I, Janel E Phetteplace, hereby submit this original work as part of the requirements for the degree of Master of Science in Genetic Counseling.

It is entitled:
Familial Hypercholesterolemia: Characterization of a pediatric population and evaluation of parental knowledge and attitudes

Student's name: Janel E Phetteplace

This work and its defense approved by:

Committee chair: Elaine Urbina, M.D.
Committee member: Erin Miller, M.S., G.C.G.
Committee member: Ashley Parrott, MS
Committee member: Valentina Pilipenko, Ph.D.
Committee member: Amy R (Garrison) Shikany, MS, LCGC
Committee member: Melanie Myers, Ph.D.
Familial Hypercholesterolemia: Characterization of a Pediatric Population and Evaluation of Parental Knowledge and Attitudes

A thesis submitted to the Graduate School of the University of Cincinnati in partial fulfillment of the requirements for the degree of Master of Science in the Department of Pediatrics of the College of Medicine

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By

Janel Phetteplace
BS, University of South Florida, 2013
Committee Chair: Erin Miller, MS, LGC
Committee Members: Robert Hinton, MD
Melanie Myers, PhD, MS, LGC
Ashley Parrott, MS, LGC
Val Pilipenko, PhD
Amy Shikany, MS, LGC
Elaine Urbina, MD, MS
Abstract

Background: Familial Hypercholesterolemia (FH) is estimated to affect one in every five hundred people. FH is a lifelong condition that if untreated can lead to premature coronary heart disease (CHD) and possibly death. In spite of the high prevalence, internationally FH is both underdiagnosed and undertreated, leaving a large population of people at a greatly increased risk for premature CHD. There are effective treatments available that can significantly improve long term survival.

Methods: The prevalence of FH at CCHMC for individuals between age 2 and 21 was calculated using i2b2, a research data warehouse at CCHMC. A chart review was completed on 180 patients who were seen between 1/1/2011 and 7/15/2013 at CCHMC and met the inclusion criteria. A 42 item survey was developed to evaluate parental knowledge, attitudes and interest in genetic counseling and genetic testing. 171 surveys were sent out, 86 included an educational brochure created by the FH Foundation and 85 did not include the brochure.

Results: The prevalence of FH in the selected population was 0.04%. 1% of individuals between the ages of 9 and 11 received cholesterol screening at CCHMC. Of the 180 individuals included in the chart review, 146 (81%) had seen a lipid specialist, 47 (26%) had a diagnosis of FH, 105 (58%) had a diagnosis of dyslipidemia or hyperlipidemia, 28 (16%) had been prescribed a statin, 0 (0%) had discussed FH with a genetic counselor. Of the 171 surveys sent, 76 surveys were completed with a response rate of 44%. Respondents whose child had been prescribed a statin had a higher mean knowledge score than respondents who children had not been prescribed a statin (p = 0.0267). If the respondent reported that a healthcare provider had talked to them about FH there was a trend towards significant associated with knowledge score (p = 0.0606). Receiving the educational brochure, having a diagnosis of FH, reported use of the internet to look up FH, annual house hold income and a reported family history of hypercholesterolemia were not associated with increased knowledge.

Conclusions: Results from this study supports existing data regarding the under diagnosis of FH and the lack of adherence to routine cholesterol screening. Participants who reported a health care provider had previously spoken to them about FH were more knowledgeable compared to those who had not. Given the important role healthcare providers play in patient education, incorporating genetic counselors into the counseling and risk assessment of these patients and families may increase overall disease knowledge as well as adherence to cholesterol screening and genetic testing guidelines.

Key words: Familial hypercholesterolemia, high cholesterol, dyslipidemia, parental attitudes, parental disease knowledge, knowledge, genetic counseling, genetic testing, cholesterol screening
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Introduction:

Familial hypercholesterolemia (FH) is an inherited common metabolic disorder that leads to early development of extensive atherosclerosis and coronary heart disease (CHD) (Goldberg, Robinson, Cromwell, Ross, & Ziajka, 2011). It is estimated that FH affects 14 to 34 million people worldwide including at least 620,000 (or 1 in 500) in the US alone (Hopkins, Toth, Ballantyne, & Rader, 2011); (Nordestgaard et al., 2013). The Copenhagen General Population Study found the prevalence of FH in Denmark to be 1 in 137; a significantly higher estimate than what had previously been reported, which may suggest a general under-estimate of disease prevalence internationally (Benn, Watts, Tybjaerg-Hansen, & Nordestgaard, 2012).

FH is defined as a group of inherited genetic defects resulting in elevated plasma concentrations of low density lipoprotein (LDL) cholesterol concentrations. Total cholesterol concentrations in individuals with heterozygous FH are usually between 350 and 550 mg/dL, (Goldberg, Hopkins, et al., 2011). Most often FH is diagnosed clinically using one of three validated sets of diagnostic criteria: US MEDPED program (Make Early Diagnosis, Prevent Early Death), the Dutch Lipid Clinic Network (DCLN) or The Simon Broome Register criteria.

Clinical genetic testing is available for the three most common genes associated with autosomal dominant FH, including LDLR, APOB and PCSK9 which account for 60-80% of cases of FH (De Castro-Oros, Pocovi, & Civeira, 2010). Over 90% of causative mutations associated with FH are found in the low-density lipoprotein receptor gene (LDLR) (Austin, Hutter, Zimmern, & Humphries, 2004b). Most patients with FH have not historically been offered genetic testing. While FH is most commonly diagnosed based on medical and family history, recommendations for genetic testing have been incorporated into the most recent guideline which state that that incorporating a clinical diagnosis of FH with molecular results is the most reliable
mode of diagnosis and recommend that all new diagnoses should have genetic testing (Watts et al., 2014). Both the Dutch Lipid Clinic Network and the Simon Broome Register Criteria for diagnosing FH include genetic testing. The US MEDPED diagnostic criteria do not include genetic testing and is based only on cholesterol levels.

CHD results in 380,000 deaths per year in the United States, making it the leading cause of death (Murphy, Xu, & Kochanek, 2013). FH results in elevated LDL cholesterol, which accelerates the rate of atherosclerosis, specifically in the coronary arteries and the proximal aorta, resulting in a significantly higher risk for premature CHD and associated complications, including myocardial ischemia and sudden cardiac death (Austin, Hutter, Zimmern, & Humphries, 2004a). Numerous studies have established that individuals with FH have a greatly increased risk for premature CHD over the general population. The risk of CHD was reported to be increased twenty-fold in patients with untreated FH (Austin et al., 2004b). Another study found that patients aged 20 to 39 with FH had a 100-fold increase in mortality from CHD and nearly a 10-fold increase in total mortality (Group, 1991). It was initially reported that approximately 50% of men and 30% of women with FH are expected to have a myocardial infarction before 60 years of age (Slack, 1969). More recently it has been reported by the National Human Genome Research Institute that the risk of myocardial infarction may be even before age 60 may be even high, up to 85% in men. Compared to the general population it is clear that there is an increased risk to develop CHD prematurely indicating that FH impacts health earlier and more severely.

