Impact of Familial Adenomatous Polyposis: An Emerging Adult Perspective

Master’s Thesis

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Gayun Chan-Smutko, Advisor

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in
Genetic Counseling

by
Nina D’Orlando

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ABSTRACT

Impact of Familial Adenomatous Polyposis: An Emerging Adult Perspective

A thesis presented to the Graduate Program in Genetic Counseling

Graduate School of Arts and Sciences
Brandeis University
Waltham, Massachusetts

By Nina D’Orlando

Familial Adenomatous Polyposis (FAP) is a rare, highly penetrant, genetic cancer predisposition syndrome typically requiring prophylactic removal of the colon. This study is an early look at the specific impact of FAP in the dynamic period of life between the ages of 18 to 25 termed “emerging adulthood.” Participants recruited through the Hereditary Colon Cancer Foundation were asked to complete an online survey designed to collect both quantitative and qualitative data that focused on areas of life typically important to emerging adults: education, career planning, relationships, and family planning. Additionally, overall adjustment was assessed via the previously validated Psychological Adaptation Scale (PAS) and participants were asked about the time of diagnosis and experiences with genetic counseling. Participants (n=33) expressed varying degrees of impact across all domains, although the PAS revealed that on average participants were relatively well adjusted to their diagnosis. Specific challenges highlighted by participants included a lack of informed providers, difficulties due to missed school or work, struggles in relationships with friends and romantic partners, and a desire to avoid passing this condition on to children. These results suggest that a longitudinal care model
of periodic follow-up with a multidisciplinary care team of genetics, gastroenterology, surgery, and mental health professionals may be beneficial for emerging adults with FAP.
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Introduction

Familial Adenomatous Polyposis

Classified as a highly penetrant cancer syndrome, Familial Adenomatous Polyposis (FAP) is a rare genetic condition characterized by the growth of polyps in the colon and rectum that, without intervention, will almost inevitably progress to malignancy. Polyp growth in affected individuals typically begins during the teen years and increases with age with the potential for hundreds to thousands to develop. Without intervention in the form of a prophylactic colectomy, the average age of a colon cancer diagnosis is 39 years. Additionally, individuals with FAP are at an increased risk for a number of extracolonic manifestations including small bowel polyps and cancer, pancreatic cancer, thyroid cancer or benign thyroid disease, CNS tumors, pediatric hepatoblastoma, gastric polyps and cancer, osteomas, dental abnormalities, CHRPE, cutaneous lesions, desmoid tumors, and adrenal masses (National Library of Medicine, 2016 & Jaspersen and Burt, 2014). In addition to the health burden of this condition, potential increased psychosocial distress or anxiety in those living with FAP threatens a widespread impact on a person’s life (Grubman & Barzi, 2015 & Andrews et al. 2007).

Emerging Adulthood

In the year 2000, Jeffrey Jensen Arnett proposed the new developmental concept of “emerging adulthood,” which he defined as the distinct period from age 18 to 25, characterized by great change and exploration of one’s world (Arnett, 2000). Having shed the dependency of adolescence but not yet having the full responsibilities of adulthood, individuals in this age group are uniquely positioned to explore and build the foundations of love and relationships, career,
and worldviews that will likely guide much of the rest of their lives (Arnett, 2000). Because this is a distinct period of growth and change in the lives of all emerging adults, the needs of these individuals who are also dealing with a hereditary cancer syndrome like FAP may differ significantly from the needs of older individuals with FAP. In outlining “what we don’t know” about adolescents and young adults who are living with colorectal cancer due to FAP and highlighting the additional challenges and concerns of the group, Grubman and Barzi call for more research and collaboration regarding age-specific medical and psychosocial care and standard treatment protocols for young individuals with FAP-related cancers (Grubman & Barzi, 2015). In his review of the psychological, social, and behavioral issues for young adults with cancer, Brad Zebrack points out that while individuals diagnosed with cancer during adolescence and young adulthood face the challenge of integrating their cancer experience into this unique developmental period, which can be distressing for some, most of these individuals function well physically and psychologically. He notes that some may integrate their cancer diagnosis and survivorship into their identity, showing resiliency and believing that overcoming this adversity has made them a better or stronger person (Zebrack, 2011).

**Physical and Psychosocial Impacts of FAP**

To the best of our knowledge, no studies have looked specifically at FAP in emerging adults. However, a number of studies have examined the impact of FAP on the lives of younger individuals. In 2006 and in 2007, Andrews *et al.* published two successive studies done in Australia about the impact of FAP on young adults aged 18-35, the first regarding attitudes toward genetic testing and support and information needs, and the second regarding quality of life outcomes (Andrews *et al.*, 2006 & 2007). Sixty-two percent of participants reported moderate/high levels of need for support in coping with at least one of the following: anxiety
related to their children developing FAP, fears of developing cancer, and uncertainty about the impact of FAP (Andrews et al., 2006). Reported impacts on quality of life in the areas of physical functioning, body image, sexual impact and negative affect differed significantly based on type of prophylactic colectomy an individual had undergone or if they had not undergone any surgery (Andrews et al. 2007). Participants who were single reported higher levels of distress and showed significantly higher FAP avoidance responses (Andrews et al. 2007).

All participants (n=11) in a similar qualitative study in Australia in 2009 reported no significant impact on their education, although clinically affected individuals reported impacts on employment, social activities, long-term relationships and marriage, attitudes toward having children and family relationships, while clinically unaffected individuals reported no or limited impact of FAP in these same areas (Mireskandari et al., 2009). In 2011, Douma et al. compared the health-related quality of life (HRQOL) between patients with FAP, at-risk individuals, and proven non-carriers (family members of participants with FAP). Participants were an average age of 43.6 years, and although HRQOL among participants was similar to that of the general Dutch population, FAP patients who had undergone surgery reported significantly poorer physical functioning and disease-related social functioning and more issues with defecation and body image than those FAP patients who had not undergone surgery (Douma et al., 2011). Around 40% of participants reported that FAP had impacted their working life and 25% that it limited recreational activities (Douma et al., 2011). Further support that HRQOL for individuals with FAP is similar to the general population comes from Fritzell et al. who found that participants seemed to have found successful coping mechanisms and ways of living with FAP as almost all reported “living a normal life” (Fritzell et al., 2010).
Testing Process

Given that the impact of living with a hereditary cancer condition like FAP can create challenges for those affected, the impact of the testing process itself may sometimes be overlooked. Interviews with ten individuals in Australia aged 12-25 who had undergone predictive genetic testing for FAP (aged 10-17 at the time of testing) uncovered five themes regarding testing: (1) the significance of the test in the patient’s life, (2) the patient’s lack of involvement in the decision making process, (3) the limited understanding of the patient at the time of testing, (4) the blood draw at the first visit, and (5) the group testing of family members (Duncan et al., 2010). These themes allowed for eight recommendations for ways in which the predictive genetic testing process for young individuals with FAP might be improved, including such ideas as providing pre-test counseling and information with more time to digest the information before the blood draw, or giving the opportunity for young individuals to see their genetic health professional alone or be tested separately from siblings to allow for more individual focus, support, and respect for their autonomy (Duncan et al., 2010).

Preimplantation genetic diagnosis (PGD) presents an opportunity to avoid having to make the decision about when to have a child tested and potentially protect children from the struggle of inheriting a genetic condition like FAP. In a 2014 study comparing the awareness and acceptance of PGD among patients with four different hereditary cancer syndromes, patients with FAP were among those with the highest percent in favor of considering using PGD (Rich et al., 2014). Andrews et al., also assessed the feelings of individuals with FAP surrounding genetic testing, including preimplantation genetic diagnosis (PGD), prenatal testing, possible termination, and testing for children. Seventy-five percent of the participants reported that they would consider prenatal testing for FAP, and 61.4% would consider PGD. However, only 20.9%
reported that they would consider termination if prenatal testing found that the fetus carried a FAP-related mutation (Andrews et al., 2006). In 1996, Whitelaw et al. obtained similar results in London finding that 64.5% of 62 interviewed individuals affected with FAP reported that they would request prenatal testing, but only 24% reported they would opt for termination if the fetus was affected (Whitelaw et al., 1996).

**Genetic Counseling and Information**

In exploring the impact of FAP on many aspects of participants’ lives, much of the reviewed literature concludes that the impact of FAP changes from the time of testing (often as a child or adolescent) throughout a person’s life warranting continued follow up with genetic counselors (Andrews et al., 2006 & Andrews et al. 2007 & Duncan et al., 2010 & Fritzell et al., 2010 & Mireskandari et al., 2009). Additionally, in 2015, a review of patients at the University of Texas MD Anderson Cancer Center who were under 35 years old, were diagnosed with colorectal cancer (CRC), and had undergone genetic counseling at the center between 2009-2013 revealed that 34.7% had a hereditary cancer syndrome (Mork et al., 2015). Notably, of the 16 patients with FAP or AFAP, 10 had apparent de novo mutations. These results support a genetic counseling referral for all patients diagnosed with CRC at 35 years or younger, regardless of family history, especially considering the high de novo mutation rate observed in individuals with FAP or AFAP (Mork et al., 2015).

In terms of informational support, Andrews et al. found that the majority of patients (94.3%) were “somewhat” or “very much” satisfied with the information about FAP that they had been given (Andrews et al., 2006). In a focus group study done in Sweden by Fritzell et al., most participants reported unmet needs in the areas of: continuity of care by healthcare providers
that are knowledgeable about FAP, practical and psychosocial support related to prophylactic surgery, FAP educational programs, and meetings with others with FAP (Fritzell et al., 2010)

**Rationale for Present Study**

As outlined above, emerging adulthood is a period of life distinctly characterized by change, instability and exploration. Examination of the existing literature related to the impacts of FAP reveals that the emerging adult population has yet to be specifically studied on its own. The goals of this study are to gain insight into the potential widespread impacts of FAP in the context of emerging adulthood and to better describe and characterize the needs of this group so that they can be most appropriately supported by their healthcare providers throughout their lives. We aim to assess how emerging adults view the impacts of a diagnosis of FAP on their education, career, relationships, family planning, and overall adjustment including coping, self-esteem, social integration, and spiritual well-being.
Methods

Study Design

This study was conducted via a confidential online survey created using Qualtrics, an online survey software supported by Brandeis. The survey method was chosen for its easy access to maximize the number of respondents. It consisted of multiple choice, Likert scale and open-ended (text entry) questions, which were utilized to gather both quantitative and qualitative data. Allowing participants to express and elaborate on their insights and opinions beyond those that were encompassed within the provided question choices allowed for a more complete picture of the overall impact of FAP on this group. This study was reviewed and approved by the Brandeis University Institutional Review Board.

Sample and Recruitment

Participants for this study were recruited through the Hereditary Colon Cancer Foundation (http://www.hcctakesguts.org/) via an email blast to all members, as well as postings about the study on associated Facebook groups. Participants were also encouraged to share the survey with others who they knew met inclusion criteria. The inclusion criteria were that participants must be between the ages of 18-25 and have a self-reported clinical or genetic diagnosis of FAP. Participants who did not have a diagnosis of FAP or did not fall within our desired age range were excluded from this study as our aim was to look specifically at the impact of FAP in the “emerging adult” population.
Data Collection Procedure

Individuals interested in participating in the study were directed to a confidential online survey. Participation in this study was voluntary and participants were free to skip any questions they did not wish to answer or to leave the survey at any time. Participants had the option to save responses and complete the survey at a later date while the survey remained open.

