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Parental Attitudes, Beliefs, and Perceptions about Genetic Testing for FAP and Colorectal Cancer Surveillance in Minors

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Abstract

Familial adenomatous polyposis (FAP) is the second most common hereditary colorectal cancer syndrome and confers a nearly 100% lifetime risk of developing colorectal cancer. Understanding factors that facilitate and inhibit genetic testing and cancer surveillance in children who are members of families affected by FAP will better equip clinicians to clarify misunderstandings and facilitate appropriate care. The aims of this study were to examine parental attitudes and beliefs regarding endoscopic surveillance and genetic testing in minors at risk for developing FAP. This cross-sectional study includes analyses of qualitative and quantitative interview data collected from parents of children with or at risk for FAP. This report includes data from 28 parents with a total of 51 biological children between 10–17 years of age. The parents had a clinical and/or genetic diagnosis of FAP. Most commonly reported facilitators included provider recommendation (surveillance) and personalized medical management (genetic testing). Most commonly reported barriers included lack of provider recommendation (surveillance) and cost (genetic testing).

Keywords

FAP; Children; Parental attitudes; Endoscopic surveillance; Genetic testing; *APC* mutation

Introduction

Familial adenomatous polyposis (FAP), the second most common hereditary colorectal cancer syndrome, is one of the few cancer susceptibility conditions that confers an almost 100% lifetime risk of developing cancer unless surgical intervention is performed. Reported incidence rates vary from approximately 1:8,000 to 1:13,500 (Bisgaard et al. 1994; Fearnhead et al. 2001; Gryfe 2006). It is an autosomal dominant condition most often caused by a mutation in the adenomatous polyposis coli (*APC*) gene located on chromosome 5q21 (Kinzler et al. 1991; Nishisho et al. 1991). Genetic testing for *APC* mutations is clinically available and is used, in combination with family history, to personalize prevention and early detection strategies. Colorectal surveillance for at-risk individuals (i.e., those with an identified familial *APC* mutation; those who meet clinical criteria for FAP; or those who have a biological first-degree relative affected with FAP) includes annual flexible sigmoidoscopy or colonoscopy to identify and remove polyps (Winawer et al. 2003). For genetically at-risk individuals who do not undergo colectomy, the mean age of cancer diagnosis is 39 years (Range: 34–43 years) (Burt and Jasperson 2008). For individuals with FAP, the average age of onset of colon polyps is 16 years (Petersen et al. 1991), although it is common for children to have clinically detectable polyps as early as age 10 years. Once the number of polyps is too great to be safely observed or removed, preventive surgery is strongly recommended, preferably before age 20 (Al-Sukhni et al. 2008; National Comprehensive Cancer Network Colorectal Screening 2009). Delayed recognition and intervention can worsen the prognosis (Augustyn and Wallerstein 2009; Croner et al. 2005).

Often because of complex social problems and individuals fear of surgery, delays in treatment occur in patients with FAP and colorectal cancer (Mills et al. 1997). Furthermore, numerous barriers to colorectal cancer screening have been observed, including physical discomfort with screening protocols, and patient embarrassment about the procedures and concern about discrimination and insurance coverage (Nichols et al. 2009). Given that FAP is an autosomal dominant condition, there is a 50% probability that each offspring of an affected individual will have the familial mutation. Consensus-approved guidelines observe that increasingly, *APC* mutation testing initially is offered during adolescence (American Society of Clinical Oncology 2003; National Comprehensive Cancer Network Colorectal Screening 2009). If a child of a mutation carrier is found not to have inherited the deleterious mutation, he or she does not need routine enhanced colorectal surveillance. However, if the child has a deleterious *APC* mutation, the recommendation is for initial colonoscopy at age 10–15 years. If a family mutation cannot be determined, or if the child is not tested and there is a known familial mutation, the recommendation is for flexible sigmoidoscopy or colonoscopy beginning at age 10–15 years and then annually until age 24. If no polyps are found, the surveillance intervals increase with age (National Comprehensive Cancer Network Colorectal Screening 2009).

