYAs.

Cue to screen

Perceptions of hereditary risk based on family medical history influenced parental decisions to screen for high cholesterol in the index AYA, often at a very young age (see illustrative quotes in Table 2, Theme 3.1). Frequently, the need for screening at a young age was included as part of the family narrative regarding hereditary risk (see AYA quote Table 2, Theme 3.1). One parent articulated the need for clinicians to recommend the screen proactively, as in her case, she had only just learned of the child's hereditary risk, (see parent quote, Table 2, Theme 3.1).

Effectiveness of treatment options

Understanding of treatment effectiveness frequently relied on the experiences of other family members. For example, one mother described how her own experiences with medications to treat high cholesterol influenced her conversations with her AYA daughters, both of whom have FH: "I've been successfully on the medication so now I'm turning to those kids ... 'Now what, girls?' You know, because this is working for me. 'My cholesterol is less than half of what yours are,' you know?. And here and so now see how my attitude changed?. It's like, 'I'm on it,' you know." As illustrated in Table 2, Theme 3.2, other parents indicated the importance of their own negative experiences when making treatment decisions as a deterrent to medication use for their AYA. AYAs, too, expressed how their perceptions of their parents' experience have influenced their perceptions of the respective treatment options (see AYA quote in Table 2, Theme 3.2).

Although these examples draw from experiences related to high cholesterol, experience with unrelated clinical conditions also informed AYA treatment decisions for high cholesterol. For example, one AYA cited a shared family narrative regarding her mother's use of medication for a noncholesterol-related condition (congenital heart disease) and how the mother's negative experiences with medication for this condition resulted in the AYA's determination not to use medications for her high cholesterol (see Table 2, Theme 3.2, parental quote 2). These experiences of other family members with treatments were easily recalled by parent and/or AYA, linked with affect or emotion, and influenced assessment of treatment effectiveness and subsequent treatment decisions for the AYA.

Perceptions of capacity to comply with treatment options

Family experiences were also a source for strategies to facilitate adherence to recommended treatment options. Respondents considered the lived experience of other family members as an asset in ensuring treatment adherence for the AYA. One mother specifically articulated her perception of the positive value of having her husband assist with reinforcing dietary recommendations (see parental quote, Table 2, Theme 4.0). This example also illustrates how shared family experiences with high cholesterol frequently meant that physician-recommended lifestyle modifications were put in place before a child's FH diagnosis to

accommodate the condition of a parent or another relative. Congruent with this finding, many AYAs refer to the healthy eating and exercise as something “that we’ve always done” as a family.

Feedback loops from AYA’s experiences

Although family experience frequently influenced perceptions of treatment effectiveness, it did not always persist after a treatment plan was developed in concert with the AYA’s provider(s) initiated for the AYA. Some respondents expressed how the AYA’s experiences in screening and treatment provided a wholly new narrative of risk for adverse events associated with FH and treatment effectiveness. As one parent said:

“It’s not a death sentence. It has been in the past for our family. It has for 3 generations but I don’t see it as a death sentence now. I see it as, based on the care that we’ve actually gotten here (pediatric preventive cardiology clinic), I see it as a team, you need a team, a team approach and (your cardiologist) has been pretty much the leader of our team.”

In another example, an AYA explained medications are more likely to be effective because, “you can’t really change genetics with behavior;” as a young adult, he drew on his own experience and those of his siblings to deduce this conclusion, stating that “(behavior) did nothing for (them)” (see Table 2, theme 5.0). These new and progressing experiences with treatments thus functioned in a feedback loop to inform and even “rewrite” the shared narrative of experiential evidence that historically influenced family preferences for cholesterol screening and specific treatment options.

Stakeholder response

“I thought the discussion on family history was very interesting. that (family history) is fragmented over time, and that knowledge about family history can be age and experience related.. how experience relates to perceived level of risk and the medical decision that are affected by this.”