The survey was developed and administered through Qualtrics and was open for a total of four weeks (January 14th, 2017 – February 14th, 2017). Participants were asked questions regarding their personal and family history of FAP and the impact of a personal diagnosis of FAP on education, career, relationships with family, friends, and romantic partners, and family planning decisions. The previously developed and validated Psychological Adaptation Scale (PAS) was administered within the survey to assess overall adaptation to a diagnosis of FAP (Biesecker et al., 2013). The survey also asked questions related to the diagnostic process for an individual and about any experiences participants may have had with genetic counseling. Participants who completed the survey were offered the option to enter a raffle for a chance to receive one of three $50 gift cards to Amazon.com as a token of appreciation for their time and insights. If they wished to do so, they were directed to a separate, unlinked Qualtrics survey at the end of the study survey where they could enter their email address for the raffle.

Data Analysis

Quantitative data was analyzed using SPSS, a statistical software program. Analyses via descriptive statistics, t-tests, chi-square analyses, ANOVA, and correlations were conducted as appropriate. We analyzed the perceived impacts of FAP on education, career planning, relationships, family planning, and overall adjustment to living with FAP. For each of these domains, we assessed impacts in terms of the following variables: current age (individual or
grouped), age at diagnosis, time since diagnosis, colectomy status, age at colectomy, time from
diagnosis to surgery, time since surgery, whether a participant has had a diagnosis of cancer,
whether the participant is the only person in their family affected with FAP, or whether the
participant has had a family member pass away due to FAP. Participant age was analyzed
according to individual age as well as by grouping participants by age (18-20 \(n=7\), 21-23
\(n=10\), 24-25 \(n=16\)). Our survey questions about the age at diagnosis and age at colectomy
utilized grouped ages, so to determine the approximate time from diagnosis to surgery, the time
since surgery, and the time since diagnosis, the median of each group was used to calculate an
approximate number of years. This number was then used in analyses. To characterize themes
that emerged from participant responses regarding the impacts of a personal diagnosis of FAP,
qualitative data resulting from the multiple open-ended questions was manually coded and
analyzed.
Results

Survey Respondents

A total of 33 participants meeting criteria for participation completed the survey in its entirety. Three participants only partially completed the survey and these responses were included in the analysis of open-ended questions. Of the 33 respondents completing the survey, 26 (78.8%) were female, 6 (18.2%) were male, and 1 individual (3%) reported that they identify as non-binary. The average age of the participants was 22 years, with the majority being 25 years old (36.4%).

Ten participants (30.3%) reported that they are the only person in their family with FAP, and all others reported at least one family member with FAP. Five participants (15.2%) reported that they have had either colorectal or thyroid cancer as a result of FAP, with 4 being diagnosed between the ages of 18 to 20 years. Just over half (51.5%) of the participants reported that a family member affected with FAP has had cancer, and 36.1% of all participants reported that a family member has passed away due to cancer caused by FAP.

Diagnosis

All participants were diagnosed with FAP between the ages of 9 and 25, with most being diagnosed between the ages of 12 and 20 (66.6%). The greatest number of participants (42.4%) were first diagnosed clinically based on their symptoms and then also had genetic testing, and almost as many were diagnosed by presymptomatic genetic testing based on their family history (39.4%). Of the 13 individuals who underwent presymptomatic genetic testing, 9 (69.2%) reported that they had little say in the decision to have testing and 2 (15.4%) that they had no say
at all. Only 2 individuals (15.4%) reported that they made the decision mostly themselves (Appendix A).

All but one participant (97%) reported that they have been diagnosed with colon polyps, with most reporting polyp diagnoses between the ages of 12 and 20 and the majority (63.6%) reporting that they have had more than 100 polyps. Twenty-one participants (63.6%) have had a colectomy, and most (80.9%) reported having the procedure between the ages of 15 and 23 years. An ileal pouch anal anastomosis (IPAA), also known as a J-Pouch, was the most commonly reported procedure (47.6%).

Eleven participants (33.3%) reported that they received only a little information about FAP at the time of their diagnosis, 10 (30.3%) received a moderate amount, 7 (21.2%) received a lot, and 2 (6.1%) received a great deal of information. Most felt that the amount of information they received at initial diagnosis was extremely (30.3%) or somewhat (27.3%) appropriate, and that the type of information was somewhat (36.4%) or extremely (24.2%) appropriate.

Open-ended questions about the time of their diagnosis revealed a few themes concerning a lack of realistic and long-term information, a lack of answers to questions, and wishes for more informed doctors. Many participants also described strong emotional responses to the diagnosis, as evidenced by selected quotes below, followed by current age (years), gender identification, participant identification number, and age (years) at diagnosis (dx):

“I was devastated. That was one thing I never wanted from my dad.” – 25 y.o. Female (p19), dx: 18-20

“i wished there was more information like pamphlets or sites dedicated to FAP and how it will affect my life. it all got a little overwhelming and felt like it was all happening way to quickly. having my first colonoscopy was very unpleasant and felt thee should have been more info about what to expect that was more accurate or real [sic].” – 25 y.o. Female (plnc.1), dx: 18-20

“I was prepared for the diagnosis. My mother is very well versed with FAP, having lived with it for so long. […]so I felt like we were more knowedgable than our GI doctor, who doesn't have any other FAP patients at all. […][sic].” – 23 y.o. Female (p6), dx: 18-20

“I didn't want to believe it and had been hoping I didn't have it. So it took me into a dark mental state and I cried hysterically because I didn't know how to deal with it.” – 24 y.o. Female (p14), dx: 21-23
On wishes for differences at the time of diagnosis:

“I wish I didn't have to have my bowel removed immediately after being diagnosed.” – 21 y.o. Female (p2), dx: 18-20

“Referred immediately to a Dr that had extensive knowledge and experience with FAP [sic].” – 20 y.o. Male (p8), dx: 15-17

“I was informally diagnosed by a general doctor, not fap specialist, who had only seen fap in a text book. I wish I had went to a gastro doctor who knew what this disease is and didn't insult me by reading the diagnosis from a text book [sic].” – 25 y.o. Female (p23), dx: 18-20

“perhaps give a little time to digest information instead of shoving the info immediately in your face [sic].” – 19 y.o. Male (p27), dx: 12-14

“wished my doctors knew a bit more and were a more empathetic [sic].” – 25 y.o. Female (pInc.1), dx 18-20

**Education**

Education level completed or currently in progress ranged from some high school to a master’s degree, with most participants having completed or currently working on a bachelor’s degree (33.3%). The most participants (36.4%, n=12) reported that their diagnosis of FAP has had both positive and negative impacts on their education, while 27.3% (n=9) reported that their diagnosis of FAP has had a negative impact on their education, and 24.2% (n=8) reported that their diagnosis of FAP has not impacted their education in any way. Perceived impacts of FAP on education were not significantly different for any of the variables assessed.

Prevalent among open-ended responses regarding the impact of FAP on one’s education were comments about missed school or the need to repeat some school, impacts on plans for school, and impacts on grades or focus. For example, on missing school:

“[…] I was still recovering from surgery when school came back into session, my last semester in community college before transferring to a university. I struggled at first. My mom had to push me to and from class in a wheelchair and I often had to leave class while my body re-taught me the difference between how gas and a bowel movement feels. I missed a lot of class, all things considered, and I struggled to absorb the material. I was worried I'd fail and my transfer would be denied, but I passed all of my classes in the end. Nothing lower than a B. I don't feel like my education has been impacted beyond that. I don't struggle any more than I used to, don't have any new problems. It feels entirely neutral [sic].” – 23 y.o. Female (p6)

“i definitely feel that it was a huge distraction, I missed a lot of high school and had to put off college for a whole semester while i was in the hospital. But it also made me want to pursue my current career [sic].” – 19 y.o Male (p27)
On impacts on plans for school:

“My diagnosis did not necessarily impact my education but the effects did. In my senior year of high school I underwent an ileostomy and reversal. I could not carry my textbooks around and I needed a pass to use the restroom whenever I needed. When I got to college I had to live near home so that I could go to all my doctors appointments. I could not attend the school I truly wanted to in Virginia until I was more stable [sic].” – 22 y.o. Female (p17)

“[…] My studies were being done part time & as such i ditched it copletely, realising that focussing in myself was key to getting through the cancer. I found myself leaving the world of standard education & moving to an introspective sort of learning, where i learnt about my body as well as my mind. I found time to ask wat i want from life, how i wanna get there & more. So while traditional learning failed during the cancer, the wisdom i walk away with feels like the greatest treasure i could have walked away with. To value life is something to be proud of. In todays wold so many people forget what a miracle just breathing is [sic].” – 25 y.o. Male (p22)

Finally, on impacts on grades and focus:

“I had to retake a year of college due to my operations and got lower grades than expected due to stress” – 25 y.o. Female (p13)

“[…] It was bad cause when I first was diagnosed/had surgery I was getting sick alot and always in and out of the hospital so I would miss school, also I was tired alo so I never felt like doing school work [sic].” – 18 y.o. Female (p31)

Career Planning

Thirteen participants (39.4%) felt that their diagnosis of FAP influenced their choice of career. Ten participants (30.3%) disagreed, feeling that FAP has not had an influence. Equal numbers of participants reported that they have either not yet decided on a career (15.2%) or that they do not know if FAP has impacted their choice of career (15.2%). Twelve participants (36.4%) reported that FAP impacts their current career on a regular basis, 10 (30.3%) reported no current impact, 9 (27.3%) are not currently working, and 2 (6.1%) do not know if FAP impacts their career.

Open-ended responses related to how FAP has influenced choice of career demonstrated that physical limitations and the need for flexibility have guided choices for many individuals. Additionally, some individuals noted that their diagnosis has inspired their choice of career. Many responses regarding the regular impact of FAP on current career related to the idea of
frequently needing time off or spending too much time in the bathroom during work. On career choice:

“It makes me want to choose a different career where I can actually help people. And to find a career where I am not working super crazy hours so I can enjoy my life. Being diagnosed was the extra push I needed to decide to find a new career.” – 24 y.o. Female (p14)

“Due to pain and such I feel a desk job is best for me.” – 25 y.o. Female (p21)

“I had to choose something with flexible hours and a bathroom always available, and the depression it has caused makes it hard to go after my dreams again” – 23 y.o. Female (p29)

“I want to be a nurse and work with kids, I feel like I understand what it is like to be in the hospital and feel awful and have to go through things that other kids my age hadn't so it made me more mature in sense. I think that I could help kids that are ick feel more comfortable and I know when ever I had a good nurse it made all the difference [sic].” – 18 y.o. Female (p31)

On everyday impacts on current career:

“I am finding my position as a teacher to be difficult with FAP. Looking into a career change. I can't be running to the bathroom or out for procedures at the rate I am.” – 25 y.o. Female (p5)

“Time off sick due to bowel issues and pain. Been put on stage 2 sickness & fear losing my job over something that I cannot change.” – 25 y.o. Female (p13)

However, some individuals noted that they do not feel held back by their diagnosis:

“I believe that I can do whatever career I want no matter what disease I have. I never let my diagnosis prevent me from doing anything that I set my mind to. I just have to make sure that I know where the nearest restroom is. It's not that big of a deal though.” – 22 y.o. Female (p17)

**Relationships**

Participants were either single (39.4%), in a committed relationship (39.4%), or married (21.2%). We asked them to rate the level of impact they feel FAP has had on six different aspects of their romantic relationships. Most participants felt that FAP has had no impact on all six aspects, however reported impacts ranged the entire scale from none at all to a great deal (Table 1). There was a significant correlation between participant age and level of impact in the area of sexual intimacy, with older participants reporting more impact ($r=0.437$, $p=0.012$). Participants who have had a colectomy felt significantly more impacted in the areas of “meeting prospective partners” ($p=0.027$) and “maintaining relationships” ($p=0.045$) than those who have not had a colectomy. Additionally, participants who have been diagnosed with cancer ($n=5$) felt
significantly more impacted overall in their romantic relationships than those who have not had cancer \((p=0.022)\).