Given the well-established association between hyperlipidemia and CHD, cholesterol screening guidelines have been published by the American Academy of Pediatrics. Current recommendations state that non-fasting cholesterol screening should be initiated between ages 9
and 11 in all children and repeated again between the ages of 18 and 21. It is recommended that all children have risk assessments beginning at age 2 and be repeated every two years until age 11 at which time risk assessments should be completed annually. Risk assessments should evaluate for a family history of myocardial infarction, angina, stroke, coronary artery bypass graft/stent/angioplasty, and a parent with known dyslipidemia. If the patient is found to have additional risk, a fasting lipid profile is recommended ("Expert panel on integrated guidelines for cardiovascular health and risk reduction in children and adolescents: summary report," 2011). Data regarding adherence to these screening guidelines in the pediatric setting have not been reported.

While diet and exercise can be effective at reducing cholesterol, medical therapy is often indicated. Statins (or HMG-CoA reductase inhibitor), a class of drugs that inhibit the enzyme (HMG-CoA reductase) that controls cholesterol production in the liver, have been shown to lower LDL by 25-40% and increase HDL by 5-10% (Perk et al., 2012). Statins are the first choice of treatment for individuals with elevated LDL and known FH (Goldberg, Hopkins, et al., 2011); (Reiner et al., 2011). A cohort study by Versmissen et al found that women (n=1026) and men (n=924) with FH who were treated with statins had a 79% and 83% reduction in risk of CHD, respectively, compared to individuals that were not treated. This study also found that individuals with FH over age 55 treated with statins had a risk of myocardial infarction close to that of the general population (Versmissen et al., 2008). In addition, data suggests there are potential and considerable health benefits if FH is diagnosed and treated beginning in childhood. A randomized, double-blind, placebo controlled study involving children between the ages of 8 and 18 with FH found that age at statin initiation was positively associated with carotid intima-media thickness (IMT). As thickening of the carotid artery is an early sign of CHD, the carotid
intima-media thickness test is used to diagnose the extent of carotid atherosclerotic vascular disease. The earlier statin treatment is initiated, the smaller the carotid IMT measurement. The outcome of this study strongly indicate that early statin treatment reduces atherosclerotic burden more vigorously than treatment started later, and support initiation of treatment in childhood (Rodenburg et al., 2007). Another randomized, double-blind, placebo-controlled trial involving children between the ages of 8 and 18 with FH found that two years of statin therapy resulted in a significant regression of carotid atherosclerosis in children and improved the lipoprotein profile. This suggests that adolescence is a critical period for correcting any atherosclerosis that has occurred as a result of FH (Wiegman et al., 2004). There are also nonprescription supplements that have been found to lower cholesterol levels. Plant sterols and stanols naturally occur in all foods from plants and supplements have been found to reduce concentrations of LDL cholesterol in individuals with hypercholesterolemia by slightly more than 10% (Parraga-Martinez et al., 2014).

Despite the high prevalence, associated risks of early CHD, well defined diagnostic criteria and availability of effective treatment, FH remains underdiagnosed and undertreated. It has been suggested that less than 1% of patients with FH receive a diagnosis in many countries (Nordestgaard et al., 2013). The under diagnosis of FH is of significant national concern such that the Office of Public Health Genomics at the Centers for Disease Control and Prevention has identified FH as one of the diseases with the strongest level of evidence for genomic and family health history applications (National Institute for Health and Care Excellence, 2008). Some estimates suggest that only 20% of patients in the United States are diagnosed and of those, only a small minority receive appropriate treatment (Goldberg et al., 2011). Despite the clear benefit of medical therapy, Benn et al. reported that less than half (48%) of patients with FH were
prescribed statins by their physician (Benn et al., 2012). Benn et al suggests that even when prescribed statin therapy, individuals with FH still had a 10-fold increased risk of CHD compared to individuals without FH. These findings were attributed to suboptimal dosing as well as initiation of treatment after severe atherosclerosis had already developed. (Benn et al., 2012). Studies have found that beginning treatment of FH in childhood can have dramatic effects on the IMT progression and disease burden (Rodenburg et al., 2007; Wiegman et al., 2004).

General lack of knowledge and lack of public awareness may be one of the primary contributing factors to under-diagnosis and treatment of FH (Goldberg, Robinson, et al., 2011). Results from a cross-sectional survey of 68 adults with FH found that most patients understood what cholesterol is, the reason for medical treatment, and self-care prevention. However, 66% of the participants lacked knowledge about genetic transmission of FH and 79% lacked knowledge about the role family history plays in FH (Hollman, Olsson, & Ek, 2006). Others have found that people with FH did not inform their relatives of the genetic risk associated with FH (van den Nieuwenhoff, Gielen, & de Vries, 2007). Reasons for nondisclosure included inadequate risk knowledge, risk denial and a perception that disclosing information was interfering. The documented lack of knowledge and communication argues for increased awareness about FH.

FH has received both national and international attention with publication of new guidelines addressing the need for improved screening, diagnosis, and treatment of this disease as well as the importance of incorporation of genetic testing and family history evaluation. Given the focus on early diagnosis in FH, we sought to characterize the population of pediatric patients with FH and examine parental beliefs and attitudes towards FH. The aims of this study were (1) to determine the prevalence of FH in a pediatric institution and further characterize the population by defining additional diagnoses, medications prescribed, and specialists seen, (2) to
assess parental beliefs and attitudes towards and knowledge of FH using a survey, and (3) to evaluate the impact of a FH educational flyer on the genetic and disease knowledge of those who have a child with a clinical diagnosis of FH.

**Methods:**

**Participants**

This study included both a retrospective chart review and cross-sectional survey performed with approval from the Cincinnati Children’s Hospital Medical Center (CCHMC) Institutional Review Board. A retrospective review of patients between the ages of 2 and 21 years who underwent cholesterol screening at CCHMC from January, 2011 to July, 2013 was completed. Patients were considered eligible if they met one or more of the following criteria: (1) had a diagnosis of FH as documented in the electronic medical record (EMR), (2) had at least one LDL level above 155mg/dL and had seen a lipid specialist at CCHMC, or (3) had one LDL level above 190mg/dL. Patients with other diagnoses known to elevate LDL including systemic lupus erythematosus, polycystic ovary syndrome, nephrotic syndrome or history of a solid organ or bone marrow transplant were excluded. All individuals meeting inclusion criteria were included in the chart review. In families with multiple affected relatives meeting inclusion criteria, the more severely affected sibling based on highest recorded LDL cholesterol level was included in the survey. Study participants invited to take part in the survey were parents/caregivers of children meeting the above inclusion criteria.