Genetic Testing of Minors

Several studies and organizations have examined issues regarding genetic testing of minors who have high prevalence rates of familial conditions, such as colorectal cancer, that do not develop until adulthood. There is consensus that genetic testing for disease susceptibility is indicated when there is direct medical benefit through medical intervention or preventive measures (Kastrinos et al. 2007; Michie et al. 2001). The American Society of Human

Genetics (ASHG), American College of Medical Genetics (ACMG), and American Academy of Pediatrics Committee on Bioethics convened expert committees to explore issues related to genetic testing of minors; consensus statements recommend that genetic testing of minors for disease susceptibility is appropriate only if preventive interventions exist (American Academy of Pediatrics Committee on Bioethics 2001; ASHG/ACMG 1995). Therefore, genetic testing for FAP in minors should be considered when manifestations occur in the pediatric period, and when proven prevention strategies are in place.

One particular barrier to genetic testing in minors, not only for FAP but also for other hereditary conditions, is the concern about adverse psychological reactions (ASHG/ACMG 1995; Borry et al. 2007; Michie et al. 2001). Examples include concerns about a negative impact on the child's self-esteem and self-worth if he or she were found to have a genetic alteration (Douma et al. 2008; Michie et al. 2001;), and concern about a change in parent-child bonding (Fanos 1997). Another possible barrier to genetic testing uptake for FAP and other conditions is that parents tend to view their own and their children's genetic status as transient (Michie et al. 2003). Consequently, those parents who hold this belief contend that genetic test results could not accurately predict future disease risk.

Several benefits of genetic testing for cancer susceptibility and cancer surveillance in children have been identified. Reduced anxiety (Fanos 1997) and avoidance of unnecessary surveillance (ASHG/ACMG 1995) are two potential benefits, especially for those children without a mutation. In addition, knowledge of one's mutation status will allow for personalized medical management (Li et al. 1992); and a clearer understanding of risk perception and perceived threat of illness will allow the family to begin planning for the future (Codori et al. 1999; Michie et al. 1996). At least one study has suggested that the benefit of knowing is better than the uncertainty of not knowing, although knowledge about increased susceptibility to develop cancer may be anxiety-provoking (Baty et al. 2006).

Conceptual Framework of the Present Study

Two validated social cognitive theories, the Health Belief Model (Rosenstock et al. 1988) and the Theory of Planned Behavior (Ajzen 1991) have been used extensively to explain determinants of preventive health behaviors in a wide variety of domains, including cancer screening and genetic testing for both childhood-and adult-onset hereditary conditions (Gooding et al. 2006; Price 2003). These two theories were used to guide the development of our interview questions and analyses because they help provide an understanding of factors that influence decision-making about genetic testing and colorectal cancer surveillance in members of FAP families. The Health Belief Model provides a framework for explaining preventive health behaviors such as *APC* mutation testing and surveillance for colorectal cancer or precursor lesions (adenomatous polyps). The central tenet underlying the model is that health behavior is determined by personal beliefs or perceptions about a particular disease and available strategies to decrease its likelihood of occurring. According to the Health Belief Model, the likelihood of performing a certain behavior increases when a person (such as the parent of a minor) believes: 1) s/he is susceptible to the health conditions, the condition is serious, and the behavior is beneficial; 2) the barriers are few and perceptions about benefits outweigh the barriers; 3) s/he has the self-efficacy to perform the behavior (belief that one is capable of performing behavior[s] to attain specific goals), and 4) s/he responds to cues to action. The Theory of Planned Behavior posits that intention is an important predictor of behavior and that differences in the intention-behavior relationship may be attributable to differences in attitudes, subjective norms [social pressure by, and actions of significant others (i.e., family members and health care providers) and perceived behavioral control [i.e., perception of the ease or difficulty of performing the behavior (self-efficacy)].

Purpose of the Study—We assessed constructs from both of these explanatory theoretical models of health behavior, including attitudes, beliefs and perceptions of the FAP-affected parent regarding surveillance and genetic testing of their at-risk minor children, as well as their child(ren)'s receipt of genetic testing results and risk-appropriate surveillance recommendations. In addition, survey measures included behavioral intentions of parents regarding their children's use of provider recommendations for genetic testing and colorectal endoscopic surveillance and the actual use of these health services. Parents' self-efficacy perceptions (confidence) regarding their ability to have their child(ren) screened and/or undergo genetic testing, as well as sociodemographic parameters, were also considered.