We administered the PAS to assess participants’ sense of social integration with family members and friends. We asked participants to rank the applicability of five statements on their relationships with family members and the same statements again regarding their friendships (Table 1). Individuals who have had cancer scored significantly higher on the social integration portion of the PAS for family relationships in the areas of “makes me more willing to help others” \((p=0.013)\) and “helped me become more aware of the love and support available from other people” \((p=0.002)\).

<table>
<thead>
<tr>
<th>Table 1. Relationships</th>
<th>None at all</th>
<th>A little</th>
<th>A moderate amount</th>
<th>A lot</th>
<th>A great deal</th>
<th>Unanswered</th>
</tr>
</thead>
<tbody>
<tr>
<td>Meeting prospective partners</td>
<td>20 (60.6)</td>
<td>4 (12.1)</td>
<td>3 (9.1)</td>
<td>3 (9.1)</td>
<td>2 (6.1)</td>
<td>1 (3.0)</td>
</tr>
<tr>
<td>Dating</td>
<td>14 (42.4)</td>
<td>6 (18.2)</td>
<td>4 (12.1)</td>
<td>4 (12.1)</td>
<td>3 (9.1)</td>
<td>2 (6.1)</td>
</tr>
<tr>
<td>Forming meaningful connections</td>
<td>16 (48.5)</td>
<td>5 (15.2)</td>
<td>7 (21.2)</td>
<td>2 (6.1)</td>
<td>2 (6.1)</td>
<td>1 (3.0)</td>
</tr>
<tr>
<td>Allowing yourself to be vulnerable with someone</td>
<td>10 (30.3)</td>
<td>5 (15.2)</td>
<td>9 (27.3)</td>
<td>5 (15.2)</td>
<td>3 (9.1)</td>
<td>1 (3.0)</td>
</tr>
<tr>
<td>Sexual intimacy</td>
<td>13 (39.4)</td>
<td>7 (21.2)</td>
<td>7 (21.2)</td>
<td>2 (6.1)</td>
<td>3 (9.1)</td>
<td>1 (3.0)</td>
</tr>
<tr>
<td>Maintaining relationships</td>
<td>17 (51.5)</td>
<td>6 (18.2)</td>
<td>3 (9.1)</td>
<td>2 (6.1)</td>
<td>4 (12.1)</td>
<td>1 (3.0)</td>
</tr>
<tr>
<td>(From PAS) Living with FAP has…</td>
<td>None at all</td>
<td>A little</td>
<td>Somewhat</td>
<td>Quite a bit</td>
<td>Very much</td>
<td>Unanswered</td>
</tr>
<tr>
<td></td>
<td>Frequency (%)</td>
<td></td>
<td></td>
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</tr>
<tr>
<td>Social Integration – Family</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Helped me know who I can count on in times of trouble</td>
<td>2 (6.1)</td>
<td>3 (9.1)</td>
<td>2 (6.1)</td>
<td>11 (33.3)</td>
<td>15 (45.5)</td>
<td></td>
</tr>
<tr>
<td>Makes me more willing to help others</td>
<td>4 (12.1)</td>
<td>3 (9.1)</td>
<td>4 (12.1)</td>
<td>9 (27.3)</td>
<td>13 (39.4)</td>
<td></td>
</tr>
<tr>
<td>Helped relationships become more meaningful</td>
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<td>3 (9.1)</td>
<td>9 (27.3)</td>
<td>7 (21.2)</td>
<td>7 (21.2)</td>
<td>1 (3.0)</td>
</tr>
<tr>
<td>Helped me become closer to people I care about</td>
<td>3 (9.1)</td>
<td>4 (12.1)</td>
<td>10 (30.3)</td>
<td>6 (18.2)</td>
<td>10 (30.3)</td>
<td></td>
</tr>
<tr>
<td>Helped me become more aware of the love and support available from other people</td>
<td>2 (6.1)</td>
<td>2 (6.1)</td>
<td>7 (21.2)</td>
<td>10 (30.3)</td>
<td>12 (36.4)</td>
<td></td>
</tr>
<tr>
<td>Social Integration – Friends</td>
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<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Helped me know who I can count on in times of trouble</td>
<td>2 (6.1)</td>
<td>4 (12.1)</td>
<td>6 (18.2)</td>
<td>6 (18.2)</td>
<td>14 (42.4)</td>
<td></td>
</tr>
<tr>
<td>Makes me more willing to help others</td>
<td>4 (12.1)</td>
<td>4 (12.1)</td>
<td>6 (18.2)</td>
<td>6 (18.2)</td>
<td>12 (36.4)</td>
<td>1 (3.0)</td>
</tr>
<tr>
<td>Helped relationships become more meaningful</td>
<td>2 (6.1)</td>
<td>5 (15.2)</td>
<td>10 (30.3)</td>
<td>7 (21.2)</td>
<td>8 (24.2)</td>
<td>1 (3.0)</td>
</tr>
<tr>
<td>Helped me become closer to people I care about</td>
<td>2 (6.1)</td>
<td>5 (15.2)</td>
<td>12 (36.4)</td>
<td>5 (15.2)</td>
<td>8 (24.2)</td>
<td>1 (3.0)</td>
</tr>
<tr>
<td>Helped me become more aware of the love and support available from other people</td>
<td>2 (6.1)</td>
<td>1 (3.0)</td>
<td>11 (33.3)</td>
<td>5 (15.2)</td>
<td>13 (39.4)</td>
<td>1 (3.0)</td>
</tr>
</tbody>
</table>
In describing their relationships, many participants noted feelings of embarrassment or anxiety about their physical needs and worries about rejection from romantic partners or friends. However, many also noted that romantic partners, family, and friends have been supportive and understanding despite these concerns. On romantic relationships:

“My husband and I were engaged when I was diagnosed. It has put a strain on our marriage.” – 25 y.o. Female (p5)

“I've never had issues with meeting people. It's when I'm in their home or vice versa where the embarrassment begins. I know it's loud when I use the toilet. I know it smells afterward. I know there's nothing I can do about it, and that's frustrating when here are new people and I already feel so judged when I'm using a public toilet. Women--strangers--have come up to me in the restroom and said things like, "Have you considered changing your diet?" And I don't want to explain it to them. In the same vein, I know I'm only going to want the kind of person who ISN'T petty enough to say, "You get feces on the bottom of the toilet seat. We have to break up." But it's still embarrassing to brush the bowl every time and, almost like paranoia, keep a stock of air reshener. I have also found that I'm much more comfortable in a sexually intimate moment after I've gone to the bathroom. Otherwise, it might feel like I'm going to have a bowel movement while having sex and that makes me extremely uncomfortable because, ith BMs like mine, you never really know if you'll hold all of it back, no matter how great your Keggels are. But I've been in a committed relationship for seven months now. He's never once complained, no matter how many times I apologize. He does activel try to go to the bathroom before I do, though. This used to bother me, but not anymore. I do the same thing with my mom when I visit home [sic].” – 23 y.o. Female (p6)

“My current partner was very informed about FAP and it was mainly due to them that I went for the tests in the first place. After they heard that my mother had had Gardner's Syndrome, they were adamant that I should get myself checked out. They searched for a lot of the information on FAP and Gardners, and as a result, was extremely understanding [sic].” – 22 y.o. Non-Binary (p16)

“I had to have a stoma (illeostomy). This alone causes so many psychological issues i cant even fathom it all. Sex was rare, especially when i felt less than adequate. I could always smell my output but no one else could, it made me feel like they were jst being nice & saying they couldn't smell it. I don't go out of fear of a bag malfunction as i had a flush stoma. I worried people would bump me, i worried id spoil the mood, i left my house 3 times in the last year for a social event, if i didn't have o leave, id stay always. I felt vulnerable constantly but i couldn't let anyone see that i thought i needed to be strong for my family i see now i should have allowed myself to be weak more because thats when you see your real strength [sic].” – 25 y.o. Male (p22)

“People don't generally like to get themselves into a relationship where they already know their partner is sick and then those that don't mind and are supportive really don't understand what you are dealing with and on top of that intimacy is difficult. Wen you have doctors and nurses touching you all the time and your dignity is gone its sometimes hard to erase the touches of a doctor and when a lover touches you it triggers something and I have a lot of anxiety and panic attacks [sic].” – 23 y.o. Female (p29)

On family relationships:

“My sister and mother have FAP and they have become my FAP support network as we go through it together.” – 19 y.o. Female (p33)
On friendships:

“...I feel awkward telling my friends that I have FAP and how this affects my body. I find that people tend to not really understand why I use the restroom more frequently than most people or why I cannot eat certain foods. I try to keep that aspect of my life to myself so that people won't think of me differently [sic].” (p17)

“...Often I'd be too ill feeling to reply to msgs or answer the phone, people had a hard time understanding that & I lost many friends for it. [...] [sic]” – 25 y.o. Male (p22)

**Family Planning**

Only three participants (9.1%) currently have children and all 3 reported that they had their children after being diagnosed with FAP. Two of these individuals conceived naturally and one participant underwent IVF with PGD to have children. Of the 30 participants who do not have any children, 17 (56.7%) plan to have children, 4 (13.3%) do not plan to have children, 1 (3.3%) has not yet thought about it, and 8 (26.7%) are undecided.

For those that answered anything other than yes (no, not yet thought about it, or undecided) to having more or any children (n=14), 6 (42.9%) feel that their diagnosis of FAP has not influenced this position at all (1 individual who does not plan to have more children, 1 individual who does not plan to have any children, and 4 individuals who are undecided) and 4 (28.6%) feel that their diagnosis has influenced this position a great deal (3 individuals who do not plan to have children and 1 who is undecided).

Participants who responded that they plan to have children (n=17) or to have more children (n=2) were asked to select all options they have considered in their planning and IVF with PGD was the most frequently considered option (Figure 1). Even with multiple options in mind the majority (52.6%)
reported that they are already undergoing or think they might ultimately undergo IVF with PGD. Equal numbers of individuals reported that they have already or think they might ultimately choose to conceive naturally (21.1%) or adopt (21.1%). Of the 19 individuals who are planning to have more or any children, the majority feel that their diagnosis of FAP has influenced these plans a great deal (68.4%) or a moderate amount (15.8%), while 10.5% feel that their diagnosis has had no influence. Specifically, for the 2 individuals who have already had children and plan to have more, one has already used IVF with PGD and one conceived naturally. Both reported that these respective methods were the only considered and likely to be used again. The individual who has used and will continue to use IVF with PGD reported that FAP has had a great deal of influence on these plans, while the individual who conceived naturally and will continue to do so reported that FAP has had no influence at all on these plans.