-Parent stakeholder panel member

As noted in the quote mentioned previously, our study findings resonated with the experiences of stakeholder panel members with important implications for clinical care. Our stakeholder panel highlighted the importance of broadening conceptualizations of family medical history from dichotomized yes or no responses to narratives based on related and unrelated clinical conditions. One parent stated that experiential evidence provides a “starting point” when she makes health care decisions for herself or her child. Building on her comment, a pediatrician emphasized the salience of experiential evidence on patient preferences in her practice; she noted that when patients were not adhering with her clinical recommendations for FH they often provided a reason that stemmed from their own or another relative’s prior experience. In responding to these insights, another clinician stated that experiential evidence is not routinely collected as part of a “family history,” which typically only assesses hereditary risk rather than prior experiences of screening and treatment within the family. The clinicians and parent stakeholders expressed the value of conceptualizing the family medical history as including not only hereditary risks and

conditions but also experiential evidence from past and current experiences that may influence the screening and treatment preferences of AYAs with FH and those of their parents.

Stakeholders also commented that mothers were often the “keepers” of these family narratives and that as AYAs took on more decision-making capacity for their own care, it would be important for parents to assure they shared this information. AYA stakeholders shared their own experience of not knowing about certain details of their family history with their own medical conditions. Together, parent, AYA, and clinician stakeholders highlighted the need for greater attention to a shared understanding across AYAs, their families, and their clinicians of this experiential evidence.

Discussion

In this study, we find that family medical history, family medical experiences, and the medical experiences of the index patient (ie, experiential evidence) influence the screening and treatment perceptions and, ultimately, decisions confronted by AYAs with FH and their parents. This narrative of experiential evidence is initially drawn from other family members (eg, parents, grandparents, uncles, aunts, and siblings), and over time, incorporates the experiences of the index patient and modifies the family narratives through a feedback loop. By using the experiences of other family members to inform preferences, experiential evidence prompted decision making regarding screening and treatment in this sample that was consistent with available guidelines. At times, unrelated medical experiences also influenced patients. Although the characteristics of the patient (eg, age and cholesterol level), disease, and treatment may not have been clinically relevant, these experiences still had an impact on the FH-related preferences expressed by the parent or AYA.

Prior research, albeit limited, is mixed regarding the influence of patients’ and families’ preferences on decisions pertaining to early identification and treatment of FH. Some research suggests that identifying FH through selective screening is challenging because patients have a limited understanding of FH pathology and inheritance.¹⁶ Alternatively, selective screening may fail to identify FH in families known to have cardiovascular risk if the projected emotional trauma from sharing a gene with a parent or a close relative who has experienced premature cardiovascular death deters families from seeking early diagnosis.^{17,18} Yet, other studies suggest that selective screening is preferred by adults, despite short-term emotional distress, to mitigate uncertainty and be fully informed about diagnoses.^{16,19,20} These studies, taken together, suggest varied preferences in deciding to receive screening for FH, but offer limited explanation for this variation. Our research suggests that family medical experiences with high cholesterol congeal into narratives that strongly influence this variation in family preferences regarding early identification of high cholesterol.

Previous exploratory studies on decision making regarding FH treatment recommendations also demonstrate discordant findings. For example, families may engage in a “culture of expectancy,” normalizing a diagnosis of FH and promoting motivation and adherence to lifestyle modifications and medication use.^{18,20} In contrast, others find lifestyle modification

requires clinicians to proactively recommend practical skills that facilitate behavioral changes.¹⁷ Although not reported in these studies, prior family experiences of associated adverse outcomes and treatment effectiveness may, in part, explain why some families hold a “culture of expectancy” and yet others require clinical intervention to facilitate behavioral change.