**Procedures**

Clinical data was abstracted for each child through chart review and entered into a secure database. The parent/caregiver survey was sent from September, 2014 to November, 2014. The questionnaire was mailed to all parents/caregivers of children meeting the inclusion criteria and
e-mailed to those who had an e-mail address in the EMR. Participants who received both the paper and electronic invitation to participate were given the option to respond via preferred method. To ensure there were not multiple responses from the same participant, each participant was given a study number and only one response could be entered into the database for each study number. Half of the study participants were sent an educational brochure produced by the FH Foundation which provides information on prevalence, diagnosis, inheritance, and risks associated with FH. Everyone in the study received a five dollar bill in the initial mailing as an incentive to complete the survey. The two groups had equal representation of children who had previously seen a lipid specialist and their socioeconomic status based on private or government health insurance to reduce any bias associated with these factors. Two weeks after the initial mailing a reminder card and e-mail were sent to all eligible participants who had not yet completed the survey. Participants whose survey had not been returned within a month of mailing were offered the option of completing the survey over the phone. See Figure 1 for an overview of the study procedures.

Clinical Data

Data are reported as means with standard deviations and frequencies, as appropriate. The clinical data abstracted via retrospective chart review included demographics, age at first elevated LDL level, prescribed medications, other medical diagnoses, body mass index (BMI), a reported family history of hypercholesterolemia, number of no show appointments, and consultations performed by any other specialists who evaluated them at CCHMC including lipid specialists, dieticians, geneticists, and genetic counselors.

The survey assessed current parental beliefs and attitudes towards and knowledge of FH as well as demographic information (Appendix A). Survey questions were developed by the
authors specifically for this study. The survey was pre-tested by a convenience sample of ten individuals without extensive knowledge about genetics or FH. Question wording was adjusted based on feedback from the pretest to improve clarity. The survey had seven sections including 42 multiple choice questions; (1) parental demographics and cholesterol status, (2) affected child’s health and diagnosis (3) parental beliefs and attitudes about risk and treatment, (4) disease and genetic knowledge (5) interest in genetic counseling and testing, (6) resources, and (7) family history.

Statistical Analysis

The prevalence of FH was calculated and reported as the proportion of those children who have FH based on the inclusion criteria previously described, compared to all individuals seen at CCHMC between the ages of 2 and 21 during January 2011 to July 2013. The number of unique individuals seen at CCHMC between the ages of 2 and 21 during January 2011 to July 2013 were generated using Informatics for Integrating Biology and the Bedside (i2b2), a research data warehouse at CCHMC. The i2b2 framework is based on the Research Patient Data Registry (RPDR) developed at Massachusetts General Hospital.

A knowledge score was calculated based on the number of disease and genetic specific questions answered correctly in the survey. Each correct response received a score of 1. Incorrect, unsure, or missing responses received a score of 0. A mean knowledge score for each group (received the educational flyer or did not receive the educational flyer) was reported and mean differences analyzed using a Wilcoxon test as the data was not normal. Additional comparisons were made between participants who had and had not seen a lipid specialist. Interest in genetic counseling or genetic testing is reported as the proportion of those in each group who were interested in genetic counseling or genetic testing compared to those who were
not. The proportions from each group were compared using a chi-square test. Parental attitudes towards FH were collected using a ten point Likert scale with 1 being strongly disagree and 10 being strongly agree. For analysis the ten point scale was collapsed into 5 groups. Responses of 1 or 2 were designated as “strongly disagree,” responses of 3 or 4 were “disagree”, responses of 5 or 6 were designated as “neutral”, responses of 7 or 8 were designated as “agree”, and responses of 9 or 10 were designated as “strongly agree.” Statistical analysis was done using JMP Genomics 7.0 (SAS Institute, Cary, NC).

**Results**

*Prevalence and patient characteristics*

Approximately 1262 individuals between the ages of 2 and 21 underwent cholesterol screening between January 1, 2011 and July 15, 2013 and had an LDL of over 155mg/dL with 180 meeting inclusion criteria. Based on the number of individuals who met the inclusion criteria and the number of total individuals seen at CCHMC for all visit types within the age and date range (n=381827) the estimated prevalence of FH was 0.04% or 4/10,000. Of the 180 individuals who met inclusion criteria, forty-seven (26%) had a diagnosis of FH in the EMR, 105 had at least one LDL greater than 190 mg/dL and 144 had a LDL greater than 155 mg/dL and saw a lipid specialist (Figure 2). At CCHMC there were 89071 patients between the ages of 9 and 11 seen in the set timeframe, of those only 948 (1%) had cholesterol screening. The racial and ethnic composition of the retrospective chart review group was primarily Caucasian and non-Hispanic (75%) with 17% Black, 2% Asian and 2% Hispanic. Based on EMR documentation, 146 of the 180 patients (81%) had previously seen a lipid specialist, including all 47 patients with a diagnosis of FH recorded in the EMR. One hundred and five (58%) had a documented diagnosis of dyslipidemia or hyperlipidemia in their medical record, 62 (34%) were diagnosed as obese in
the EMR, 23 (13%) were diagnosed with diabetes, 50 (27%) were prescribed a statin and 28 (16%) were prescribed plant sterols and stanols. Not surprisingly, patients who had previously seen a lipid specialist were more likely to have been prescribed a statin (32%) whereas only 9% who had not seen a lipid specialist were prescribed a statin. Patients who had seen a lipid specialist were also more likely to be prescribed a plant sterol/stanol (18%) whereas only 3% of those who had not seen a lipid specialist had been prescribed a plant sterol/stanol. Table 1 summarizes additional characteristics of the chart review sample. There were no significant differences in the demographics of the participants that received the brochure and those who did not.

Of the 180 individuals who met the inclusion criteria, nine families had multiple individuals meeting criteria. Of the 171 surveys sent, 76 surveys were completed with a response rate of 44%. There were no statistically significant difference between the demographics of the responders and non-responders. Table 2 describes the demographics of the survey participants. Sixty three (83%) participants were female and 58 (76%) identified as the biological mother of the patient. Sixty (79%) participants were white and 73 (97%) identified as non-Hispanic.

Parental beliefs and attitudes

Parental beliefs and attitudes about FH and statin use were evaluated using four ten point Likert scale questions. Only 8% of participants felt it likely or very likely that their child would have a heart attack in the next 10 years (18% responded neutral and 74% unlikely or very unlikely); whereas 52% felt it likely or very likely that their child would have a heart attack by the age of 60 (26% responded neutral and 23% unlikely or very unlikely). When asked about concern for side effects caused by statins, 47% of participants were concerned or very concerned, 21% were neutral and 32% were unconcerned. There was more variability about the perceived
safety of treating children who have FH with statins. Only 29% agreed or strongly agreed that statin treatment was safe in children (34% responded neutral and 33% disagreed or strongly disagreed).