Seventeen individuals used the open-ended response to comment on family planning. None of these individuals already have children and 11 are planning to have children, 3 are planning not to have children, and 3 are undecided. Concern over the potential of passing FAP on to children and how this has led to the consideration of other options for having children was expressed by some participants, although others noted that their diagnosis has not dissuaded them from having biological children. On other methods for having children:

“I did IVF with PGD and it failed. My husband and I are currently unsure how we will proceed but until my colon comes out, a pregnancy is not an option with twice a year colonoscopies.” – 25 y.o Female (p5)

“I only recently started to think about having children, and it was largely influenced by the difficulty post-surgical FAP patients allegedly have with bearing children. I, however, didn't want to risk cancer and wait to have children before my surgery. So here I am, weighing all my options like I haven't had to before, when I wasn't even thinking about kids.” – 23 y.o. Female (p6)

“My father had 3 children with 2 different wives & we all have FAP. It is simply irresponsible to bring life into this world knowing you have a 3/3 hit rate. For me, the testing of the egg is option number 1. I'd love a child of my own but if there's even n option that they miss the mutation because it presents differently I'd rather find donors or adopt. I cannot let my child go through what i have [sic].” – 25 y.o. Male (p22)
“I do not want my children to have this disease, and I have always wanted to foster and adopt children. I would like to have a large family, and especially with FAP I am planning on looking into ways of ensuring I do not pass it on genetically. I am still in college however so I am not quite there yet [sic].” – 19 y.o. Female (p33)

On having biological children:

“Not sure if I want to naturally have kids and go through with the possibility of having Hepatoblastoma/FAP.” – 25 y.o. Female (p19)

“I always wanted kids. Worried my fiance wouldn't want "our" kids. He is fully supportive of having natural kids only if my body can handle carrying a child” – 22 y.o. Female (p25)

“I am extremely uncertain of whether I will have children. It is possible, but I can say nothing more. As far as the influence FAP has on having children, it would only be relevant to the degree that it's relevant to my partner. To me, the possibility of passing FAP onto my children currently does not sway me in any particular direction [sic].” – 25 y.o. Male (p26)

**Living with FAP**

As previously described, we administered the PAS to assess overall adaptation to living with FAP (Appendix B). The 20-question scale is designed to assess coping efficacy, self-esteem, spiritual well-being, and social integration. In our survey, we gave participants the social integration questions in the context of both family relationships and friendships, as described above (Table 1). We scored participants overall (including social integration questions twice), and then scored each domain of adaptation separately. Participants who have had a cancer diagnosis scored significantly higher in the areas of: “helped me accept the way things work out” (p=0.039), “taught me to adjust to things I cannot change” (p=0.021), “helped me take things as they come” (p=0.047), “helped me look at things in a more positive way” (p=0.030), “helped me become a better person” (p=0.003), and “helped me develop a deeper sense of purpose in life” (p=0.031) than those who have not had cancer. Additionally, participants who have had cancer scored significantly higher on the overall domain score for coping efficacy (p=0.037) than those who have not had cancer. Individuals who are the only person in their family affected with FAP scored significantly higher in the areas of “helped me become a stronger person” (p=0.016), and “helped me become a better person” (p=0.044) than those who have affected family members.
We asked participants to elaborate on their answers to the PAS and some commented on how their diagnosis has made them a stronger person and given them a greater appreciation for life, while others noted that FAP has been part of them but hasn’t changed them from who they have always been. On living with FAP:

“Yes, FAP has taught me how to face and deal with adversity and possible disasters life tends to throw, but I've always been an optimist. FAP hasn't influenced that. I've also always felt like life is as meaningful as we make it, which FAP hasn't changed. I've always had a great appreciation for life, though I've never felt like I have a distinct purpose or calling; I've always felt peaceful, which FAP has not changed, and have never been particularly spiritual.” – 23 y.o. Female (p6)

“I don't allow tough times to keep me down.” – 25 y.o. Female (p19)

“I faced many ups and downs before being diagnosed. Being diagnosed was the icing on the cake for me. I have learned though 3 family deaths in 3 years what life means. But is has given me a better out look on life [sic].” – 22 y.o. Female (p25)

“In general, living with FAP has not helped me in my personal strength etc. (it has mostly been a background nuisance I don't give much mind). It has to a small degree reinforced the fact that there are some things you just have to deal with.” – 25 y.o. Male (p26)

**Genetic Counseling**

Twenty-two individuals (66.7%) have seen a genetic counselor regarding their diagnosis, and 85% of these felt that their genetic counseling session(s) were extremely useful (35%), very useful (25%), or moderately useful (25%). Of the 20 individuals who had seen a genetic counselor and remember their session, there was some variability in how they felt about it, although most expressed some level of agreement favoring a more positive experience (Table 2).

On genetic counseling experiences:

“I had a wonderful genetic counselor when I was first diagnosed who took on the role of providing me information when my doctor was too busy. […]” – 25 y.o. Female (p5)

“I wish there were more genetic counselors familiar with FAP. The last time I saw a genetic counselor, she specialized in breast cancer genetics and couldn't tell me more than I could read online but she was the only option.” – 24 y.o. Female (p18)

Participants were also asked whether they are in need of more support or have more questions related to FAP than their genetic counselor and/or other healthcare providers have provided or answered. On unmet needs:
“I am still feeling confused regarding how getting pregnant could affect my body due to this disease. I currently have desmoid tumors and am on Gleevec, and I know that I cannot stay on this medication if I get pregnant. I would like to speak with my doctor regarding this issue [sic].” – 22 y.o. Female (p17)

“I think that there should be more mental health support. You don’t realize how much this wears you down until you stop rushing through life for a moment.” – 23 y.o. Female (p29)

“The main distress of living with FAP for me has been not knowing how much to trust surgeons. They have a variety of motivations for impressing upon me the urgency of the need for surgery, and I know this isn’t the only approach because my uncle in Europe has a surgeon who think it is better to wait. I wish I could get more perspectives, I feel that I need to make a huge decision with very little information [sic].” – 25 y.o. Male (p26)

On things not addressed in this survey:

“I used to have no trouble with losing weight and I had energy. Now I have no energy and I have gained a lot of weight since my ileostomy reversal.” – 25 y.o. Female (p19)

“Nutrition is hard for me. Trying to find out what is best for me to eat and what I should eat. There is no clear diet or recommendations on what to eat.” – 22 y.o. Female (p25)

“I would just like to connect with others my age that have this disease that aren’t my siblings.” – 23 y.o. Female (p29)

<table>
<thead>
<tr>
<th>The genetic counselor…</th>
<th>Strongly Agree</th>
<th>Disagree</th>
<th>Somewhat Agree nor Disagree</th>
<th>Neither Agree nor Disagree</th>
<th>Strongly Disagree</th>
<th>Disagree</th>
<th>Somewhat Disagree</th>
<th>Neither Agree nor Disagree</th>
<th>Frequency (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Presented me with an appropriate amount of information</td>
<td>1 (3.0)</td>
<td>0</td>
<td>0</td>
<td>2 (6.1)</td>
<td>3 (9.1)</td>
<td>11 (33.3)</td>
<td>3 (9.1)</td>
<td></td>
<td></td>
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<tr>
<td>Answered all of the questions that I had in a satisfactory manner</td>
<td>0</td>
<td>0</td>
<td>2 (6.1)</td>
<td>0</td>
<td>3 (9.1)</td>
<td>10 (30.3)</td>
<td>5 (15.2)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Was sensitive and attentive to my needs and emotions</td>
<td>1 (3.0)</td>
<td>1 (3.0)</td>
<td>0</td>
<td>0</td>
<td>4 (12.1)</td>
<td>11 (33.3)</td>
<td>3 (9.1)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Made me feel supported and comfortable</td>
<td>1 (3.0)</td>
<td>0</td>
<td>2 (6.1)</td>
<td>1 (3.0)</td>
<td>3 (9.1)</td>
<td>9 (27.3)</td>
<td>4 (12.1)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Is someone I would like to see at regular intervals as I have questions throughout my life</td>
<td>1 (3.0)</td>
<td>1 (3.0)</td>
<td>3 (9.1)</td>
<td>1 (3.0)</td>
<td>6 (18.2)</td>
<td>3 (9.1)</td>
<td>5 (15.2)</td>
<td></td>
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</tbody>
</table>
Discussion

We surveyed 18 to 25-year-old individuals with Familial Adenomatous Polyposis to characterize how this condition has impacted the lives of these emerging adults. We focused particularly on areas of development that are often important to emerging adults: education, career planning, relationships, and family planning. Additionally, we assessed overall adjustment (via the Psychological Adaptation Scale), how participants felt at the time of diagnosis, and how they felt about their experiences with genetic counseling. Our goals were to characterize the unique challenges for emerging adults with FAP and to highlight the needs of this group so they can be most appropriately supported by their healthcare providers throughout their lives.

Diagnosis

Similar numbers of participants were diagnosed clinically prior to genetic testing (n=14, 42.4%) and presymptomatically by genetic testing based on their family history (n=13, 39.4%). Of those diagnosed by presymptomatic genetic testing, 11 (84.6%) reported that they had little or no say in the decision. Duncan et al. also discussed a lack of patient involvement in the decision-making process when testing young individuals for FAP, suggesting that this happens frequently. Considering that many of our participants were minors at the time of diagnosis, it is somewhat understandable that many of these individuals may have had little or no say in the decision to undergo presymptomatic testing. However, in looking at the ages of diagnosis of the 11 participants who reported little or no say in the decision, 3 individuals were between the ages of 18 to 23 and 4 individuals were between the ages of 15 to 17. While it is understandable that family members and/or providers might have strongly influenced these individuals to have
genetic testing given the high rate of cancer in the absence of preventative surgery, it seems that many of these individuals were old enough that they should have been given more input into the decision-making process. For example, when asked about how she felt at the time of her diagnosis, one 25-year-old female participant diagnosed between the ages of 15 to 17 by presymptomatic genetic testing who indicated that she had no say in the decision to undergo testing remarked that she felt “like it was a death sentence as my dad had just died of it a few months before,” and that she wished she “had had more input.” Additionally, these 7 individuals who were between the ages of 15 to 23 at the time of diagnosis reported receiving varying amounts of information about FAP (from a little to a great deal), yet almost all felt that the amount they received was extremely or somewhat appropriate or neutral. This suggests that a discussion prior to genetic testing about the decision and why it was important for them may have been well received and useful to these individuals. Our results do not suggest that these young individuals who underwent presymptomatic genetic testing were significantly adversely affected by this lack of control during the diagnostic process. However, it seems that taking the needs and input of all patients undergoing this testing into account – particularly those old enough to have some understanding of the implications – would be beneficial. Addressing their concerns and helping them better understand the need for testing may help them better cope with the results. This supports the finding by Duncan et al. that predictive genetic testing for FAP in individuals aged 10 to 17 was viewed as a major life event, thus requiring appropriate support and counseling (Duncan et al., 2010).