Notably, our findings likely inform future research on health care decision making among AYAs and their parents. AYAs with genetic conditions may be especially influenced not only by their own personal experiences but also by the experiences of other family members. Our findings extend a robust literature on the role of personal and family medical history in medical decision making among adults. Among adults, availability and affect heuristics have been written about extensively in relation to risk perception and communication among adults.^{21,22} The availability heuristic suggests the ease with which examples of a hazard, or potential negative event, can be brought to the mind serves as a cue for estimating the probability of that hazard. Past experiences have been conceptualized as a “cue” that is heavily relied on to assess risk²²; these have been studied in adults with disorders such as FH,¹⁹ cancer,²³ and in natural disasters.²⁴ Contrastingly, the affect heuristic posits that strong emotional experiences with a hazard may be important for increasing perceived risks.²² Recent literature suggests that risk perception incorporates availability and affect heuristics through ease of recall, particularly for those remembered images that are connected with affect.²¹ Extant literature has focused primarily on adults, with limited attention to AYAs who are increasingly likely to be engaged, themselves, in shared decision making with their parents and clinicians.

Our findings suggest that ease of recall suggested by the availability heuristic, and the associated affect referred to in the affect heuristic, may derive from both personal and other family members’ experiences, whether intergenerationally or intragenerationally. These findings emphasize the import for future research of AYA decision making to consider not only the familial medical histories but also the ease of recall and associated affect of AYAs’ own experiences (as emphasized in the literature on availability and affect heuristics among adults^{9–14}). In addition, our findings highlight that AYA and family understanding of disorders are often influenced by the experiences of other family members, even if these experiences are with conditions that are not clinically related to FH; our findings parallel perspectives highlighted in a recent article on attitudes toward taking medications for heart disease.²⁵

These findings have 2 central implications to clinical practice and policy. First, our findings suggest that an understanding of prior experiences, both of the family and the adolescent, may help the clinician unearth assumptions that exist and help to inform the development of a treatment plan that incorporates both families’ experiential evidence and up-to-date research and clinical evidence. Families’ assumptions may be especially important to address as patients and parents may be drawing from prior experiences with cardiovascular risk factors, events, or treatments that are not clinically related to FH; this may result in compromising the health literacy of the patient and parent, specifically limiting their ability, as defined by the Institute of Medicine, “to obtain, process, and understand health information necessary for making appropriate health decisions.”²⁶ Notably, eliciting

families' experiential evidence and providing patient and parent education may address the potential for experiential evidence to compromise the health literacy of AYAs and families. Second, medical history and treatment experiences of AYAs may influence older (and higher risk) relatives to receive evidence-informed screening and treatment. With advancements in the treatment of AYAs with FH, many respondents indicated that they learned about the promise of new treatments informed by research and clinical evidence that are available to manage high cholesterol and deter future adverse events. In these cases, clinicians are well positioned to encourage the patient and/or parent to have family members also receive screening and treatment for FH.

The selected sample for the present study derives from 2 pediatric preventive cardiology clinics associated with an urban academic tertiary hospital. As a population that is engaged in services, we anticipate that this sample holds greater interest in seeking FH care. Although this sample facilitates explorations into the role of experiential evidence, we recognize it does not likely account for the full heterogeneity of experiences of AYAs with FH, especially those who are not engaged in care. This sampling bias might help to explain why we did not have study participants who reported preferences for 2 potential treatment options, specifically the use of statins alone and the decision to pursue no treatment when diagnosed with high cholesterol. However, our findings do highlight that even in this engaged sample, experiential evidence influenced screening and treatment decision making. Future research could further examine these findings in larger and more diverse samples. In addition, outside of our stakeholder panel, we did not incorporate providers' viewpoints on the juxtaposition of families' experiential evidence and research and clinical evidence. Research is needed to explore best practices for incorporating families' experiential evidence and research and clinical evidence in shared decision making.