Genetic Counseling and Resources

Survey participants were asked about prior experiences with genetic counseling and testing as well as interest in future genetic counseling services. On chart review, 13 (7%) patients had seen a genetic counselor at CCHMC. Four (5%) survey participants reported speaking to a genetic counselor about FH. Upon further review, only one of these four had documentation confirming a prior visit with a genetic counselor and there was no documentation that FH was discussed. Five (7%) survey participants reported that their child had genetic testing for FH. However, based on the chart review, no genetic testing for FH was documented for any of the 180 individuals meeting inclusion criteria. Forty eight (63%) reported that they would consider speaking to a genetic counselor about FH and 37 (49%) would consider genetic testing for FH for their child. Participants were also asked about educational and support resources that are available to the public about FH. Only 1 (1%) reported having heard of the FH Foundation and 0% reported having heard of the CASCADE FH Registry. Twenty-five (33%) reported having looked up information about FH on the internet.

Family history and communication

Forty-two respondents (55%) reported never discussing FH with their siblings, parents or children. When asked on a scale of 1 (not informed) to 10 (very informed) how informed they felt about their family member’s health, 28 participants (37%) responded they felt informed.

Reported family history of FH, statin use, and cholesterol screening is summarized in Table 3. Twenty-eight respondents (37%) reported having a family history of FH and 41 (54%) reported a
family history of CHD. Seventy respondents (92%) reported a family history of the use of cholesterol lowering medications.

When asked if their child had been diagnosed with FH 37 participants (49%) responded yes, 13 (17%) responded no and 3 (4%) responded that their child had not been diagnosed with FH, but had been diagnosed with a different condition that caused increased cholesterol in the blood. Twenty-two (29%) responded that they did not know if their child had been diagnosed with FH. Of the 27 participants whose child had been diagnosed with FH in the EMR, only 17 (63%) responded that their child had a diagnosis.

*Disease knowledge*

Participant knowledge of FH was measured using six questions. Every participant correctly responded that if untreated, FH may lead to early heart disease and heart attacks. Due to the lack of variability in responses, this question was subsequently not used to calculate mean knowledge scores. Therefore, the mean knowledge score was calculated based on response to the remaining five knowledge questions. Overall, the participants who received the educational brochure had a higher mean knowledge score (2.8) than those who did not receive the brochure (2.5), however this difference was not statistically significant. Only 53% of those who received the brochure and 60% of those who did not correctly responded with LDL to the question “someone who has FH likely has high levels of what in their blood?” (*Table 4*).

Respondents who reported that a statin had been prescribed to their child had higher mean knowledge scores (3.1) than respondents whose child was not prescribed a statin (2.5) (*p* = 0.0267). Although not significant, mean knowledge scores were higher among participants who reported speaking with a HCP (3.0) than those who did not speak with a HCP (2.5) (*p* = 0.0606). Factors not associated with higher mean knowledge scores included a reported diagnosis of FH
having looked up information about FH on the internet \( p = 0.1811 \), reported annual household income of over $60,000 \( p = 0.1852 \), and a reported family history of hypercholesterolemia \( p = 0.3148 \).

**Discussion:**

Our results support previous findings that suggest FH is underdiagnosed. Additionally we found that the most recent guidelines for lipid screening from the American Academy of Pediatrics were not routinely followed, which likely contributed to the underdiagnosed of FH in this pediatric population. Those who reported that a healthcare provider had spoken to them about FH tended to have a higher mean knowledge score, which suggests that speaking with a healthcare provider may have increased subsequent knowledge and understanding of the disease. Lastly, the majority of the population had interest in genetic testing and genetic counseling suggesting that the genetics community has the opportunity to make an important impact on the improving care for families with FH. Taken together, there are several substantial opportunities for improvement in the care of patients with elevated cholesterol.

The prevalence of FH was calculated to be 0.04% compared to an expected international estimate of 0.2% or 1 in 500. In 2011 the American Academy of Pediatrics updated their recommendation to start universal cholesterol screening between the ages of 9-11 ("Expert panel on integrated guidelines for cardiovascular health and risk reduction in children and adolescents: summary report," 2011). Of the almost 90000 patients between the ages of 9 and 11 seen at CCHMC, less than 1000 (1%) received cholesterol screening. Given that some of the individuals seen at CCHMC received specialist care and not primary pediatric care, it is possible that routine cholesterol screening was facilitated elsewhere and that our reported prevalence is an underestimate of the number of individuals who received cholesterol screening. This data may
also suggest that the new cholesterol screening guidelines have not been universally adopted. The poor adherence to cholesterol screening guidelines observed may be reflective of cholesterol screening utilization in pediatric hospitals nationally.

The evidence for pediatric cholesterol screening in the 2011 guidelines focused on the correlation seen between lipoprotein disorders and the age of onset and severity of atherosclerosis in adolescents and adults. Additional evidence suggested that early identification and control of dyslipidemia in children with FH is associated with reduced subclinical evidence of atherosclerosis ("Expert panel on integrated guidelines for cardiovascular health and risk reduction in children and adolescents: summary report," 2011). A study by Rodenburg et al found that early initiation of statin therapy in children with FH may be beneficial in the prevention of atherosclerosis in adolescence (Rodenburg et al., 2007). Despite good evidence and formal guidelines recommending cholesterol screening in childhood, adherence is suboptimal. It does seem, however, that routine cholesterol screening may be more feasible in the pediatric setting than the adult setting. Pediatric patients are seen by a physician for routine well child care, which would offer an opportunity for cholesterol screening. The challenges limiting more widespread use of routine cholesterol screening are unclear but may include limited physician time, lack of awareness about the guidelines, parent’s concern regarding blood draw, or lack of perceived value of screening. Further study regarding barriers to implementing screening recommendations is needed.

Healthcare provider involvement in improving the diagnosis and management of FH

We found a non-significant trend between healthcare providers speaking to participants about FH and higher mean knowledge scores. However, receiving the educational brochure about FH was not associated with mean knowledge scores. These findings may suggest that
written information is not an optimal educational tool for this disease, that revisions to the educational brochure are needed, or that the brochures are more effective when partnered with physician interaction. It is also possible that while the educational brochure clearly communicated information, those reviewing the brochure did not view the information as important or relevant to their own health. Data from our study also suggested that individuals with FH are largely unaware of existing resources about FH. Only one person responded that they had previously heard of the FH Foundation and no one reported familiarity with the CASCADE FH registry. The FH Foundation is a nonprofit organization dedicated to raising awareness of FH through education, advocacy and research (http://thefhfoundation.org/). The lack of knowledge about existing resources places greater importance on the education healthcare providers’ provide about FH for patients and families.