Many participants felt that the amount and type of information about FAP they received at their initial diagnosis was extremely or somewhat appropriate, although this was discordant with many of the open-ended responses, which expressed frustration with a lack of
knowledgeable providers and information. This frustration has been observed previously, for example by Fritzell et al., who found the topic of providers with a lack of knowledge or interest in FAP frequently discussed by participants in their focus group study (Fritzell et al., 2010). Of those in our study who commented on a lack of information, there was considerable variability in their responses although they tended more toward inappropriate amount and type of information than those who did not comment on a lack of information at the time of diagnosis. This suggests that those who were dissatisfied with the information they received may have been more likely to respond to this question or that memories of emotions felt at the time of diagnosis were more vivid than memories regarding information given, thus leading some participants to comment only on emotion.

**Education and Career Planning**

Although none of the variables we analyzed were found to have a statistically significant impact on education or career, the potential impact is evidenced by the variability in participant responses, as well as by responses of those who answered open-ended questions. In terms of education, it seems the most negative impact was related to the time of surgery, with participants citing missed time in school, difficulty planning for the future around an impending surgery, negative impacts on grades and focus, or negative impacts of medical expenses. For example:

“Before and after being diagnosed, although I dealt with the situation fairly well, it was still difficult to come to terms with what would be happening. Having to miss quite a few lectures/lab sessions for tests and appointments resulted in me having to rsit the university year, although I graduated after going through with the ileostomy this year [sic].” – 22 y.o. Non-Binary (p16)

“I had finished my bachelors before I was diagnosed and with my medical bills, it's not realistic to return to school for a masters [sic].” – 25 y.o. Female (p5)

“In middle and high schools it did not affect me much other than annual colonoscopies. Now that I am in college, I am facing surgery within the next 1-5 years depending on my prognosis. It is affecting my academic plan, housing for next year, co op and stdy abroad plans, and more. Especially on a quarter system it is very difficult to plan ahead when I may have to take anywhere from 3-12 months off [sic].” – 19 y.o. Female (p33)
Despite this, many felt that their diagnosis of FAP has overall had both positive and negative impacts on their education, for example:

“FAP has been both positive and negative for me when it comes to school. I do sometimes have hard days but I have also learned a lot of about my disease. It has actually helped me find my career field, as I hope to become a nurse and work with patients that share my disease [sic].” – 23 y.o. Female (p7)

“Had to repeat a year at age which delayed going to uni with my friends, but am more mature and able to handle uni better than may have [sic].” – 20 y.o. Male (p8)

In terms of career planning, due to the young age of our study population, many participants may still be at the beginning stages of setting their career goals or entering their working lives and this may account for the varied responses regarding the impacts of FAP on choice of career and current career. This may be more representative of the period of emerging adulthood rather than a diagnosis of FAP. However, it is clear that some of the individuals in our study have been impacted by FAP in either their career choices or everyday work life. For example, some commented on the negative effects of FAP regarding physical limitations and the need for flexibility and accommodations at work:

“I can't sit or stand too long. When I sit for long I have shooting pain up my tailbone. Standing too long gives me upper back pain. I already have chronic back pain and can't lift up to 50 lbs like many places expect. I'm in the restroom for 15 minutes or longer each time I go, and I'm always tired.” – 25 y.o. Female (p19)

“I have a career as an office administrator currently, but when looking for new jobs I always find myself looking at the bathroom location due to the smelly and loud bowel movements I have multiple times a day. I also tend to get sick often which doesn't allow me to stay at a job too long [sic].” – 25 y.o. Female (plnc.3)

Other participants noted that FAP has inspired current career goals due to their experiences:

“I am working on my health management major. I had hospital staff that didn't have the training to help me after surgery and the hospital didn't have the supplies I needed. Wanted to make sure I could make patients had what they needed [sic].” – 22 y.o. Female (p25)

“I would love to be a nurse, possibly even working in the colonrectal field. My diagnosis has led me in this direction [sic].” – 23 y.o. Female (p7)

Overall, surgery and the resulting physical limitations seem to be reasons individuals are most likely to miss or struggle in school or work. Our results support findings by Mireskandari et al., which were that FAP had a limited long-term, overall impact on education, but that impacts
on career included a lack of privacy, missed time at work, and physical limitations (Mireskandari et al., 2009). Healthcare professionals who care for young individuals with FAP need to be prepared to provide support in making appropriate plans for surgery to limit the impact as much as possible. Additionally, these individuals may need help strategizing regarding communications with their school or workplace, or advice for how to practice self-care while simultaneously achieving success in their desired education and career.

**Relationships**

Romantic relationships were most significantly impacted in participants who have had a colectomy or a cancer diagnosis. This is not surprising given that both situations are life- and body-altering, with the potential to leave an individual feeling vulnerable, self-conscious and anxious. This supports the reviewed literature, in particular Zebrack’s review, which highlights the challenges for young adults with cancer regarding sexuality and intimacy, telling partners about a diagnosis or its affects, and negative self-regard (Zebrack, 2011).

Romantic relationships in emerging adulthood are generally very variable, therefore the variability in responses regarding romantic relationships may be a result of having FAP, although it may also be the result of being an emerging adult. Comparing responses of emerging adults with and without FAP might help clarify how much of this variability is truly attributable to FAP. Respondents who commented on their relationships touched upon impact such as the strain that this disease puts on a relationship, dealing with embarrassing bodily functions, anxiety around telling partners about the condition, and feeling unwanted, highlighting those that are attributable to FAP. Some also mentioned the support and understanding they have received from their partners despite these concerns.
In contrast to the negative overall impact of a cancer diagnosis on romantic relationships, participants who have had cancer scored significantly higher on two areas of the social integration portion of the PAS for family relationships. This suggests that their diagnosis has actually helped them to become more well-adjusted in these areas, possibly due to the increased support they received from family members while dealing with cancer. For example, one 22-year-old female who has been diagnosed with cancer says:

“Having FAP made me realize how much my family loves me. My mom attends every appointment with me that she can and has always been by my side through everything. My extended family has also been very supportive of me.” (p17)

Other participants commented on how FAP has strengthened their relationships with family members and friends. For example, when describing how supportive her friends have been of her, one 24-year-old female participant says:

“I have found out that as I told my friends because if I told them then it's real and it's there. That they have all been by my side supporting me and my decision for surgery. I am overcome with the fact that they all would come see me in a heartbeat just as I would for them. Just didn't expect it back so it makes my heart so full and happy to know I have people there for me [sic].” (p14)

However, others noted that the challenge of learning to live with FAP has cost them friendships or that friendships have been difficult due to the awkwardness of discussing the impact of this condition, for example one 22-year-old female says:

“I'm still a bit new to my diagnosis I haven't quite learned how to live with it and my poor coping mechanisms have cost me some very good friends.” – 23 y.o. Female (p29)

Overall, healthcare providers caring for emerging adults with FAP need to be aware of these types of impacts on relationships with family, friends, and romantic partners, which may be exacerbated by being an emerging adult in general. Encouraging conversation with partners, friends, family, or professionals about the diagnosis or related challenges may help in the coping process.
Family Planning

Andrews et al. highlighted the anxiety of individuals with FAP surrounding their children developing FAP and the need for support in coping with this possibility (Andrews et al., 2006). In our study, IVF with PGD has been considered by the majority (73.7%) of participants who are planning to have more or any children, more frequently than other potential methods (Figure 1). IVF with PGD is also the method that the majority (52.6%) of participants reported that they are already undergoing or think they might ultimately undergo. These results support Andrews’ finding of increased anxiety about the potential of children developing FAP, in that many of our participants are considering or planning to use IVF with PGD to avoid passing FAP on to their children. Although this may be an option for many of our participants, others in our study expressed that this would not be an option for them. For example, one 19-year-old male participant says “i will most definitely adopt my children, there is no way we would be able to afford invetro [sic]” (p27). Additionally, 3 of the 4 individuals who do not have or plan to have any children stated that their diagnosis of FAP has influenced this decision a great deal, with one saying, “do not want children to go through this disease” (25 y.o. male, p28). We did not ask those who answered anything other than yes to having more or any children about what methods they might consider or choose and it is possible that those who do not plan to have children or who are struggling with this decision are not fully aware of all available options.

This idea that the potential to pass FAP on to children can be anxiety provoking was supported further by those who commented on family planning, although these may be biased toward those planning on having children (11 out of 17 who commented). Participants discussed options to avoid passing this condition on, and for some, the possibility of IVF with PGD seems to have provided a sense of optimism that has changed their minds regarding having children:
“When diagnosed at 15 I made my mind up not to have children and risk passing it on only finding out about PGD has changed my mind.” – 25 y.o. Female (p13)

“I want to have kids and like 3 or 4 but since ive been diagnosed with fap I’ve thought alot about how I want to conceive my kids. Before being diagnosed I thought that in vitro was strange and unnatural but now I'm considering it [sic].” – 18 y.o. Female (p31)

Another commonality that emerged highlights the resiliency of these individuals in that even though they have been impacted by this condition they still wish to have biological children:

“It [FAP] hasn't [influenced my decision] because I'm fine and my family who has it are fine. Yes we all are going to have had the same surgery but we're all alive.” – 24 y.o. Female (p14)

“I realize that my children have a 50/50 chance of inheriting FAP from me. I still want to have biological children despite this though; I feel that having FAP has made me a stronger person and I know that if my children are anything like me, they will handle this disease just fine. I also realize that I may not be able to have children because of this disease, and in that case, I do not mind adopting children [sic].” – 22 y.o. Female (p17)

Our population of emerging adults (aged 18 to 25) is younger than the population surveyed by Andrews (aged 18 to 35) and most of our participants do not yet have children, therefore rather than needing support for coping with the potential for existing children to develop FAP, our results suggest that emerging adults with FAP are likely to need information about available options for having children, particularly IVF with PGD. Most importantly, these individuals need support for any decision that they make.

**Living with FAP**

The 20-question PAS is designed to assess coping efficacy, self-esteem, spiritual well-being, and social integration by providing statements related to each of these domains and asking participants to rate how living with FAP has impacted each statement (scale: 1 = not at all, 2 = a little bit, 3 = somewhat, 4 = quite a bit, 5 = very much) (Appendix B and Table 1). Although responses across the PAS varied, the average overall score among all participants was around the middle of the scale at 3.40 (higher scores indicate better adjustment). Separate domain average scores were similarly in the middle of the scale: coping efficacy: 3.15, self-esteem: 3.59, spiritual well-being: 3.09, social integration – family: 3.66, and social integration – friends: 3.60. Average
scores for each individual question were almost all in the middle of the scale, indicating that, on average, participants are well adjusted across the scale. Scores for “helped me become a stronger person” and “helped me know who I can count on in times of trouble” (for family relationships) were particularly high at 4.09 and 4.03, respectively, indicating that FAP has had “quite a bit” of impact for most participants in these areas. Participants who commented on their responses noted how their diagnosis has made them stronger, given them a greater appreciation for life, and has not changed them from who they’ve always been. On average, our participants are reportedly relatively well adjusted to their diagnosis, which provides support for Fritzell’s (2010) findings that most individuals with FAP report “living a normal life.”