Conclusions

By focusing on the process by which familial and personal medical experiences (coined "experiential evidence") influence screening and treatment decisions, we provide new insight into shared decision making in AYAs with genetic conditions. Our findings emphasize how experiential evidence drawn from shared family narratives influences the screening and treatment decisions of AYAs with FH and their parents. By recognizing the importance of both familial and personal experiences in addition to family medical history, clinicians are provided with the opportunity to understand their influence and assist patients in understanding these past experiences as part of the shared decision-making literature. Consistent with recommendations from the stakeholder panel (see Feedback loops from AYA's experiences in Findings section), future studies might explore how to incorporate prior family experiences into the assessment of a patient's recorded family medical history. Current efforts to build medical homes and develop electronic medical records that capture patient preferences might be especially well positioned to capture and accommodate influential experiential evidence in screening and treatment decisions for AYAs and parents affected by FH.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

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References

1. Gooding HC, Rodday AM, Wong JB, et al. Application of Pediatric and Adult Guidelines for Treatment of Lipid Levels Among US Adolescents Transitioning to Young Adulthood. *JAMA Pediatr.* 2015;169(6):569–574. [PubMed: 25845026]
2. Simon Broome Steering Committee. Risk of fatal coronary heart disease in familial hypercholesterolaemia. Scientific Steering Committee on behalf of the Simon Broome Register Group. *BMJ.* 1991; 303(6807):893–896. [PubMed: 1933004]
3. Brown MS, Goldstein JL. Familial hypercholesterolemia: a genetic defect in the low-density lipoprotein receptor. *N Engl J Med.* 1976; 294(25):1386–1390. [PubMed: 177875]
4. Daniels SR, Greer FR, Committee on Nutrition. Lipid screening and cardiovascular health in childhood. *Pediatrics.* 2008;122(1):198–208. [PubMed: 18596007]
5. United States Preventive Services Task Force. Screening for lipid disorders in children. 2007; Available at: <http://www.uspreventiveservicesattaskforce.org/upstf/upspchlip.htm> Accessed December 14, 2014.
6. Haney EM, Huffman LH, Bougatsos C, Freeman M, Steiner RD, Nelson HD. Screening and treatment for lipid disorders in children and adolescents: systematic evidence review for the US Preventive Services Task Force. *Pediatrics.* 2007;120(1):e189–e214. [PubMed: 17606543]
7. Daniels SR, Benuck I, Christakis DA, et al. Expert panel on integrated guidelines for cardiovascular health and risk reduction in children and adolescents Full Report. Bethesda, MD: National Heart, Lung, and Blood Institute, U.S. Department of Health and Human Services; 2011.
8. Daniels SR, Gidding SS, de Ferranti SD. Pediatric aspects of familial hypercholesterolemias: recommendations from the National Lipid Association Expert Panel on Familial Hypercholesterolemia. *J Clin Lipidol.* 2011;5(3):S30–S37. [PubMed: 21600527]
9. Nordestgaard BG, Chapman MJ, Humphries SE, et al. Familial hypercholesterolaemia is underdiagnosed and undertreated in the general population: guidance for clinicians to prevent coronary heart disease Consensus Statement of the European Atherosclerosis Society. *Eur Heart J.* 2013;34(45):3478–3490. [PubMed: 23956253]
10. Slovic P, Finucane ML, Peters E, MacGregor DG. Risk as analysis and risk as feelings: some thoughts about affect, reason, risk, and rationality. *Risk Anal.* 2004;24(2):311–322. [PubMed: 15078302]
11. Informed Medical Decisions Foundation. What is shared decision making? 2014; Available at: <http://www.informedmedicaldecisions.org/what-is-shared-decision-making/> Accessed December 10, 2014.
12. Stata Statistical Software: Release 12 [computer program]. College Station, TX: StataCorp LP; 2011.