Patients who had been seen by a lipid specialist were more likely to have been prescribed a statin or a plant sterol/stanol, have a diagnosis of FH listed in the EMR, and have been seen by a dietician. Nearly half of the survey participants were concerned or very concerned about the side effects of statins and over half did not agree that statins are safe for use in children. The data suggests that a lipid specialist is essential in coordinating comprehensive care for individuals with FH and educating patients about the risks associated with statin use. However, as pediatric lipid specialists may not be widely available to all individuals with FH, pediatricians should also be active in managing children with FH. Despite the expected improvement in diagnosis and management of FH associated with lipid specialist involvement, the disease remains underdiagnosed and under treated.

In addition, individuals seen by a lipid specialist were more likely to have a diagnosis of FH listed in the EMR. Only 47 of the 180 individuals meeting the inclusion criteria had a
diagnosis of FH documented in the EMR. While it is possible that some of the 180 individuals meeting the inclusion criteria would not meet clinical diagnostic criteria for FH, the numbers suggest that either provider documentation or diagnosis of disease is limited. There were 41 individuals who had multiple elevated LDL levels including one over 190 mg/dL and prior evaluation with a lipid specialist who did not have a diagnosis of FH in the EMR. In addition to limited EMR documentation of diagnosis, the data suggests that a lipid specialist is essential in coordinating comprehensive care for individuals with FH and educating patients about the risks associated with statin use.Pediatricians should also be active in managing children with FH, as pediatric lipid specialists may not be widely available to all individuals with FH. It is essential that pediatricians become more familiar with diagnosing and managing FH.

Need for increased awareness and support among FH population

Participant responses to the survey questions indicated limited understanding of inheritance and risks associated with FH. Other studies have also demonstrated a lack of knowledge about the genetic transmission of FH. Hollman et al found that 66% of their sample lacked knowledge about genetic transmission of FH and 79% lacked knowledge about the role family history plays in FH (Hollman et al., 2006). Additionally, the majority of respondents (55%) in our study reported never discussing FH with family members. Reasons for limited communication regarding FH with family members could be lack of awareness of the diagnosis and lack of understanding of the inheritance of FH. Only 37% of survey respondents reported a family history of FH. However, nearly all of the survey respondents reported a family history of high cholesterol, which suggests that parents/caregivers may not know that their child has a specific diagnosis of FH. Another study completed by van den Nieuwenhoff et al also found that people with FH did not inform most of their first degree relatives of their genetic risk and were
generally unwilling or unable to alert more distant relatives (van den Nieuwenhoff et al., 2007). There were many reasons stated for nondisclosure such as inadequate risk knowledge, risk denial and a perception that disclosing information was interfering. Given the autosomal dominant inheritance and near complete penetrance of the disease, the vast majority of participants should have a family history of FH thus making family communication extremely important in identifying other at-risk and affected individuals.

Participants were asked about their risk perceptions related to a diagnosis of FH. Only 39 (51%) responded that they felt it likely or very likely their child would have a heart attack by age 60. According to the National Human Genome Research Institute, the risk of heart attack by age 60 in men with FH could be as high as 85%. Another study surveyed 81 adults who had genetic testing to confirm their diagnosis of FH and found that they perceived their risk of cardiovascular disease to be relatively low (26.88 out of 100) (Claassen, Henneman, Kindt, Marteau, & Timmermans, 2010). These findings suggest a need for increased awareness regarding the risks associated with untreated high cholesterol.

Guidelines for the diagnosis and management of FH now incorporate genetic testing. Participants in our study expressed an interest in both genetic counseling and genetic testing for FH, however none of the families included in the study received either based on chart review. While any healthcare provider can order genetic testing, it can be optimized in a genetic counseling environment. Although genetic counselors have not widely been involved in the care of those with FH; their training and experience facilitating communication among families about their health would likely be beneficial in this population. Genetic counseling has been associated with increased patient knowledge of inheritance and natural history in other diseases (Lobb et al., 2004), as well as increased family communication and heightened adherence to
recommendations for at risk family members (Forrest, Burke, Bacic, & Amor, 2008). Overall, to improve the diagnosis of FH there must be improved cholesterol screening. All individuals with FH must have access to genetic testing and genetic counseling as individuals may benefit from an increased understanding of the disease. As discussed, genetic counselors have not been widely utilized in this population and may need additional education about FH.

Future Directions

The barriers to routine cholesterol screening in pediatrics remain unclear. It is possible that adherence may be limited in part by lack of awareness among pediatric care providers; however, further investigation is needed. In addition, further studies into the impact of genetic counseling and testing on disease-knowledge, family communication and uptake of cholesterol screening are needed, as are studies focused on the impact of education on the outcomes of FH are warranted. Individuals with FH represent a subset of the population with hyperlipidemia that is at incredibly high risk for early onset CHD. The FH population requires unique care that is likely best provided in coordination with pediatricians, lipid specialists and genetic counselors. Collaboration between these groups is likely to be important in making strides to improve FH diagnosis and management.

Conclusion

This study presents the first prevalence estimate for FH in a pediatric medical center and supports existing data regarding the under diagnosis of FH and the lack of adherence to routine cholesterol screening. Participants who indicated a healthcare provider had spoken to them about FH tended to be more knowledgeable about FH compared to those who had not. While the educational brochure did not have a significant impact on knowledge about FH, the brochure used in conjunction with provider discussion may be helpful. Given the important role healthcare
providers play in patient education, incorporating genetic counselors into the counseling and risk assessment of these patients and families may increase overall disease knowledge as well as adherence to cholesterol screening and genetic testing guidelines.
References:


Table 1: Characteristics of Patient Population

<table>
<thead>
<tr>
<th>Characteristics</th>
<th>Chart review Sample N=180 (%)</th>
<th>Responded to survey N=76 (%)</th>
<th>Did not respond to survey N=95* (%)</th>
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<tr>
<td>Age</td>
<td>Mean: 16.23 St Dev 3.56</td>
<td>Mean: 16.05 St Dev: 3.59</td>
<td>Mean: 16.58 St Dev: 3.43</td>
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<tr>
<td>Gender</td>
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<td>42 (55)</td>
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</tr>
<tr>
<td></td>
<td>Female 92 (51)</td>
<td>34 (45)</td>
<td>52 (55)</td>
</tr>
<tr>
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<td>2 (3)</td>
<td>2 (2)</td>
</tr>
<tr>
<td></td>
<td>Non-Hispanic 176 (98)</td>
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<td>21 (22)</td>
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<tr>
<td></td>
<td>Prescribed plant sterols/stanols 28 (16)</td>
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<td>Clinical diagnosis of FH</td>
<td>47 (26)</td>
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<td>18 (19)</td>
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<td>Seen by a lipid specialist</td>
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<td>Seen by dietician</td>
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<td>Age at first elevated LDL</td>
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<td>Mean: 11.59</td>
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<td>Mean: 26.9</td>
<td>Mean: 28.28</td>
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<tr>
<td>Additional diagnoses</td>
<td></td>
<td></td>
<td></td>
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<td>Dyslipidemia or hyperlipidemia</td>
<td>105 (58)</td>
<td>47 (62)</td>
<td>63 (66)</td>
</tr>
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<td>Obesity</td>
<td>62 (34)</td>
<td>31 (41)</td>
<td>25 (26)</td>
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<td>Hypertension</td>
<td>39 (22)</td>
<td>15 (20)</td>
<td>22 (23)</td>
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<td>Diabetes</td>
<td>23 (13)</td>
<td>10 (13)</td>
<td>13 (14)</td>
</tr>
<tr>
<td>None</td>
<td>29 (16)</td>
<td>11 (14)</td>
<td>14 (15)</td>
</tr>
</tbody>
</table>