Participants who have had a cancer diagnosis (n=5) scored significantly higher than those who have not had cancer on 6 questions, 4 of which are in the domain of coping efficacy, which explains why those with cancer also scored significantly higher in this individual domain. This provides support for the idea that these individuals have integrated the cancer diagnosis into their identity and are able to cope with this adversity (Zebrack, 2011). For example, in elaborating on their responses to the PAS, one 22-year-old female who has had cancer commented:

“I feel that if I did not have FAP, I would not be the person I am today. I am strong, courageous, and persistent because of the trials I have been through with this disease. I will fight through anything and not give up. Sometimes I get discouraged and life seems unfair, but for the most part I have learned to deal with the uncertainty. It has definitely made me rely on God more through the difficult times [sic].” (p17)

Although some individuals who have dealt with a diagnosis of cancer may have learned how to cope successfully, the process is difficult for others. For example, one 23-year-old participant who scored 3.68 overall on the PAS, acknowledged in some of her comments her own poor coping mechanisms and mentioned her struggles with anxiety and depression resulting from her diagnosis. Her comment elaborating on her PAS responses illustrates how coping mechanisms may take the place of the actual coping necessary to heal from a diagnosis of cancer and FAP:
“I feel like any diagnosis of a chronic illness is a heavy thing to carry around but I find that the sadder I feel inside the more I want to make others around me smile and laugh. It has definitely amped up my empathy game.” (p29)

All our participants who have had cancer reported being first diagnosed during emerging adulthood. These results suggest that a diagnosis of cancer in emerging adults with FAP might actually lead to better overall coping, although due to the small number of individuals in our study who have had cancer (n=5), it is hard to say whether this result is generalizable.

**Implications for Genetic Counseling**

Most participants who have seen a genetic counselor felt that the experience was useful and positive, and 42.5% expressed agreement with the possibility of seeing a genetic counselor at regular intervals. Comments by some participants support the idea that regular visits with a genetic counselor experienced with FAP could be useful:

“\(\text{I was very young when I met the counselor and I didn't have any questions for her, so I don't have any complaints. However, if I were to make any big decisions (surgery), I would want to speak to one.}\)" – 25 y.o. Male (p26)

“I feel comfortable with where I am at, but I do have many questions about what my life will be like in the future and what other risks may play a part in my life.” – 23 y.o. Female (p7)

“Because I was told it wasn't a big deal, it didn't affect me much. I wish I had done more research and understood what would happen now that I'm in college and really dealing with it.” – 19 y.o. Female (p33)

This idea is also supported by our results that many participants that underwent presymptomatic genetic testing reported little or no involvement in the decision or that some individuals were too young to really understand what was going on when they were first diagnosed. Another commonly noted response was a strong emotional reaction to the diagnosis, with some participants reporting being devastated, numb, shocked, and at a loss for how to cope. Much of the reviewed literature concludes that the impact of FAP changes from the time of testing (often as a child or adolescent) throughout a person’s life, warranting continued follow up with genetic counselors, and our results further support this idea (Andrews et al., 2006 & Andrews et al. 2007 & Duncan et al., 2010 & Fritzell et al., 2010 & Mireskandari et al., 2009).
It seems that in addition to necessary medical appointments related to FAP, a longitudinal care model of periodic follow-up every 1 to 2 years after initial diagnosis for young individuals with FAP might be appropriate. This follow-up may include individual meetings with a genetics or FAP expert, gastroenterologist, and/or mental health professional, although it seems that the most beneficial approach might be multidisciplinary care through a team of all the above professionals when available. Due to limited availability of healthcare professionals with experience and expertise in FAP, telehealth appointments may be necessary to expand care to individuals in need.

**Limitations**

An obvious limitation to our study is the small sample size (n=33). Our analyses are limited in their ability to find significant differences between participants and a larger study might reveal that factors we have looked at have actually had significant impacts on the lives of emerging adults with FAP. Additionally, our analyses of open-ended response are limited in that not all participants responded to all (or any) open-ended questions. Therefore, commonalities in open-ended response that we described may be biased as they are based on the responses of only some participants. Finally, we recruited participants through the Hereditary Colon Cancer Foundation and its associated Facebook support groups, therefore our results are exclusive of emerging adults with FAP who are not associated with these groups. However, these individuals are likely more representative of the general population with FAP than if we had recruited individuals through an institution-based research registry or other cohort. Still, our results may not be representative of all emerging adults with FAP due to the limited sample size of our study.
Future Research

This study is an early look at the impact of FAP on the lives of emerging adults and we believe that a similar, larger study might yield more significant results. This might be achieved through alternate methods of recruitment or larger windows of time for surveys to be active. Additionally, comparing results from emerging adults with FAP to older individuals with FAP might provide more insight into the unique needs of this group.
Conclusions

Despite being a period of life typically characterized by variability, growth, and change, emerging adults living with FAP face some unique challenges as they learn to incorporate this diagnosis into their lives. While many of our participants demonstrated resiliency and strength and were relatively well adjusted, others are struggling more with the diagnosis. Our results suggest that a longitudinal care model of periodic follow-up with a multidisciplinary care team might be appropriate for emerging adults with FAP. Healthcare professionals involved in the care of these individuals need to be prepared to provide support in strategizing ways to integrate this condition into their academic, work, and personal lives.
<table>
<thead>
<tr>
<th>Control in Presymptomatic Genetic Testing</th>
<th>Age at diagnosis</th>
<th>Only had genetic testing based on family history (no symptoms)</th>
</tr>
</thead>
<tbody>
<tr>
<td>When you underwent genetic testing based on your family history, how much did you feel that this was your own decision?</td>
<td></td>
<td></td>
</tr>
<tr>
<td>I made this decision mostly by myself. Family members, providers, or others had a little influence.</td>
<td>12 – 14</td>
<td>1</td>
</tr>
<tr>
<td>(n = 2)</td>
<td>21 – 23</td>
<td>1</td>
</tr>
<tr>
<td>My decision was mostly made for me by family members, providers, or others. I had little say.</td>
<td>9 – 11</td>
<td>1</td>
</tr>
<tr>
<td>(n = 9)</td>
<td>12 – 14</td>
<td>3</td>
</tr>
<tr>
<td></td>
<td>15 – 17</td>
<td>2</td>
</tr>
<tr>
<td></td>
<td>18 – 20</td>
<td>1</td>
</tr>
<tr>
<td></td>
<td>21 – 23</td>
<td>2</td>
</tr>
<tr>
<td>My decision was entirely made for me by family members, providers, or others. I had no say.</td>
<td>15 – 17</td>
<td>2</td>
</tr>
<tr>
<td>(n = 2)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Total:</td>
<td></td>
<td>13</td>
</tr>
</tbody>
</table>
Appendix B: Psychological Adaptation Scale

<table>
<thead>
<tr>
<th>Psychological Adaptation Scale</th>
<th>Not at all</th>
<th>A little bit</th>
<th>Somewhat</th>
<th>Quite a bit</th>
<th>Very much</th>
<th>Unanswered</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Coping Efficacy</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Helped me accept the way things work out</td>
<td>6 (18.2)</td>
<td>6 (18.2)</td>
<td>7 (21.2)</td>
<td>5 (15.2)</td>
<td>9 (27.3)</td>
<td>-</td>
</tr>
<tr>
<td>Helped me learn to deal better with uncertainty</td>
<td>9 (27.3)</td>
<td>4 (12.1)</td>
<td>9 (27.3)</td>
<td>3 (9.1)</td>
<td>8 (24.2)</td>
<td>-</td>
</tr>
<tr>
<td>Taught me how to adjust to things I cannot change</td>
<td>3 (9.1)</td>
<td>10 (30.3)</td>
<td>5 (15.2)</td>
<td>5 (15.2)</td>
<td>10 (30.3)</td>
<td>-</td>
</tr>
<tr>
<td>Helped me take things as they come</td>
<td>2 (6.1)</td>
<td>8 (24.2)</td>
<td>10 (30.3)</td>
<td>3 (9.1)</td>
<td>10 (30.3)</td>
<td>-</td>
</tr>
<tr>
<td>Helped me to look at things in a more positive way</td>
<td>7 (21.2)</td>
<td>4 (12.1)</td>
<td>10 (30.3)</td>
<td>3 (9.1)</td>
<td>9 (27.3)</td>
<td>-</td>
</tr>
<tr>
<td><strong>Self Esteem</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Helped me learn to handle difficult times</td>
<td>3 (9.1)</td>
<td>2 (6.1)</td>
<td>7 (21.2)</td>
<td>6 (18.2)</td>
<td>15 (46.5)</td>
<td>-</td>
</tr>
<tr>
<td>Helped me become more comfortable with who I am</td>
<td>8 (24.2)</td>
<td>6 (18.2)</td>
<td>7 (21.2)</td>
<td>4 (12.1)</td>
<td>8 (24.2)</td>
<td>-</td>
</tr>
<tr>
<td>Helped me become a stronger person</td>
<td>3 (9.1)</td>
<td>2 (6.1)</td>
<td>2 (6.1)</td>
<td>8 (24.2)</td>
<td>18 (54.5)</td>
<td>-</td>
</tr>
<tr>
<td>Helped me feel better about my ability to handle problems</td>
<td>5 (15.2)</td>
<td>3 (9.1)</td>
<td>7 (21.2)</td>
<td>8 (24.2)</td>
<td>10 (30.3)</td>
<td>-</td>
</tr>
<tr>
<td>Helped me become a better person</td>
<td>5 (15.2)</td>
<td>1 (3.0)</td>
<td>7 (21.2)</td>
<td>9 (27.3)</td>
<td>11 (33.3)</td>
<td>-</td>
</tr>
<tr>
<td><strong>Spiritual Well-Being</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Helped me learn my life is more meaningful</td>
<td>6 (18.2)</td>
<td>3 (9.1)</td>
<td>6 (18.2)</td>
<td>7 (21.2)</td>
<td>10 (30.3)</td>
<td>1 (3.0)</td>
</tr>
<tr>
<td>Given me a greater appreciation for life</td>
<td>5 (15.2)</td>
<td>4 (12.1)</td>
<td>2 (6.1)</td>
<td>10 (30.3)</td>
<td>11 (33.3)</td>
<td>1 (3.0)</td>
</tr>
<tr>
<td>Helped me develop a deeper sense of purpose in life</td>
<td>9 (27.3)</td>
<td>1 (3.0)</td>
<td>7 (21.2)</td>
<td>5 (15.2)</td>
<td>10 (30.3)</td>
<td>1 (3.0)</td>
</tr>
<tr>
<td>Helped me feel peaceful</td>
<td>13 (39.4)</td>
<td>5 (15.2)</td>
<td>4 (12.1)</td>
<td>3 (9.1)</td>
<td>7 (21.2)</td>
<td>1 (3.0)</td>
</tr>
<tr>
<td>Helped me find strength in my faith or spiritual beliefs</td>
<td>14 (42.4)</td>
<td>1 (3.0)</td>
<td>4 (12.1)</td>
<td>4 (12.1)</td>
<td>9 (27.3)</td>
<td>1 (3.0)</td>
</tr>
</tbody>
</table>
The Hereditary Colon Cancer Foundation hereby agrees to assist Nina D’Orlando in recruiting suitable individuals with Familial Adenomatous Polyposis (FAP) for her Master’s thesis project. We understand that the goal is to recruit as many individuals aged 18 to 25 as possible, with either a clinical or genetic diagnosis of FAP for participation in an online survey. The goal of the survey will be to assess the overall impact of a diagnosis of FAP on multiple different domains of life relevant to emerging adults.