To our knowledge, no previous studies have specifically examined parents' cognitions about perceived barriers concerning colorectal cancer surveillance and *APC* genetic testing of their minor children. While other studies have evaluated perceived barriers and facilitators, as well as other issues related to cancer screening tests in a variety of at-risk populations (Li et al. 1992), our study addressed beliefs and attitudes related to genetic testing and surveillance of minors as perceived by the affected parents of children at increased risk for developing FAP. Identification of barriers and facilitators, as well as other important cognitions related to utilization of *APC* gene mutation testing and colorectal cancer surveillance in minors may enhance personalized risk communications and informed decision making in clinical and other settings.

Methods

Study Design

This cross-sectional study assessed parents' cognitions and health behaviors relevant to genetic testing and colorectal cancer screening for minors with or at risk for FAP.

Sample

Participants were identified through the Huntsman Cancer Institute's Familial Colorectal Cancer Registry at the University of Utah and by expanding participants' pedigrees to include other at-risk relatives. Eligibility criteria for participation in this study included: (1) parent had a clinical and/or genetic diagnosis of FAP, and (2) parent had at least one biological child between the ages of 10 and 17 years.

Procedures

All procedures were approved by the Huntsman Cancer Institute Clinical Cancer Investigations Committee, the University of Utah Institutional Review Board, and the Resource for Genetic and Epidemiologic Research Review Committee. Potential participants were invited by mail to participate in the study (Kinney et al. 2007). Written informed consent was obtained from each participant after the nature of the study had been fully explained to them, and prior to a computer-assisted telephone semi-structured interview conducted by a trained interviewer. During the interviews, information was collected on demographics and on clinical and psychosocial factors, including risk perceptions and perceptions about colorectal cancer surveillance. Interviews related to this analysis were conducted between March 2000 and July 2004.

Measures

Sociodemographics

Sociodemographic factors that were assessed included: race/ethnicity, household income, health insurance status, and marital status.

Clinical Factors

Genetic Counseling and Testing and Surveillance Behaviors—The participants were asked about previous experiences with genetic counseling and/or *APC* mutation testing (yes, they had undergone genetic counseling and/or pursued genetic testing; vs. no, they had not done so). They also were asked whether their child(ren) had ever undergone *APC* mutation testing or received a diagnosis of FAP (yes vs. no). Other items assessed included the number of polyps found in the child(ren) and the age of each child at the time of *APC* mutation testing. Utilization of endoscopic surveillance and/or prior genetic counseling for the at-risk minor child(ren) were assessed based on consensus-approved guidelines in use at the time (National Comprehensive Cancer Network Colorectal Screening 2003). We asked participants if they or their children had ever undergone sigmoidoscopy/or colonoscopy (yes vs. no vs. don't know).

Behavioral Intentions—We assessed the participants' future plans regarding colorectal cancer surveillance and *APC* mutation testing for their at-risk child(ren) with two items assessing when/if the participant planned on having the child(ren) screened with sigmoidoscopy/colonoscopy and when/if the participant planned on their child(ren) having *APC* mutation testing.

Psychosocial Factors

Provider Recommendation—Parents were asked whether they ever received advice from a physician or other health care provider about genetic testing and/or regular sigmoidoscopy or colonoscopy for their minor child(ren) (yes vs. no).

Behavioral Intentions—Another interview item assessed the parents' behavioral intentions to have their minor children undergo *APC* mutation testing. Two questions assessed actual behaviors including 1) whether the child(ren) ever had *APC* mutation testing and 2) whether the child(ren) has ever undergone sigmoidoscopy or colonoscopy.

Self-efficacy—A single question assessed the parent's confidence that they could follow through with the recommended schedule of sigmoidoscopy or colonoscopy for their minor child(ren) on a 1–5 scale (1 = not at all confident, to 5 = very confident). This question was only asked of those parent's who had reported their child(ren) had previously undergone lower endoscopy screening.