13. Willms DG, Best JA, Taylor DW, et al. A systematic approach for using qualitative methods in primary prevention research. *Med Anthropol Q.* 1990;4(4):391–409.
14. Harry B, Sturges KM, Klingner JK. Qualitative data analysis: mapping the process. *Educational Researcher.* 2005;34(2):3–13.
15. Dedoose Version 5.0.11, web application for managing, analyzing, and presenting qualitative and mixed methods research data [computer program]. Los Angeles, CA: Sociocultural Research Consultants, LLC; 2014.
16. Agard A, Bolmsjo IA, Hermeren G, Wahlstrom J. Familial hypercholesterolemia: ethical, practical and psychological problems from the perspective of patients. *Patient Educ Couns.* 2005;57(2):162–167. [PubMed: 15911189]
17. Hardcastle SJ, Legge E, Laundry CS, et al. Patients' perceptions and experiences of familial hypercholesterolemia, cascade genetic screening and treatment. *Int J Behav Med.* 2015;22(1):92–100. [PubMed: 24585182]
18. Weiner K, Durrington PN. Patients' understandings and experiences of familial hypercholesterolemia. *Community Genet.* 2008;11:273–282. [PubMed: 18493125]
19. Tonstad S Familial hypercholesterolaemia: a pilot study of parents' and children's concerns. *Acta Paediatr.* 1996;85(11):1307–1313. [PubMed: 8955457]
20. Hollands GJ, Armstrong D, Macfarlane A, Crook MA, Marteau TM. Patient accounts of diagnostic testing for familial hypercholesterolaemia: comparing responses to genetic and non-genetic testing methods. *BMC Med Genet.* 2012;13(1):13–87. [PubMed: 22397687]
21. Slovic P, Peters E, Finucane ML, MacGregor DG. Affect, risk, and decision making. *Health Psychol.* 2005;24(4S):S35–S40. [PubMed: 16045417]
22. Keller C, Siegrist M, Gutscher H. The role of the affect and availability heuristics in risk communication. *Risk Anal.* 2006;26(3): 631–639. [PubMed: 16834623]
23. Farrell MH, Murphy MA, Schneider CE. How underlying patient beliefs can affect physician-patient communication about prostate-specific antigen testing. *Eff Clin Pract.* 2002;5(3):120–129. [PubMed: 12088291]
24. Siegrist M, Gutscher H. Flooding risks: a comparison of lay people's perceptions and expert's assessments in Switzerland. *Risk Anal.* 2006; 26(4):971–979. [PubMed: 16948689]
25. Rosenbaum L Beyond belief—how people feel about taking medications for heart disease. *N Engl J Med.* 2015;372(2):183–187. [PubMed: 25564902]
26. Nutbeam D Health literacy as a public health goal: a challenge for contemporary health education and communication strategies into the 21st century. *Health Promot Int.* 2000;15(3):259–267.

Figure 1.
Role of experiential evidence in shared decision making.

Table 1**Sample description**

Sample Characteristics	Parent, n = 12, n (%) or mean (SD)	AYA, n = 12, n (%) or mean (SD)
Race/ethnicity		
Asian	0	1 (9)
Black or African American	1 (10)	1 (9)
White	9 (90)	9 (82)
Missing	2	1
Female	10 (83)	6 (50)
Latino or Hispanic	1 (8)	2 (17)
Insurance		
Private	9 (82)	7 (58)
Public	2 (18)	1 (8)
Don't know	0	4 (33)
Missing	1	0
Age, y	49.3 (7)	18.4 (1)
Self- or parent-reported hereditary risk of AYA		
Early heart attack, heart procedure, or angina		
None	2 (18)	1 (9)
Parent or sibling	1 (9)	3 (27)
Grandparent, aunt, or uncle	7 (64)	4 (36)
Don't know	1 (9)	3 (27)
Missing	0	1
Early stroke		
None	5 (50)	5 (50)
Parent or sibling	0	0
Grandparent, aunt, or uncle	5 (50)	2 (20)
Don't know	0	3 (27)
Missing	2	2
High cholesterol		