*9 individuals were not sent surveys as they had siblings in the patient sample
(There were a total of 180 individuals in the chart review and only 171 sent a survey)
<table>
<thead>
<tr>
<th>Characteristics</th>
<th>N=76 (%)</th>
<th>Brochure N=34 (%)</th>
<th>No brochure N=42 (%)</th>
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<tbody>
<tr>
<td><strong>Age</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Mean age: 45.9</td>
<td></td>
<td>Mean age: 47.5</td>
<td>Mean age: 44.7</td>
</tr>
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<td>StDev: 7.6</td>
<td></td>
<td>StDev: 7.0</td>
<td>StDev: 7.9</td>
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<tr>
<td><strong>Gender</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Male</td>
<td>13 (17)</td>
<td>4 (12)</td>
<td>9 (21)</td>
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<tr>
<td>Female</td>
<td>63 (83)</td>
<td>30 (88)</td>
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</tr>
<tr>
<td>Non-Hispanic</td>
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<td>34 (100)</td>
<td>40 (95)</td>
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<td></td>
<td></td>
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<tr>
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<td>Black or African American</td>
<td>9 (12)</td>
<td>3 (9)</td>
<td>6 (14)</td>
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<td>Asian</td>
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<td>2 (6)</td>
<td>4 (10)</td>
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<td>American Indian or Alaskan Native</td>
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<td>1 (3)</td>
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<td><strong>Education</strong></td>
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<td>High school degree of equivalent (e.g., GED)</td>
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<td>4 (10)</td>
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<td>Some college but no degree</td>
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<td><strong>Household Income</strong></td>
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<td>Under $20,000</td>
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<td>$20,001 to $40,000</td>
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<td>1 (3)</td>
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<td><strong>Relationship to child</strong></td>
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<tr>
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<td>58 (76)</td>
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<td>28 (67)</td>
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<tr>
<td>Biological Father</td>
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<td>3 (9)</td>
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<tr>
<td>Adoptive Mother</td>
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<tr>
<td>Adoptive Father</td>
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<td>0 (0)</td>
<td>1 (2)</td>
</tr>
<tr>
<td>Step parent</td>
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<td>1 (3)</td>
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<td>Other</td>
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<tr>
<td>Days per week spent with child</td>
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<td>(N=32)</td>
<td>(N=39)</td>
</tr>
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<td>0 days</td>
<td>5 (7)</td>
<td>3 (9)</td>
<td>2 (5)</td>
</tr>
<tr>
<td>1-2 days</td>
<td>5 (7)</td>
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<td>3 (7)</td>
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<tr>
<td>3-4 days</td>
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<td>2 (6)</td>
<td>0 (0)</td>
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<tr>
<td>5-6 days</td>
<td>4 (5)</td>
<td>3 (9)</td>
<td>1 (2)</td>
</tr>
<tr>
<td>7 days</td>
<td>58 (78)</td>
<td>22 (69)</td>
<td>36 (86)</td>
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Table 3: Reported Family History of Disease, Medication use, and Screening

<table>
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<tr>
<th>Characteristics</th>
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<tr>
<td>Any reported history of FH</td>
<td>28 (37)</td>
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<tr>
<td>First degree affected</td>
<td>26 (34)</td>
</tr>
<tr>
<td>Second degree affected</td>
<td>23 (30)</td>
</tr>
<tr>
<td>Family history of high cholesterol</td>
<td></td>
</tr>
<tr>
<td>Any reported history of high cholesterol</td>
<td>75 (99)</td>
</tr>
<tr>
<td>First degree affected</td>
<td>66 (87)</td>
</tr>
<tr>
<td>Second degree affected</td>
<td>66 (87)</td>
</tr>
<tr>
<td>Family history of coronary heart disease</td>
<td></td>
</tr>
<tr>
<td>Any reported history of CHD</td>
<td>41 (54)</td>
</tr>
<tr>
<td>First degree affected</td>
<td>5 (7)</td>
</tr>
<tr>
<td>Second degree affected</td>
<td>39 (51)</td>
</tr>
<tr>
<td>Family history of heart attack before age 60</td>
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</tr>
<tr>
<td>Any reported history of heart attack</td>
<td>42 (55)</td>
</tr>
<tr>
<td>First degree affected</td>
<td>7 (9)</td>
</tr>
<tr>
<td>Second degree affected</td>
<td>37 (49)</td>
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<tr>
<td>Family history of taking cholesterol lowering medicine such as statins</td>
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<tr>
<td>Any reported history of statin use</td>
<td>70 (92)</td>
</tr>
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<td>First degree</td>
<td>57 (85)</td>
</tr>
<tr>
<td>Second degree</td>
<td>60 (79)</td>
</tr>
<tr>
<td>Family history of xanthomas</td>
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<tr>
<td>Any reported history of xanthomas</td>
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</tr>
<tr>
<td>First degree affected</td>
<td>7 (9)</td>
</tr>
<tr>
<td>Second degree affected</td>
<td>7 (9)</td>
</tr>
<tr>
<td>Family history of cholesterol screening</td>
<td></td>
</tr>
<tr>
<td>Any reported history of screening</td>
<td>70 (92)</td>
</tr>
<tr>
<td>First degree</td>
<td>66 (87)</td>
</tr>
<tr>
<td>Second degree</td>
<td>60 (79)</td>
</tr>
<tr>
<td>Family history of genetic testing for FH</td>
<td></td>
</tr>
<tr>
<td>Any reported history of genetic testing</td>
<td>4 (5)</td>
</tr>
<tr>
<td>First degree</td>
<td>2 (3)</td>
</tr>
<tr>
<td>Second degree</td>
<td>3 (4)</td>
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Table 4: Impact of Educational Brochure on Survey Knowledge Score