Risk Notification—The participants were asked if their at-risk child(ren) was aware of a possible FAP diagnosis and/or risks of the disorder (yes vs. no); and if so, who communicated that information to the child and the child's age at the time.

Open-Ended Questions about FAP

Participants were given an opportunity to respond to eight open-ended questions about surveillance and genetic testing for their minor child(ren). They were asked to describe their beliefs about colorectal screening for children whose family members have FAP, and to provide comments or concerns regarding colorectal cancer surveillance or genetic testing as relevant to their child(ren). These questions assessed parental attitudes, and perceived potential barriers and benefits to genetic testing. Another open-ended question gave the participants an opportunity to impart what they had been told by their health care provider regarding colorectal cancer surveillance for their minor child(ren).

When applicable, the participants also described what the surveillance procedure was like for their child(ren). Parents shared their beliefs regarding the age at which at-risk children should begin colorectal cancer screening procedures. Interview questions assessed parental

beliefs about a) reasons why their child(ren) should, or should not, be tested for the *APC* gene mutation; and b) what they thought children under 18 years old should be told about genetic test results. Interview questions are available upon request.

Data Analysis

Analysis of closed-ended questions included calculating descriptive statistics such as frequency counts, proportions, and measures of central tendency using SAS software (version 9.1.3, Cary, NC). Participants' responses to the open-ended questions were analyzed using standard content analysis techniques described by Krippendorff (1980). A distinguishing feature of content analytic approaches to analyze qualitative data is the use of a consistent set of codes to delineate data segments containing similar material. In our study, we initially developed a preliminary coding system based on a literature review. Next, participants' responses to open-ended survey questions were independently reviewed by two of the authors (JC, FL) to identify themes that emerged from the data. After these were verified with a third author (AK), the final set of themes was determined. Then, JC and FL independently coded and rated all participant responses; and AK resolved discrepancies. Kappa statistics were calculated to determine inter-rater reliability of the qualitative data coding scheme (Landis and Koch 1977; Viera and Garrett 2005). Upon completion of the coding, the codes were summarized with frequency counts and percentages according to procedures outlined by Miles and Huberman (1994).

Results

Sample Sociodemographics

Table 1 summarizes the sociodemographic and clinical variables for the 28 parent participants. The majority were white and non-Latino (89%), were married/living as married (79%), had health insurance (82%), and earned greater than \$50,000/year (54%). Table 2 summarizes the demographic and clinical variables regarding the participants' 51 minor children, whose mean age was 14 years (Range: 10–17 years). Sixty-four percent (18/28) of participants reported that they had partial or full insurance coverage for colorectal cancer screening tests for their children. Of the remaining participants, three had no coverage for these services for their children; six did not know whether or not their insurance covered these services; and one parent did not answer the question. However, we observed a different trend for participant insurance coverage for genetic counseling/testing services for their children. Only five participants reported partial or full insurance coverage for these services, while 9 had no such coverage. Almost half (13/28) reported that they did not know whether or not genetic counseling/testing was a covered service. One participant did not respond to this question.

Clinical Factors

Genetic Counseling and Testing and Surveillance Behavior—Half of the twenty-eight parent participants reported previous genetic counseling experiences. These same individuals reported that they had undergone *APC* mutation testing. Among the fourteen parents tested, twelve had an *APC* mutation, and two had an indeterminate result. We were able to validate self-reported genetic test results with clinical reports for twenty of the parents for whom medical records were available for our review. Of the participants' whose medical records were available for our review, there was perfect agreement between self-reported and documented genetic testing results. Seventeen of the parent participants reported having utilized endoscopy previously.