Once the project has IRB approval, we will assist Nina in recruiting by sending email notices to members of the Hereditary Colon Cancer Foundation and associated support groups. Additionally, we will post information on the Hereditary Colon Cancer Foundation website, www.HCCTakesGuts.org.

Sincerely,

Travis H. Bray, PhD
Previvor, Founder, and Executive Director
Hereditary Colon Cancer Foundation
Subject: Do you have FAP? Are you between the ages of 18 to 25?

What are we asking from you?

We are asking for your participation in an anonymous online survey. We want to gather information about what life is like for individuals aged 18-25 who have a diagnosis of Familial Adenomatous Polyposis (FAP). Anyone who is currently between the ages of 18-25 and who has a clinical and/or genetic diagnosis of FAP can take the survey, even if you were diagnosed before age 18. We also ask that you encourage family members and/or friends who have a diagnosis of FAP and who are between the ages of 18-25 to take the survey. The results of this survey will help educate medical professionals about the unique needs of young individuals with FAP.

What should you expect?

This survey will ask questions about how your diagnosis of FAP has impacted your life. We are curious about multiple areas of your life including education, job choices, and planning a family. We would also like to know how FAP has impacted your relationships with family, friends, and partners. We are very interested in your insights. Your answers will help genetic counselors, doctors, and other healthcare providers better support and care for patients, like you, who have FAP.

What do you have to do?

Simply click the link below to go to the survey.

Some important things to know about this study:

- The survey is open to anyone currently aged 18 to 25 who has a diagnosis of Familial Adenomatous Polyposis (FAP).
- The survey will take approximately 20-30 minutes to complete.
- The survey is anonymous and participation is voluntary.
- You can skip any question(s) you are not comfortable answering and may exit the survey at any time.
- If you exit the survey your answers will be saved and you can choose to continue answering questions at a later date while the survey is open (until [date]).
- All participants who complete the survey may enter a raffle for one of three $50 gift cards to Amazon.com. Your survey responses will not be connected to your contact information in
any way.
• The survey will be available until [date].

Who is conducting this study?
My name is Nina D’Orlando, and I am a graduate student in the Brandeis University Genetic Counseling program. This survey is part of my thesis research titled “Impact of Familial Adenomatous Polyposis: An Emerging Adult Perspective.”

This study was reviewed and approved by the Brandeis University Institutional Review Board. If you have any questions about this research project please contact Nina D’Orlando at ndorlando@brandeis.edu, or the Brandeis University faculty sponsor, Gayun Chan-Smutko, at gchansmutko@brandeis.edu.

“Click Here to Take the Survey” (will hyperlink to Qualtrics online survey).

Thank you in advance for your time and participation. We look forward to hearing from you!

Sincerely,

Nina D’Orlando, BS
MS candidate, Brandeis University
Genetic Counseling Program
Thank you for your interest in participating in our study!

This survey will ask you about your life with FAP. We ask that you be honest and open when answering questions so that we can use this information to improve the care for young adults living with FAP. Your insights and opinions are extremely valuable to us.

You will be asked a few eligibility questions and some demographic information followed by questions in the following sections:

I. Personal and Family History
II. Diagnostic Process
III. Education
IV. Career Planning
V. Relationships
VI. Family Planning
VII. Living with FAP
VIII. Genetic Counseling

A few things to remember:

- The survey is anonymous and participation is voluntary.
- You are free to skip any questions that you do not wish to answer.
- You may choose to exit the survey at any time.
- If you exit the survey your answers will be saved and you can choose to continue answering questions at a later date while the survey is open (until [date]).
- The survey will take approximately 20-30 minutes to complete.
- All participants who complete the survey may enter a raffle for one of three $50 gift cards to Amazon.com. Your survey responses will not be connected to your contact information in any way.

We appreciate you taking the time to complete this survey. If you have questions about your rights as a research participant please contact the Brandeis Institutional Review Board at 781-736-8133 or irb@brandeis.edu.

By clicking next, you consent to participate in our study.

Q2 Do you have a diagnosis of Familial Adenomatous Polyposis (FAP)?
   ☒ Yes (1)
   ☐ No (2)
   If No Is Selected, Then Skip To End of Survey

Q3 Are you currently between the ages of 18-25 years?
   ☒ Yes (1)
   ☐ No (2)
   If No Is Selected, Then Skip To End of Survey
Q4 How old are you?

<table>
<thead>
<tr>
<th>Age</th>
<th>18 (1)</th>
<th>19 (2)</th>
<th>20 (3)</th>
<th>21 (4)</th>
<th>22 (5)</th>
<th>23 (6)</th>
<th>24 (7)</th>
<th>25 (8)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>☑</td>
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</tr>
</tbody>
</table>

Q5 Which of the following do you identify as?

- Female (1)
- Male (2)
- Transgender (3)
- Prefer to self-describe: (4) ____________________
- Prefer not to say (5)

Q6 I. Personal and Family History

The following set of questions will ask you about your own medical history and your family history in relation to FAP.

Q7 How old were you when you were first diagnosed with FAP?

- 0-2 (1)
- 3-5 (2)
- 6-8 (3)
- 9-11 (4)
- 12-14 (5)
- 15-17 (6)
- 18-20 (7)
- 21-23 (8)
- 24-26 (9)
- I'm not sure (10)

Q8 Have any of your family members or relatives ever been diagnosed with FAP? (Select all that apply)

- I am the only person in my family with FAP (1)
- Yes, my mother has FAP (2)
- Yes, my father has FAP (3)
- Yes, my sister/brother has FAP (4)
- Yes, my daughter/son has FAP (5)
- Another relative has FAP. Please list all other relatives with FAP (For example: Uncle, Cousin, Grandfather, etc): (6) ____________________

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Q9 How many of your sisters or brothers have been diagnosed with FAP?
   Number of sisters with FAP: (Please enter a number) (1)
   Number of brothers with FAP: (Please enter a number) (2)

Q10 How many of your daughters or sons have been diagnosed with FAP?
   Number of daughters with FAP: (Please enter a number) (1)
   Number of sons with FAP: (Please enter a number) (2)

Q11 Who was the first person in your family to be diagnosed with FAP?
   I was the first person in my family (1)
   My mother/father was first (2)
   My sister/brother was first (3)
   My daughter/son was first (4)
   Another relative was first. Please specify (For example: Uncle, Cousin, Grandfather, etc): (5)

Q12 Have you ever been diagnosed with colon polyps?
   Yes (1)
   No (2)

Q13 How old were you when you were first diagnosed with polyps?
   0-2 (1)
   3-5 (2)
   6-8 (3)
   9-11 (4)
   12-14 (5)
   15-17 (6)
   18-20 (7)
   21-23 (8)
   24-26 (9)
   I'm not sure (10)
### Q14 How many polyps have you had?
- 1-4 (1)
- 5-19 (2)
- 20-39 (3)
- 40-59 (4)
- 60-79 (5)
- 80-99 (6)
- More than 100 (7)
- I’m not sure (8)

### Q15 Have you had a colectomy?
- Yes (1)
- No (2)

### Q16 How old were you when you had a colectomy?
- 0-2 (1)
- 3-5 (2)
- 6-8 (3)
- 9-11 (4)
- 12-14 (5)
- 15-17 (6)
- 18-20 (7)
- 21-23 (8)
- 24-26 (9)
- I’m not sure (10)

### Q17 What type of surgery did you have?
- Ileorectal anastomosis (IRA) (1)
- Ileal pouch anal anastomosis (IPAA), also known as a J-Pouch (2)
- Ileostomy (3)
- I don’t know what type of procedure I had (4)
- Other. Please list: (5) ____________________

### Q18 Have you ever been diagnosed with cancer?
- Yes (1)
- No (2)
Q19 How old were you when you were first diagnosed with cancer?
- 0-2 (1)
- 3-5 (2)
- 6-8 (3)
- 9-11 (4)
- 12-14 (5)
- 15-17 (6)
- 18-20 (7)
- 21-23 (8)
- 24-26 (9)
- I'm not sure (10)

Q20 What type of cancer were you diagnosed with?

Q21 Have you ever been diagnosed with a second (or more) cancer?
- Yes (1)
- No (2)

Q22 Have any of your family members or relatives with FAP ever been diagnosed with cancer?
- Yes (1)
- No (2)

Q23 Please specify which of your relatives with FAP have been diagnosed with cancer. (For example: Mother, Uncle, Cousin, Grandfather, etc.).

Q24 Have any of these family members or relatives passed away due to cancer caused by FAP? (Please list relatives).

Q25 II. Diagnostic Process

The following set of questions will ask you about how you were diagnosed with FAP and how you felt during that process.

Q26 How were you diagnosed with FAP?
I was **clinically** diagnosed based on my symptoms. I have **never** had genetic testing. (1)

I was **clinically** diagnosed based on my symptoms, and then I **also** had genetic testing. (6)

I **only** had genetic testing **based on my symptoms**. (2)

I **only** had genetic testing **based on my family history** (I did not have any symptoms at the time of my testing). (3)

I don't know how I was diagnosed. (5)

---

**Answer**

If How were you diagnosed with FAP? I was diagnosed clinically based on my symptoms, and then I also had genetic testing. Is Selected

Or How were you diagnosed with FAP? I only had genetic testing based on my symptoms Is Selected

Or How were you diagnosed with FAP? I had genetic testing based on my family history (I did not have any symptoms at the time of my testing) Is Selected

---

**Q27** What were the results of your genetic testing?

- Positive result (a mutation was found) (1)
- Negative result (a mutation was not found) (2)
- Uncertain result (a variant of uncertain significance (VUS) was found) (3)
- I'm not sure what the result was (4)

---

**Answer**

If How were you diagnosed with FAP? I had genetic testing based on my family history (I did not have any symptoms at the time of my testing) Is Selected

---

**Q28** When you underwent genetic testing based on your family history, how much did you feel that this was **your own decision**?

- I made this decision **entirely** by myself. Family members, providers, or others had **no influence**. (1)
- I made this decision **mostly** by myself. Family members, providers, or others had a **little influence**. (2)
- I made this decision **somewhat** by myself. Family members, providers, or others had **some influence**. (3)
- My decision was **mostly made for me** by family members, providers, or others. I **had little say**. (4)
- My decision was **entirely made for me** by family members, providers, or others. I **had no say**. (5)

---

**Q29** How much information about FAP were you given **at the time of your diagnosis**?

- A great deal (1)
- A lot (2)
- A moderate amount (3)
- A little (4)
- None at all (5)
- I have no memory of my diagnosis (6)
**Q30** How did you feel about the information about FAP you were given at the time of your diagnosis?