Sample Characteristics	Parent, n = 12, n (%) or mean (SD)	AYA, n = 12, n (%) or mean (SD)
None	0	0
Parent or sibling	9 (82)	7 (58)
Grandparent, aunt, or uncle	2 (18)	3 (25)
Don't know	0	2 (17)
Missing	1	0
Diabetes		
No	5 (46)	2 (18)
Parent or sibling	1 (9)	0
Grandparent, aunt, or uncle	4 (36)	5 (46)
Don't know	1 (9)	4 (36)
AYA health care decisions and service use		
Parent is "somewhat" or "very" involved in AYA health care decisions	12 (100)	12 (100)
Agree/strongly agree to gathering information in health care decisions	12 (100)	11 (92)
Agree/strongly agree to being fearful of new medical information	4 (33)	8 (67)
Current use of prescription medicines to lower cholesterol by AYA	10 (83)	9 (75)

AYA, adolescents and young adults; SD, standard deviation.

Table 2

Illustrative quotes

Sample	Illustrative quote
1.0. Perceived hereditary risk	
1.1. Perceived etiology of high cholesterol	
Parent	"(My daughter) had high cholesterol and that she was a thin kid and she was eating well and she was exercising so it was pointing towards hereditary cause. And her father and her father's family had high cholesterol so it was not a big surprise."
AYA	"I know for a fact that my whole family has it and I know it increases your chance of heart attack, possibly strokes... It means that you could be like in danger of like what could possibly happen like in the future if you don't change anything."
1.2. Perceived patterns of high cholesterol and severity of associated events	
Parent	"A maternal uncle died at the age of 19 of a stroke. So, with that knowledge, we raised that knowledge to the pediatrician who then took a blood test, saw that there was high cholesterol, and then recommended we see a specialist."
Parent	"[My husband] has the hyperlipidemia and his family, his father never was tested for high cholesterol, but he died at age 33 from a heart attack. He had an uncle, his father's brother, died at 34 from a heart attack and there's a third brother that's living, he's in his 80s now... And then the brother that has a child, she does not have the high cholesterol. (name) has 2 sisters, his older...from his father. His oldest sister has the high cholesterol. And one of her...one or both of her children have hyperlipidemia... And then in our family 2 out of the 3 girls have high cholesterol. So it seems to run 2 out of 3."
AYA	"A lot of my family members on my dad's side didn't really make it since high cholesterol. My cousin died at age 21 from her heart ... high cholesterol and heart disease and she was the oldest one with the heart disease to live for a girl. And my grandpa died at 50 or 52. And it's a worry because they started off with high cholesterol and they were pretty healthy and then it got more intense."
1.3. Perceived concern for hereditary risk despite "normal" cholesterol level in screen	
Parent	"The cholesterol could become an issue as you get older with changes, hormonal changes I mean that could...my mom didn't get high cholesterol till her 40s. But, she's not active at all and her diet wasn't good. So, it came out but I mean there's still something you have to be aware of and monitor and still make healthy choices... Yeah, make sure you get it checked every year. Make sure you're feeding your body right."
2.0. Perceived risk of associated adverse outcomes (ie, cardiovascular disease)	
Parent	"I know we have a long family history. I'm involved in it because I was detected at an early age as well. It's part of life really. So we're dealt cards and we deal with them... I was taking it as similar as possible to my own situation having grown up with it."
Parent	"...they are children and they don't fully understand everything and they're sitting here looking at their own parent who has given it to them and this parent is not unhealthy. You see, if you take me back to my childhood, my parents were unhealthy with this situation because the technology wasn't there yet. They didn't know to check women after menopause. They didn't know to check men who were overweight or overstressed. You know, the stress factors weren't in there. It was the late 1960s early 70s when my parents had their first incidents. So I was a young person when I saw this. So when I found out I had it I wasn't all that surprised because my parents had it."
AYA	"You get risk of heart attacks... you can't, it affects your running. I know a lot of people that when they run they can't run for a certain amount of time. It decreases your stamina. I know that it's a very high risk of heart attacks because my grandfather has high cholesterol. I know that when I play basketball, he can't play basketball; he can't really run as long as he can, not like me. It affects you really badly."
3.0. Influence of shared family narratives across the treatment trajectory	
3.1. Cue to Screen	
Parent	"...And, you know, because pediatricians ask you that sometimes, you know, because if the mom's doing good then the kids are probably doing good too... "Well, (HC provider)," I said, "I just got back from the doctor and I found out that my cholesterol's really high and I'm probably going to have to go on statins." And he said, "Oh, really?" And I said, "Yeah, and I probably inherited it from my dad or something." And he said, "You know what we need to do, (name)?" And I said, "No, what?" He said, "We need to