The same background information was obtained for the minor children of the participants: of the 51 at-risk children, 41% (21/51) had genetic testing. Medical records of the children

were not available for our review. The age at which testing was conducted varied from 1 to 16 years (mean=10 years; median=11 years). Children were not considered to be candidates for genetic testing if their affected parent had not yet pursued genetic testing for themselves. Per parent report, 20% (10/51) of the children had been diagnosed with FAP based on positive *APC* genetic test results. In our study sample, only 33% (17/51) of the at-risk children previously had undergone lower bowel endoscopy. The seventeen children who had undergone lower bowel endoscopy included both those who had tested positive for the known familial *APC* mutation (10 children) and those who had not had genetic testing (7 children). Of the seventeen children who had undergone lower bowel endoscopy, eight had been diagnosed with polyps of the colon. Of the eight children diagnosed with polyps of the colon, three were found to have an *APC* mutation; the remaining five children had not undergone genetic testing for the familial *APC* mutation.

Behavioral and Psychosocial Factors

Behavioral Intentions—When the parents were asked when they planned on their child(ren) having their initial or next sigmoidoscopy or colonoscopy, the most common response was ‘within the next year’ (9/17). This question was only asked if participants had reported they were planning to have their child(ren) screened, or that their child(ren) already had colorectal screening via lower endoscopy. The majority of the children (42/51) did not have a scheduled endoscopy appointment as of the time of the interview. When asked about plans for genetic testing of their minor child(ren), ten parents had already tested their children; four were committed to having their child(ren) tested; three thought they should probably have their child(ren) tested; one parent was considering having their child(ren) undergo genetic testing someday; five parents including two who had indeterminant results) stated they had no plans for having their child(ren) tested; and two responded that it should be the child’s decision once he or she turns 18. Three parents did not know what their plans were regarding genetic testing for their child(ren).

Self-efficacy—Of the nine participants who intended to have their child(ren) screened within the next year, eight reported they were very confident that they could follow through with regular screening. One participant indicated being not at all confident about proceeding with surveillance recommendations.

Provider Recommendation—When asked if a health care provider had recommended regular endoscopic surveillance for polyps or colon cancer, seventeen responded “yes,” while eleven responded “no.” About half (28/51) of the children’s parents had been given recommendations about screening intervals, while 43% (22/51) had not received provider suggestions. One of the parents was not aware of whether her child’s healthcare provider had made recommendations about timing of surveillance. Nineteen parent participants reported receiving recommendations concerning genetic testing for children under age 18.

Risk Notification—The participants also were asked if their child(ren) were aware of their at-risk status. Per parent report, 71% (36/51) of the children were aware of their FAP risk. For the purposes of this study, children who tested negative for the familial *APC* mutation or whose parent had an indeterminant result were not included in the following analysis. Thirty-six of the 38 at-risk children were aware of their increased risk for FAP; eight of the ten children who tested positive for an *APC* mutation were aware of their risk, and 28 of the 30 at-risk children who did not have *APC* mutation testing were informed of their increased risk based on their family history of FAP. In most of these cases (33/36) a parent was the individual who shared the child’s risk with her or him. We did not ask the participants whether the affected, unaffected, or both parents were the one(s) to inform the child of his/her risk. The remaining three children were informed of the risk or diagnosis of FAP by

another, unspecified individual. The mean age at which the child was informed of his or her genetic risk was 10 years (Range: 7–16 years).

Open-ended Questions—Kappa statistics were calculated to assess inter-rater coding agreement for each of the eight open-ended questions (Viera and Garrett 2005) and they ranged from 0.63–1.0—representing substantial (0.61–0.80) to almost perfect (0.81–0.99) agreement. Several themes emerged regarding the parents' beliefs and perceptions surrounding screening and genetic testing of their minor children for FAP. Sixteen of the 28 participants reported the belief that screening at-risk children who have a family member with FAP is important. Seven parents expressed opinions about when screening should begin, ranging from age 10 to the mid-twenties. Table 3 includes examples of themes from parents' open-ended responses.

Most of the participants (22/28) reported communication with their primary care provider regarding this topic; however, the provider recommendation varied regarding both the age that surveillance should begin for at-risk children and the intervals at which screening should be done. The most commonly reported recommendation was that screening should be done once per year (5/22). Three participants received a recommendation from a health care provider to start screening between ages 12 and 17, while another three were advised to wait until the child reached the age of 18. Two participants reported receiving provider recommendations to not have a child or additional children due to a family history of FAP.