<table>
<thead>
<tr>
<th>Survey FH Knowledge Questions (correct response)</th>
<th>Brochure N=34* (%)</th>
<th>No brochure N=42* (%)</th>
<th>P-value</th>
</tr>
</thead>
<tbody>
<tr>
<td>FH can be passed from parent to child. (True)</td>
<td>32/32 (100)</td>
<td>39/42 (93)</td>
<td>P= 0.123</td>
</tr>
<tr>
<td>If one parent has the most common type of FH what is the chance that their son or daughter will also have FH? (50%)</td>
<td>25/32 (78)</td>
<td>31/42 (74)</td>
<td>P= 0.668</td>
</tr>
<tr>
<td>If someone has the most common type of FH what is the chance that their brother or sister will also have FH? (50%)</td>
<td>20/31 (65)</td>
<td>22/42 (52)</td>
<td>P= 0.300</td>
</tr>
<tr>
<td>How common is FH? (1 in 500 individuals)</td>
<td>19/30 (63)</td>
<td>15/42 (36)</td>
<td>P= 0.021**</td>
</tr>
<tr>
<td>If untreated, FH may lead to early heart disease and heart attacks. (True)</td>
<td>34/34 (100)</td>
<td>42/42 (100)</td>
<td>n/a</td>
</tr>
<tr>
<td>Someone who has FH likely has high levels of what in their blood? (LDL (low-density lipoprotein) cholesterol)</td>
<td>17/32 (53)</td>
<td>25/42 (60)</td>
<td>P= 0.582</td>
</tr>
</tbody>
</table>

*number of potential responders

**P-value is statistically significant (P<0.05)
Figure 1: Study Procedures

Patients (age 2-21) seen between 1/1/2011 and 7/15/2013 with LDL cholesterol above 155mg/dL
N= 1262

Assessed for Eligibility N=845

Chart review completed on N=180

Survey sent to N=171

Received brochure N=86

No brochure N=85

Returned survey N= 42

Returned survey N= 34

Excluded total: N= 417
- Incomplete lab values available

Excluded Total: N= 665
- Diagnosis of Lupus, polycystic ovary syndrome, nephrotic syndrome, solid organ or bone marrow transplant N=348
- Anyone who had not seen a lipid specialist AND did not have an LDL above 190 N=317

Excluded Total: N= 9
- Multiple family members met criteria
FH Diagnosis
N=47

1

16

29

1

41

34

LDL over 190mg/dL
N=105

LDL over 155mg/dL and saw a lipid specialist
N=144

Figure 2: Inclusion Criteria
Appendix A: Survey

Please answer the following questions about your **CHILD** whose name is listed on the envelope:

1. What is the age of your child? ____________

2. Does your child see any of the following doctors at least once a year? (please circle all that apply)
   a. Cardiologist (doctor who specializes in diagnosing or treating heart disease)
   b. Lipid specialist (doctor who specializes in treating problems with cholesterol, or the fats present in blood)
   c. Pediatrician/primary care provider (PCP)
   d. Unsure

3. Has your child been diagnosed with familial hypercholesterolemia?
   a. Yes
   b. No
   c. No, they have been diagnosed with a different condition that causes increased cholesterol in the blood
   d. I don’t know

4. Has a dietician ever talked to you or your child about eating a diet low in cholesterol and fat?
   a. Yes
   b. No
   c. I don’t know

The following questions ask about your thoughts on health risks and treatment related to familial hypercholesterolemia. This portion of the survey is not to evaluate your knowledge.

5. What is the likelihood of your child having a heart attack in the next ten years?
   Not likely 1 2 3 4 5 6 7 8 9 10
   Very likely

6. What is the likelihood of your child having a heart attack by age 60?
   Not likely 1 2 3 4 5 6 7 8 9 10
   Very likely
7. How concerned are you about side effects from statin medication?
   Not concerned  Very concerned
   1  2  3  4  5  6  7  8  9  10

8. Which side effects are known to be associated with statins? Please circle all that apply.
   a. Upset stomach including diarrhea, constipation or nausea
   b. Muscle aches and pains
   c. Liver disease
   d. Memory loss
   e. Mental confusion
   f. Diabetes
   g. Other
   h. None of the above
   i. I don’t know

9. Treating children who have familial hypercholesterolemia with statins is safe.
   Disagree  Agree
   1  2  3  4  5  6  7  8  9  10

10. Medications are NOT effective at treating familial hypercholesterolemia.
    Disagree  Agree
    1  2  3  4  5  6  7  8  9  10

The following questions will ask about familial hypercholesterolemia and genetics, please answer the questions to the best of your ability.

11. Familial hypercholesterolemia CAN be passed from parent to child.
    a. True
    b. False

12. If one parent has the most common type of familial hypercholesterolemia what is the chance that their son or daughter will also have familial hypercholesterolemia?
    a. 0%
    b. 25%
    c. 50%
    d. 75%
    e. 100%
13. If someone has the most common type of familial hypercholesterolemia what is the chance that their brother or sister will also have familial hypercholesterolemia?
   a. 0%
   b. 25%
   c. 50%
   d. 75%
   e. 100%

14. How common is familial hypercholesterolemia?
   a. 1 in 100 individuals
   b. 1 in 500 individuals
   c. 1 in 1000 individuals
   d. 1 in 10000 individuals

15. If untreated, familial hypercholesterolemia may lead to early heart disease and heart attacks.
   a. True
   b. False

16. Someone who has familial hypercholesterolemia likely has high levels of what in their blood?
   a. LDL (low-density lipoprotein) cholesterol
   b. HDL (high-density lipoprotein) cholesterol
   c. Triglycerides (a type of fat in your blood)
   d. None of the above

The following four questions ask about your experiences and thoughts on genetic counseling and genetic testing:

17. Would you consider having your child undergo genetic testing for familial hypercholesterolemia?
   a. Yes
   b. No
   c. My child already had genetic testing for familial hypercholesterolemia
   d. Unsure

18. Has anyone in your family other than the child whose name is listed on the envelope had genetic testing for familial hypercholesterolemia?
   a. Yes
   b. No
   c. Unsure

19. Have you ever spoken to a genetic counselor about familial hypercholesterolemia?
   a. Yes
   b. No
c. Unsure

20. Would you consider speaking to a genetic counselor about familial hypercholesterolemia?
   a. Yes
   b. No
   c. I have already spoken to a genetic counselor about familial hypercholesterolemia
   d. Unsure

The following questions ask about possible sources of information you may have heard of or used to learn more about familial hypercholesterolemia:

21. Has a health care provider ever discussed familial hypercholesterolemia with you?
   a. Yes
   b. No
   c. I don’t know

22. Did you use the pamphlet from the FH Foundation that was included with the questionnaire to answer any of the questions?
   a. Yes
   b. No
   c. I did not receive a pamphlet

23. Have you ever heard of the FH Foundation?
   a. Yes
   b. No
   c. Unsure

24. Have you ever heard of the CASCADE FH Registry through the FH Foundation?
   a. Yes
   b. No
   c. Unsure

25. Have you ever looked up information about familial hypercholesterolemia on the internet?
   a. Yes
   b. No
   c. I don’t know
The following questions are about your family’s health history:

26. Please answer the following questions about your child’s biological family. Biological family members are those that are related to the child by blood. Please check all that apply:

Example) Who in your family has ever been diagnosed with familial hypercholesterolemia… your child’s mother, your child’s father, your child’s aunt… etc.

