Information and support needs of adolescents with Familial Adenomatous Polyposis

Rebecca Chan
Second Year Medicine (Graduate)
Notre Dame University (Sydney)

Rebecca Chan completed her Bachelor of Medical Science degree before going on to a Graduate Diploma of Genetic Counselling. During this time, she developed a strong interest in the psychological impact of genetic conditions, and hopes that her medical degree will lead her towards a career in genetic medicine.

Familial Adenomatous Polyposis (FAP) is a dominantly inherited bowel cancer predisposition syndrome presenting with hundreds of premalignant polyps in the colon. The standard form of treatment is preventative surgery which involves removal of the entire colon. The rectum and colon may also be removed. Predictive testing is usually done at ten to fourteen years of age, and surgery is recommended by the age of 20. Thus, adolescents face a wide variety of difficult decisions and situations. The aim of this review is to critically evaluate existing literature which examines the experiences of these young people and their families, including their information and support needs, psychosocial adjustments and satisfaction with current genetic services.

Introduction

Familial adenomatous polyposis (FAP) is a dominantly inherited colorectal cancer predisposition syndrome which occurs due to the inheritance of germ-line mutations in the APC tumour suppressor gene. [1] It is characterised by the progressive development of hundreds to thousands of adenomas within the colon which, if left untreated, may eventually develop into carcinomas. [2] It is thus imperative that there is early diagnosis and treatment. As the polyps can develop at very early ages, current management calls for annual endoscopic screening, starting at approximately ten years of age. [3]

As FAP is an autosomal dominant disorder with a penetrance of almost 100%, the chance of a child being born from an affected parent is one in two. [4] Therefore, in addition to screening, genetic diagnosis is available from a young age, usually ten to fourteen, and this in itself can result in issues regarding age, autonomy of the child and the desires of parents.

State-based Family Cancer Services are responsible for the provision of genetic testing, as well as subsequent counselling and support following diagnosis. Staff include genetic counsellors, medical geneticists and oncologists who work together to provide patients with information regarding their individual risk, screening options and cancer risk-reduction strategies. [5] Once a diagnosis of FAP is made, patients face complex decisions regarding care, with prophylactic surgery the standard form of treatment. This is a difficult decision to make, particularly for individuals who are otherwise young and healthy, making it challenging to weigh future cancer risks alongside the more immediate impact of surgery. [6]

Such adolescents face a wide variety of difficult decisions and situations. The aim of this review is to critically evaluate existing literature which examines the experiences of these young people and their families, including their information and support needs, psychosocial adjustments and satisfaction with genetic services currently in place. This will provide a greater understanding of the impact such a diagnosis can have, and perhaps lead to the establishment of a variety of age-appropriate services and support.

Methods

Medline and EMBASE databases were used to carry out a search of the literature for English language studies published between 1988 and 2009. The following search terms were used individually and in combination: (Familial Adenomatous Polyposis OR hereditary colorectal cancer OR adenomatous polyposis coli) AND (psychology OR psychosocial aspects OR genetic testing OR genetic counselling OR quality of life OR needs OR support OR information OR surgery). Studies were included if they described empirical research relating to the information and support needs of adolescents diagnosed with FAP, their psychosocial adjustment and satisfaction with current genetic services. Qualitative studies were included due to a limited number of quantitative studies measuring information and support needs of adolescents. Due to the paucity of literature evaluating patient satisfaction with FAP-specific genetic services, studies which evaluated familial cancer clinics regardless of the type of cancer were also considered for the purposes of this review.

Familial adenomatous polyposis: the mucosal surface of the colon is carpeted by numerous polypoid adenomas. (Copyright, UNSW Department of Pathology, from the 'Images of Disease' collection.)

Two hundred and sixty-seven articles were identified through the literature search. Forty-five were retrieved for more detailed evaluation and the final sample included fourteen studies to be reviewed. Reasons for exclusion included articles which did not consider the psychological aspects of FAP, patient cohorts which had been diagnosed with FAP as adults and studies of at-risk patients with a negative test result. Case studies, review articles, conference abstracts and commentaries were also excluded.

Surgery-related impact and needs

The many aspects of FAP mean that deciding when and how it is most suitable to treat patients is difficult and requires careful assessment. The two main forms of surgery are restorative prophylactic proctocolectomy with ileal pouch anal anastomosis (IPAA) or total colectomy with ileorectal anastomosis (IRA).