Slightly over half of the parents (15/28) believed that minor children with a parent who has FAP should have regular lower bowel endoscopy (54%; 15/28). Three stated that they did not believe children under age 18 should be screened by sigmoidoscopy and/or colonoscopy. Almost one third (9/28) asserted that endoscopy in this age group should depend on the unique circumstances/presentation of each family and on provider recommendations. We asked parent participants to comment about their child's perceptions of the surveillance procedure. Most (10/17) responded that overall, the child had no problems with the procedure. However, one parent reported that their child was fearful and/or anxious about the procedure, another parent reported adverse psychological consequences in the child, and two parents perceived that the procedure and/or prep were painful and uncomfortable for their child.

Many participants (17/28) reported that they believed there was no reason why their at-risk child(ren) should not be tested for the *APC* gene. For the remaining parents who identified reasons for not testing, discrimination (both insurance and employment) (3/28) and cost (3/28) were the most commonly cited reasons. For instance, some participants thought an insurance company might become suspicious due to increased frequency of colorectal cancer surveillance testing, and therefore would deny coverage or raise premiums. Two parents reported that children under 18 should not be tested for FAP because "the child's genes would change as he/she got older." Another parent stated that "children should not be tested because they are too young and the test is not conclusive." The remaining two participants did not respond to the question.

Parent participants were asked about their reasons for having their child(ren) tested for mutations in the *APC* gene, and a few provided more than one response. Early detection and personalized management were the most common reasons for having child(ren) tested for a known familial mutation within the *APC* gene (11/28). Other reasons included knowledge (8/28); reducing anxiety and uncertainty (5/28); family history (5/28); and making decisions about increasing surveillance or stopping unnecessary screening (2/28). The final question focused on whether children under the age of 18 should be told the result of their test. A majority of participants (20/28) believed that children under age 18 should receive their test

result, while a few (6/28) stated that results should be shared with children under age 18 on a case-by-case basis. Two parents reported that they believed results should not be disclosed to children less than 18 years of age.

Discussion

This is among the first studies that utilize qualitative and quantitative data collection methods which are guided by theory in order to investigate parent perspectives concerning screening and genetic testing in minors at risk for FAP. Because individuals with FAP can develop polyps as early as age 10, it is important for clinicians to understand parents' perceived barriers, benefits, and facilitators to colorectal cancer surveillance and genetic testing in their children. An important benefit of *APC* mutation testing for children is that colorectal cancer surveillance can be individualized according to the child's genetic test results. Effective screening and preventive measures are available for those who test positive, while those who test negative for a known familial mutation are spared the discomfort and anxiety associated with regular screening.

Age for Testing and Surveillance

The average age at which known mutation testing was initiated in our study participants' children was 10, but ranged from 1 to 16 years. This is noteworthy because of the considerable age range of the children who underwent genetic testing. For many of the participants' children, *APC* testing was done at very young ages, much earlier than the recommended surveillance guidelines. For others, initial testing was done several years later than the recommended age for beginning colorectal surveillance (i.e., age 10–12 years). These findings suggest a departure from standards of care for risk assessment and medical management (American Society of Clinical Oncology 2003). Furthermore, some of the participants' children were tested before the age of 10, before they had reached the legal age for becoming an active participant in the decision-making process (i.e., to formally provide assent if over the age of 8 years).

Perceived Barriers to Testing

In our study, one commonly perceived barrier was lack of health care provider recommendation. This is consistent with previous observations that provider recommendations positively correlated with colorectal cancer screening and genetic testing uptake; and that completion of screening increased if providers addressed areas of concern such as family history, mutation status, and test-specific barriers (Gennarelli et al. 2005; Janz et al. 2007). Participants in another study expressed feelings of empowerment when the direct assessment of their cancer risk was followed by a variety of options upon which they could act (Sweet et al. 2003). These findings underscore the importance of familial/hereditary cancer risk assessment, as well as the need for effective risk notification and behavioral counseling that address patient concerns and specific barriers to genetic testing and relevant preventive health care services (Janz et al. 2007).