<table>
<thead>
<tr>
<th></th>
<th>Extremely appropriate (1)</th>
<th>Somewhat appropriate (2)</th>
<th>Neither appropriate nor inappropriate (3)</th>
<th>Somewhat inappropriate (4)</th>
<th>Extremely inappropriate (5)</th>
<th>I have no memory of my diagnosis (6)</th>
</tr>
</thead>
<tbody>
<tr>
<td>I felt that the <strong>amount</strong> of information I was given at the time of my diagnosis was... (1)</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>I felt that the <strong>type</strong> of information I was given at the time of my diagnosis was... (2)</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
</tbody>
</table>

**Q31** Please use the space below to tell us anything else about how you felt at the time of your diagnosis with FAP:

**Q32** Is there anything you wish had been different when you were diagnosed with FAP?

**Q33** III. Education

The following questions will ask you to think about how your diagnosis of FAP has or has not impacted your education.
Q34 What is the highest degree or level of education you have completed or are currently working on?
- Some High School (1)
- Completed High School / GED (2)
- Some College (3)
- Trade / Technical / Vocational Training (4)
- Associate Degree (5)
- Bachelor's Degree (6)
- Master's Degree (7)
- Doctorate Degree (8)

Q35 How do you feel about the following statement: "My diagnosis of FAP has impacted my education"
- I agree, my diagnosis of FAP has had a **positive** impact on my education (1)
- I agree, my diagnosis of FAP has had a **negative** impact on my education (2)
- I agree, my diagnosis of FAP has had **both positive and negative** impacts on my education (3)
- I disagree, my diagnosis of FAP has not impacted my education in any way (4)
- I'm not sure if my diagnosis of FAP has impacted my education (5)

Q36 Please use the space below to tell us more about how your diagnosis of FAP has or has not impacted your education:

Q37 IV. Career Planning

The following questions will ask you to think about how your diagnosis of FAP has or has not impacted your career choices and work life.

Q38 Do you feel that your diagnosis of FAP has had an influence on your choice of career?
- Yes (1)
- No (2)
- I have not yet decided on a career (3)
- I don't know (4)

Q39 Please use the space below to tell us more about how your diagnosis of FAP has or has not influenced your choice of career:

Q40 Do you feel that your diagnosis of FAP impacts your current career on a regular basis?
- Yes (1)
- No (2)
- I am not currently working (3)
- I don't know (4)
Q41 Please use the space below to tell us more about how your diagnosis of FAP has or has not impacted your current career:

Q42 V. Relationships

The following questions will ask you to think about how your diagnosis of FAP has or has not impacted your relationships with romantic partners, family members, and friends.

Q43 What is your current relationship status?
- Single (1)
- In a committed relationship (2)
- Married (3)
- Common Law (4)
- Domestic Partner (5)
- Separated (6)
- Divorced (7)
- Widow/Widower (8)

Q44 How much would you say that your diagnosis of FAP has impacted the following aspects of your romantic relationships?

<table>
<thead>
<tr>
<th></th>
<th>None at all (1)</th>
<th>A little (2)</th>
<th>A moderate amount (3)</th>
<th>A lot (4)</th>
<th>A great deal (5)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Meeting prospective partners (1)</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>Dating (2)</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>Forming meaningful connections (3)</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>Allowing yourself to be vulnerable with someone (4)</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>Sexual Intimacy (5)</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>Maintaining relationships (6)</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
</tbody>
</table>

Q45 Please use the space below to elaborate on any of the above aspects or tell us more about how your diagnosis of FAP has or has not impacted your romantic relationships:
**Q46** Please rate the following statements in terms of how they apply to only your relationships with *family members* (immediate or extended).

Living with FAP has...

<table>
<thead>
<tr>
<th></th>
<th>Not At All (1)</th>
<th>A Little Bit (2)</th>
<th>Somewhat (3)</th>
<th>Quite A Bit (4)</th>
<th>Very Much (5)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Helped me know who I can count on in times of trouble (1)</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>Makes me more willing to help others (2)</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>Helped relationships become more meaningful (3)</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>Helped me become closer to people I care about (4)</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>Helped me become more aware of the love and support available from other people (5)</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
</tbody>
</table>

**Q47** Please use the space below to tell us more about how your diagnosis of FAP has or has not impacted your relationships with family members (immediate or extended):
**Q48** Please rate the following statements in terms of how they apply to only your relationships with **friends**.

Living with FAP has...

<table>
<thead>
<tr>
<th></th>
<th>Not At All (1)</th>
<th>A Little Bit (2)</th>
<th>Somewhat (3)</th>
<th>Quite A Bit (4)</th>
<th>Very Much (5)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Helped me know who I can count on in times of trouble (1)</td>
<td>✔</td>
<td>☐</td>
<td>☑</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>Makes me more willing to help others (2)</td>
<td>✔</td>
<td>☐</td>
<td>☑</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>Helped relationships become more meaningful (3)</td>
<td>✔</td>
<td>☐</td>
<td>☑</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>Helped me become closer to people I care about (4)</td>
<td>✔</td>
<td>☐</td>
<td>☑</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>Helped me become more aware of the love and support available from other people (5)</td>
<td>✔</td>
<td>☐</td>
<td>☑</td>
<td>☐</td>
<td>☐</td>
</tr>
</tbody>
</table>

**Q49** Please use the space below to tell us more about how your diagnosis of FAP has or has not impacted your relationships with friends:
Q49 VI. Family Planning

The following questions will ask you to think about how your diagnosis of FAP has or has not impacted your decisions about having children.

One useful definition for this section:

**In vitro fertilization (IVF) with preimplantation genetic diagnosis (PGD):**
IVF with PGD is an assisted reproductive technique that allows an embryo - created using a couple's own sperm and egg - to be tested for a specific FAP-causing mutation known to be present in one partner before it is implanted into the woman's uterus. Only embryos that do not have the mutation are implanted.

Q51 Do you have children?
- Yes (1)
- No (2)

Answer If Do you have children? Yes Is Selected

Q52 Did you have your children before you were diagnosed with FAP?
- Yes (1)
- No (2)

Answer If Did you have your children before you were diagnosed with FAP? No Is Selected

Q53 Please check all that apply:
- I had my child(ren) without using assisted reproductive techniques (I conceived naturally) (1)
- I had my child(ren) using IVF with PGD (2)
- I had my child(ren) using a donor egg or donor sperm (3)
- My child(ren) is/are adopted (4)

Answer If Do you have children? Yes Is Selected

Q54 Do you plan to have more children?
- Yes (1)
- No (2)
- I haven't thought about it yet (3)
- I am undecided (4)

Answer If Do you have children? No Is Selected

Q55 Do you plan to have children?
- Yes (1)
- No (2)
- I haven't thought about it yet (3)
- I am undecided (4)
Answer If Do you plan to have more children? No Is Selected Or Do you plan to have more children? I haven't thought about it yet Is Selected Or Do you plan to have more children? I am undecided Is Selected Or Do you plan to have children? No Is Selected Or Do you plan to have children? I haven't thought about it yet Is Selected Or Do you plan to have children? I am undecided Is Selected

Q56 To what extent has your diagnosis of FAP influenced this position?
- A great deal (1)
- A lot (2)
- A moderate amount (3)
- A little (4)
- None at all (5)

Answer If Do you plan to have more children? Yes Is Selected Or Do you plan to have children? Yes Is Selected

Q57 Which of the following have you considered for having future children? (Check all that apply)
- Having children without using assisted reproductive technologies (conceiving naturally) (1)
- Adopting children (2)
- Using IVF with PGD (3)
- Using a donor egg or sperm (4)

Answer If Do you plan to have more children? Yes Is Selected Or Do you plan to have children? Yes Is Selected

Q58 Which process do you think you might undergo in the future, or are you undergoing already?
- Having children without using assisted reproductive technologies (conceiving naturally) (1)
- Adopting children (2)
- Using IVF with PGD (3)
- Using a donor egg or sperm (4)

Answer If Do you plan to have more children? Yes Is Selected Or Do you plan to have children? Yes Is Selected

Q59 To what extent has your diagnosis of FAP influenced these plans for having future children?
- A great deal (1)
- A lot (2)
- A moderate amount (3)
- A little (4)
- None at all (5)

Q60 Please use the space below to tell us more about how your diagnosis of FAP has or has not influenced your family planning:
Q61 VII. Living with FAP

The following questions will ask you to think about how your diagnosis of FAP has or has not impacted you on a personal level.

Q62 Please rate the following statements in terms of how they have been impacted by your diagnosis of FAP.

Living with FAP has...
<table>
<thead>
<tr>
<th></th>
<th>Not At All (1)</th>
<th>A Little Bit (2)</th>
<th>Somewhat (3)</th>
<th>Quite A Bit (4)</th>
<th>Very Much (5)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Helped me accept the way things work out (1)</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>Helped me learn to deal better with uncertainty (2)</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>Taught me how to adjust to things I cannot change (3)</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>Helped me take things as they come (4)</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>Helped me to look at things in a more positive way (5)</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>Helped me learn to handle difficult times (6)</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>Helped me become more comfortable with who I am (7)</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>Helped me become a stronger person (8)</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>Helped me feel better about my ability to handle problems (9)</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>Helped me become a better person</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
</tbody>
</table>
Q63 Please use the space below to elaborate on any of the above answers:

Q64 VIII. Genetic Counseling

The following questions will ask you to think about your experience(s) with a genetic counselor (if any) and/or other healthcare providers.

Q65 Have you ever seen a genetic counselor regarding your diagnosis of FAP?
○ Yes (1)
○ No (2)
○ I don't know (3)
**Q66** How would you rate your overall experience with your genetic counselor?

<table>
<thead>
<tr>
<th></th>
<th>Not at all useful (5)</th>
<th>Slightly useful (4)</th>
<th>Moderately useful (3)</th>
<th>Very useful (2)</th>
<th>Extremely useful (1)</th>
<th>I don’t remember my genetic counseling session (6)</th>
</tr>
</thead>
<tbody>
<tr>
<td>I thought that my genetic counseling session(s) was/were... (1)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
**Q67** To what extent do you agree/disagree with the following aspects of your genetic counseling session(s):

<table>
<thead>
<tr>
<th></th>
<th>Strongly disagree (1)</th>
<th>Disagree (2)</th>
<th>Somewhat disagree (3)</th>
<th>Neither agree nor disagree (4)</th>
<th>Somewhat agree (5)</th>
<th>Agree (6)</th>
<th>Strongly agree (7)</th>
</tr>
</thead>
<tbody>
<tr>
<td>The genetic counselor presented me with an appropriate amount of information (1)</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>The genetic counselor answered all of the questions that I had in a satisfactory manner (2)</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>The genetic counselor was sensitive and attentive to my needs and emotions (3)</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>The genetic counselor made me feel supported and comfortable (4)</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
<tr>
<td>The genetic counselor is someone</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
<td>○</td>
</tr>
</tbody>
</table>

Answer:

- Have you ever seen a genetic counselor regarding your diagnosis of FAP? Yes
- Is your overall experience with your genetic counselor rated as follows? - I don't remember my genetic counseling session
- Is Not Selected
I would like to see at regular intervals, as I have questions throughout my life (5)

Q68 Do you feel that you are **in need of more support** and/or **have more questions** related to FAP than your genetic counselor and/or other healthcare providers have provided or answered?

Please use the space below to share your thoughts about unmet needs you have (if any) and how you think your genetic counselor and/or healthcare team could best meet these needs:

Q69 If you would like to tell us anything else about your experience(s) with your genetic counselor and/or other healthcare providers, please use the space below:

Q70 If there is anything else that you would like to share with us about living with FAP that we have not addressed in this survey, please use the space below:
References:


7. Grubman, J., Barzi, A. (2015). We don’t know what we don’t know about adolescent and young adult patients with familial adenomatous polyposis-related colorectal cancer. *Journal of Adolescent and Young Adult Oncology, 4*(3), 105-107. doi: 10.1089/jayao.2015.0008