<table>
<thead>
<tr>
<th>Who in your family has ever been diagnosed with familial hypercholesterolemia?</th>
<th>Your child's Mother</th>
<th>Your child's Father</th>
<th>Your child's Siblings</th>
<th>Your child's Aunt (Mother's side)</th>
<th>Your child's Uncle (Mother's side)</th>
<th>Your child's Aunt (Father's side)</th>
<th>Your child's Uncle (Father's side)</th>
<th>Your child's Grandparent (Mother's side)</th>
<th>Your child's Grandparent (Father's side)</th>
<th>None</th>
<th>I don't know</th>
</tr>
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<tbody>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
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<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Who in your family has ever been diagnosed with high cholesterol?</th>
<th>Your child's Mother</th>
<th>Your child's Father</th>
<th>Your child's Siblings</th>
<th>Your child's Aunt (Mother's side)</th>
<th>Your child's Uncle (Mother's side)</th>
<th>Your child's Aunt (Father's side)</th>
<th>Your child's Uncle (Father's side)</th>
<th>Your child's Grandparent (Mother's side)</th>
<th>Your child's Grandparent (Father's side)</th>
<th>None</th>
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<tr>
<th>Who in your family has been diagnosed with coronary heart disease (narrowing of blood vessels)?</th>
<th>Your child's Mother</th>
<th>Your child's Father</th>
<th>Your child's Siblings</th>
<th>Your child's Aunt (Mother's side)</th>
<th>Your child's Uncle (Mother's side)</th>
<th>Your child's Aunt (Father's side)</th>
<th>Your child's Uncle (Father's side)</th>
<th>Your child's Grandparent (Mother's side)</th>
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<th>Who in your family has had a heart attack before the age of 60?</th>
<th>Your child's Mother</th>
<th>Your child's Father</th>
<th>Your child's Siblings</th>
<th>Your child's Aunt (Mother's side)</th>
<th>Your child's Uncle (Mother's side)</th>
<th>Your child's Aunt (Father's side)</th>
<th>Your child's Uncle (Father's side)</th>
<th>Your child's Grandparent (Mother's side)</th>
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<th>Who in your family takes cholesterol lowering medication such as statins (Lipitor, Zocor, Mevacor, Pravachol)?</th>
<th>Your child's Mother</th>
<th>Your child's Father</th>
<th>Your child's Siblings</th>
<th>Your child's Aunt (Mother's side)</th>
<th>Your child's Uncle (Mother's side)</th>
<th>Your child's Aunt (Father's side)</th>
<th>Your child's Uncle (Father's side)</th>
<th>Your child's Grandparent (Mother's side)</th>
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<th>Who in your family has fatty skin deposits called xanthomas over part of their hands, elbows, knees, ankles and around the cornea of the eye (corneal arcus)?</th>
<th>Your child's Mother</th>
<th>Your child's Father</th>
<th>Your child's Siblings</th>
<th>Your child's Aunt (Mother's side)</th>
<th>Your child's Uncle (Mother's side)</th>
<th>Your child's Aunt (Father's side)</th>
<th>Your child's Uncle (Father's side)</th>
<th>Your child's Grandparent (Mother's side)</th>
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32
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<tr>
<th>Who in your family has had cholesterol screening?</th>
<th>None</th>
<th>I don't know</th>
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</table>

| Who in your family has had genetic testing for familial hypercholesterolemia? | None | I don't know |
27. On average, how often do you discuss familial hypercholesterolemia with your immediate family members? Immediate family members include your siblings, parents, and children.
   a. Never
   b. Once a year
   c. Once a month
   d. Once a week

28. How informed do you feel regarding the health of your biological family members?

<table>
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<tr>
<th>Not Informed</th>
<th>Very Informed</th>
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<td>1</td>
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<td>3</td>
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<td>9</td>
<td>2</td>
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<tr>
<td>10</td>
<td>1</td>
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Please answer the following questions about **YOURSELF**:

29. What is your age? ____________

30. What is your gender?
   a. Female
   b. Male

31. What is your ethnicity?
   a. Hispanic (Mexican, Puerto Rican, Cuban, or other Spanish background)
   b. Non-Hispanic
   c. Don’t know

32. What is your race?
   a. White
   b. Black or African-American
   c. American Indian or Alaskan Native
   d. Asian
   e. Native Hawaiian or other Pacific Islander
   f. None of the above

33. What is the highest degree or level of school you have completed?
   a. Less than a high school degree
   b. High school degree or equivalent (e.g., GED)
   c. Some college but no degree
   d. Associate degree
   e. Bachelor’s degree
   f. Graduate degree
34. What is your total household annual income before taxes?
   a. Under $20,000
   b. $20,001 to $40,000
   c. $40,001 to $60,000
   d. $60,001 to $80,000
   e. $80,001 to $100,000
   f. More than $100,000

35. What is your marital status?
   a. Single, never married
   b. Married or domestic partnership
   c. Widowed
   d. Divorced
   e. Separated

36. Have you personally had cholesterol screening done?
   a. Yes, I have high cholesterol
   b. Yes, I have low or normal cholesterol
   c. Yes, I had high cholesterol that is treated with medication
   d. No
   e. Unsure

37. What is your relationship to your child whose name is listed on the envelope?
   a. Biological Father (related by blood)
   b. Biological Mother (related by blood)
   c. Adoptive Mother
   d. Adoptive Father
   e. Step parent
   f. Legal Guardian
   g. Other (specify) ____________

38. On average, how many days per week do you spend with your child whose name is listed on the envelope?
   a. 0 days
   b. 1-2 days
   c. 3-4 days
   d. 5-6 days
   e. 7 days
39. How often do you attend doctor’s appointments with your child?
   a. Always
   b. Very often
   c. Sometimes
   d. Rarely
   e. Never

40. How many children do you have? __________

41. How many of your children have had cholesterol screening? __________

42. What are some reasons one or more of your children have had cholesterol screening? (check all that apply)
   a. Family history of high cholesterol or heart disease
   b. Routine screening
   c. Weight concerns (obesity)
   d. Other reason not listed
   e. Unsure
   f. My children have never had cholesterol screening

Thank you for participating in this research study. Your feedback is greatly appreciated