Although our study was not designed to assess insurance barriers in depth, it is recognized that third party coverage affects mutation testing and surveillance. Issues and concerns about insurance coverage for genetic testing and colorectal cancer screening also have been documented in previous studies (Penziner et al. 2008; Rawl et al. 2002). Our participants' concern about insurance company denial of coverage or premium increases, due to increased frequency of colorectal cancer surveillance testing, is consistent with a recent study of adolescents at risk for Huntington's disease and FAP (Duncan et al. 2008). Furthermore, this is a fear even for those with adequate coverage (Duncan et al. 2008). Fear of genetic discrimination also is expressed by people who are at increased risk for many different

presymptomatic genetic conditions (Duncan et al. 2008; Hall and Rich 2000). We conducted our interviews prior to the passage of the Genetic Information Nondiscrimination Act (GINA) in 2007, and it is possible that concerns about genetic discrimination could have changed and/or lessened since GINA was enacted.

Reasons for Testing

Parents' Reasons for Testing their Children—Early detection and the ability to individualize medical management were the most frequently reported reasons for having child(ren) tested for a known familial *APC* gene mutation. The benefits of personalized medical management are best recognized if individuals at risk for FAP undergo *APC* mutation testing, and if they also adhere to guidelines for risk-appropriate screening. This benefit has been substantiated in other areas, including genomic medicine, for a variety of conditions such as diabetes and hemochromatosis (Khoury et al. 2007).

Some participants indicated their belief that germline mutations change with age and the *APC* test results were not conclusive before age 18. This belief has been previously reported (Michie et al. 1996). Genetic counselors can help dispel misconceptions and increase knowledge about these and other beliefs. Correct information may facilitate informed decisions about genetic testing and promote risk-appropriate cancer screening for at-risk children. By becoming aware of accurate information about *APC* mutation testing, a parent may choose to utilize genetic testing, and subsequent screening, for high-risk child(ren) at an appropriate age and with the long-term goal of preventing colorectal cancer.

Study Limitations and Research Recommendations—A relatively small sample size and racial/ethnic representation primarily limited to non-Latino Caucasians, were among the limitations of this study. Another limitation is that the study sample was recruited primarily through a high-risk registry at one cancer center. These factors could have introduced a potential selection bias. Consequently, our findings may not be generalizable to other settings. Furthermore, qualitative data are not intended to be generalized to the population.

Our study was designed to assess cognitions and behaviors among FAP patients and their at-risk relatives. The analyses presented here were based on responses of parents with a clinical or genetic diagnosis of FAP, who had affected or at-risk biological children between 10–17 years of age and who may have not pursued genetic testing for themselves. It is conceivable that unaffected parents may differ from affected parents with regards to their attitudes and beliefs about FAP, genetic testing and colorectal cancer screening. Any or all of these factors may limit the generalizability of the results. Because many counseling sessions involve both affected and unaffected parents, and in view of the scant literature on unaffected parents' concerns about genetic testing and colorectal cancer screening, further studies are needed to examine the attitudes, beliefs, and perceptions of both affected and unaffected parents regarding genetic testing for FAP and colorectal cancer surveillance in minors. An additional limitation was that our study's conceptual framework did not consider a stress and coping perspective which has relevance in this context (Gooding et al. 2006). More research is needed in diverse populations regarding both cognitive and emotional factors in order to gain a more in-depth understanding of parental beliefs that impact the decision-making process regarding colorectal cancer screening and genetic testing for children at risk for developing FAP.

Practice Implications

A major role for genetic counselors is to enhance informed decision-making by clearly communicating risk and other educational information to high-risk adults and children, as

well as to their primary care providers and to medical specialists involved in their care. In addition, psychosocial counseling is often beneficial in helping patients to cope with the genetic information and to make decisions about genetic testing and screening (Codori et al. 1999). While it is beyond the genetic counselor's control to ensure that the patient retains all of the information discussed in a session, it is within the counselor's control to be knowledgeable about common misconceptions in at-risk populations (e.g., a belief that a child's gene will alter as an adult). Awareness of perceived barriers and facilitators to colorectal cancer screening and genetic testing uptake in affected parents of minors at risk for FAP enhances a genetic counselor's ability to help with decisions that are consistent with their clients' values and preferences.