get the children checked.” And it was a big light bulb went on for me. It was like, “Oh really? Oh.” And at that point I didn’t know that any of my children had this. And so I had a two year old, a six year old, an eight year old and a ten year old. And two of those four children had cholesterol as high as I did at that time.”

AYA

“So I was tested for high cholesterol when I was two years old because my father has it and his father, so my grandfather, passed away when he was thirty-two years old of a sudden heart attack. At that time they didn’t know really anything about high cholesterol because one, it was back then, and two, he was in Brazil...anyway, so because of that my dad wanted to get us tested because it ran in the family and he wanted to make sure that if we had it we could start treatment early, which happened.”

3.2. Effectiveness of treatment options

Parent

Interviewer: “And how much does having had (a negative medication) experience yourself sort of influence how you think about this for your child? Interviewee: “Hugely... More than hugely... Because this is a child. This is somebody that you bore and now you’re going to make them take something that you yourself has had a problem with. I’m going to put them behind the wheel of a car that I know gets a flat tire every time you turn around? Oh, no, you never put your kids in the car that ... the junker...”

Parent

“(My mom) has a congenital heart disease, so she was on medicine... since she was born. So I know she has had the actual effects of having to eat a lot of medication... I want to say the chemicals wore out enamel and stuff. So she knows the side effects of it and how annoying. It’s not annoying but it really affects her health and the system. Ever since we were young, she has always kind of been against just eating the medicine. If you don’t need it, then don’t eat it or take it... So then that. I’ve been in that mind sense since I was really young, so that’s why I personally want to, don’t really want to take medicine unless I have to.”

AYA

“I always have seen my dad taking pills and stuff so I didn’t really think it was a huge deal. I mean he’s not like the healthiest guy on earth—he’s a little overweight and everything—but I figured (taking medications for high cholesterol) wasn’t really a big deal because he seems to live a pretty normal life.”

4.0. Perception of adherence to treatment options

Parent

“It affected me because I don’t have the condition, you know, (her dad), he’s been living with it since he has known about it at age ten. And you know he made the accommodations for it. You know I had to kind of... besides that fact you know I knew he had high cholesterol so I’d obviously tried to cook in a way to help him. But it just sort of put it you know, it kinda just said okay, this is my new life and this my lifestyle and we’re gonna you know have to do things a certain way. You know you just do it, so. But yeah, so for me it was, yeah. It just seemed more...cause he could help me, you know where she couldn’t, I had to do it for her.”

AYA

“I think it depends on your family history. For me, because of a strong family history, there was kind of no question about the medication because we’d already been doing the healthy diet and the healthy activity regimen beforehand, so we knew that wasn’t the case. So there’s no question that we’d be on medication.”

5.0. Feedback Loop of personal experience

Parent

Interviewee: “I think that knowledge though, me as a parent, I’m cautious because it’s my kids. Knowing what I know now I would recommend kids that have a family history of hypercholesterolemia that I would recommend consideration of statins, where as I wouldn’t probably 12 years ago.”

Interviewer 1: What’s made the difference would you say?

Interviewee: “Experience... So, seeing where my kids are and that there hasn’t been any detriment so far, I think though I would question whether my 16 year old should start ramping up until she’s probably 18 to 20 years old.”

AYA, adolescents and young adults