Because manifestations of FAP may begin in childhood, it is recommended that at-risk children be screened for colon polyps. However, parental beliefs and perceptions about genetic counseling and screening may negatively impact adherence to the recommended screening guidelines for their high-risk children. The diversity of parental attitudes and beliefs in our study population suggest the complexity inherent in consideration of genetic testing and colorectal surveillance of minors at risk for FAP. To successfully implement effective genetic counseling and cancer screening programs for minors, it is important for practitioners to provide interventions that address the concerns of both parents and children.

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Table 1

Demographic and Clinical Variables of Parent Participants

Variable	N=28 (n)	(%)
Education		
High school or less	7	25.0
Technical/Vocational School	3	10.7
Some College/College graduate	18	64.3
Religious affiliation		
Church of Jesus Christ of Latter Day Saints	12	42.9
Other	16	57.1
Race/Ethnicity		
White, not Latino	25	89.3
White, Latino	3	10.7
Marital Status		
Single	6	21.4
Married/Living as married	22	78.6
Income		
< \$50,000	13	46.4
≥ \$50,000	15	53.6
Insurance Coverage		
Yes	23	82.1
No	1	3.6
Other	3	10.7
Don't know	1	3.6
Age Diagnosed with FAP		
≤ 18 years	4	14.3
19–25	4	14.3
26–31	3	10.7
32–40	6	21.4
41–50	3	10.7
Don't know	8	28.6
Had APC testing		
Yes	14	50.0
No	14	50.0
Genetic test result		
APC-mutation Positive	12	85.7
Indeterminate	2	14.3
Prior Colorectal Endoscopy		
Yes	17	60.7
No	10	35.7
Don't know	1	3.6

Table 2

Demographic and Clinical Data for Participants' Minor Children (Ages 10–17 years)

Age of children (n=51)	(n)	(%)
10 years–12 years	17	33%
13 years–15 years	16	31%
16 years–17 years	18	35%
*Mean=13.96 years SD 2.37		
Number of children who had genetic testing ^a (n=51)	21	41%
Age at genetic testing (n=21)		
≤ 5 years	2	9%
6 years–10 years	7	33%
11 years–15 years	10	48%
16 years–17 years	1	5%
Do not know	1	5%
*Mean=10.65 years SD 3.44		
Number of children who had ever been diagnosed with polyps of the colon (n=51)	8	16%
Number of children who had been diagnosed with FAP (n=51)	10	20%
Number of children who were aware of his/her risk for FAP (n=36) *does not include those who tested negative or indeterminate		
APC mutation positive	8	22%
At-risk based on family history, no genetic testing	28	78%
Age at which at-risk children were told his/her risk for FAP (n=36)		
≤ 5 years	2	5%
6 years–10 years	14	39%
11 years–15 years	19	53%
16 years–17 years	1	3%
*Mean=10.43 years SD=3.40		

^a Per parent report

Table 3**Examples of Themes from Participants' Qualitative Responses**

Themes and Categories	Quotes
Benefits to screening/genetic testing	
○ Early detection/Personalized management	"The gene will always be there. It will never change. If they could know at an early age, then they could make some intelligent decisions and / or have a little bit of peace of mind."
○ Knowledge	"So that they know their risk of cancer or they know whether or not they have to have the annual exam."
○ Family history	"Cause I had it."
Barriers to screening/genetic testing	
○ No reason not to	"I can't think of one."
○ Discrimination	"Only if it would affect their future chance of employment, or future chance of insurance."
○ Cost	"None whatsoever—unless it was cost prohibited."
Attitudes towards screening (sigmoidoscopy/colonoscopy)	
○ Important/good idea	"I think as far as I am concerned and as far as I know it should be done."
○ Opinion about age to start screening	"I think it should be done once per year, starting around age 11."
○ Physician recommendation	"If suggested by the doctor."
○ No problems with the procedure	"He was fine with it. It didn't bother him."
Attitudes towards genetic testing	
○ Necessary/important	"Important that they have it done."
○ Physician recommendation	"No [concerns].....you're just kind of going off what the doctor thinks or recommends."