IPAA involves excision of the entire colon and rectum, leaving the anus and sphincter muscles. A small pouch is fashioned from a loop of the terminal ileum, and an anastomosis is created to attach it directly to the anus. While this heals, a temporary opening in the abdomen, called an ileostomy, is created. This is reversed a few months later, and patients begin to pass normal bowel movements through the anus. [7] In an IRA, the patient's colon is removed and the surgeon leaves 13cm of the rectum which is surgically joined to the small intestine. This is a one-stage operation with a relatively low complication rate, meaning patients generally have normal bowel function afterwards. However, for patients with FAP, there is the persistent risk of developing rectal cancer. [8]

Three papers identified through the literature search looked at the impact surgery has on the psychosocial adjustment of those with FAP. [4,9,10] Notter's study [9] used a purposive sample of women, and conducted semi-structured interviews to assess the main surgery-related impacts on psychosocial adjustment. Findings included distress at the major change brought about by surgery and a feeling of being "disfigured, less feminine, less of a woman"; shock and disgust at the appearance of an ileostomy; and a high level of pain. The study by Osterfield *et al.* [10] used pre- and post-operative interviews to prospectively examine quality of life, personal experience and adjustments in patients who underwent an IPAA. It was reported that prior to having surgery, almost all patients were in good or excellent health and most were afraid of the surgery. Post-operative feelings were similar to those found in Notter's study, with strong feelings of disgust and shame and difficulty adjusting. Severe problems in sex life were a major issue, as was deterioration in work and leisure activities. The findings by Andrews *et al.* [4] echoed those above, with adverse body image, sexual impact and physical functioning being the main factors. Further, all studies suggested that postoperative counselling may improve adjustment to the impact of the surgery, particularly in females.

These studies used three different methods of participant recruitment. That of Andrews *et al.* [4] was a convenience sample, recruited from the Hereditary Bowel Cancer Registries; Notter's [9] was a purposive sample of 50 women, and that of Osterfield *et al.* [10] was a consecutive sample of patients who underwent an IPAA at Heidelberg Medical Centre in a one-year period. All three methods leave room for sample bias, as those who agreed to participate may not be representative of the general community. Despite this, the similarity in findings provides some validity for the conclusions drawn and highlights the important need for careful pre- and post-operative counselling, particularly in the case of young people who may be undergoing surgery at a time when sexuality and sexual functioning is just being established, making it more difficult for them to deal with the issues of body image and sexuality. [9]

Information needs

The need and desire for information about the condition was a consistent theme amongst many studies. [6,11-14] The most preferred source of information was consultation with a medical or genetic expert, [11] and while most patients reported feeling that the professional had provided them with adequate information during a consultation, additional complementary information was often desired. [12] This was most often desired as written material to use as a resource for making a decision about prophylactic surgery.

One worrying aspect of this was the results of a study by Neuman *et al.*, [6] which considered the adequacy of the internet as a resource for making decisions about prophylactic surgery. In this study, a representative internet search was performed which was designed to mirror that of patient searches, and qualitatively assessed the first 50 sites from each search. Of sites identified, 75% failed to include any data relevant to the surgical treatment of FAP, and even those which the researchers considered 'excellent' did not supply details on surgical procedures or postoperative outcomes necessary for decision making. It is possible that the search may be limited due to the simplistic search strategy employed and that more information could be found if a more intensive search strategy was used. However, Neuman *et al.* highlight that this was designed to be a representative sample of available information from a patient's perspective and that it could be easily replicated.

Social support needs

Five studies were identified which documented the impact of social support on the psychological adjustment of persons diagnosed with FAP. Three used self-administered mail-out questionnaires involving itemised rating scales, and one also included open-ended questions. [4,10,14,15] One used semi-structured interviews, [16] and one used a focus group of participants to identify key issues and concerns. [14]

In their study, Esplen *et al.* [14] found that perceived social support was high amongst participants. Scores also indicated a high level of family functioning. However, these two factors were not found to be significantly associated with health-related quality of life. This result contrasts with another study by Osterfield *et al.*, [10] which found that favourable social resources helped the majority of patients to successfully adapt to the challenges and impairments of undergoing surgery for FAP. Similarly, Carlsson *et al.* [15] determined that perceived social support helped to maintain a direct positive effect on mental health-related quality of life for patients with hereditary cancer. It should be noted, however, that the sample surveyed by Esplen used a purposive sample, with very specific characteristics – a diagnosis of both FAP and desmoid tumour, whereas Carlsson *et al.*'s study consisted of a convenience sample and encompassed various types of hereditary cancer, with colorectal cancer being just one form. Thus, it is difficult to directly compare the results of the two studies given the possibility of bias as a results of sampling methods.

However, Carlsson *et al.* concluded that the social support which made the most significant contribution to health-related quality of life was self-esteem and appraisal support - the perceived availability of someone with whom to discuss one's problems. [17] This indicates that it is important for individuals to feel loved, valued and competent. [15] This is supported by Esplen *et al.*'s finding that marital status was a significant and independent predictor for better adjustment, and thus marital status may in fact be a proxy for social support in their study. [14] This finding is also matched by that of Andrews *et al.*, [11] who restricted their study to a FAP affected population and found that being single is associated with higher levels of distress. As FAP is generally diagnosed in early adolescence, it is less likely that these young people will have this form of relationship. This therefore suggests a need to create more age-appropriate resources which may help to provide a further source of support. In response to open ended questions regarding improvements in support services, [11] some adolescents requested support groups be set up locally or on the internet and this is an important possibility to investigate.

Many of these studies however had small sample sizes and several of the sample types accessed may introduce a level of bias. For example, both Esplen *et al.* [14] and Carlsson *et al.* [15] used study groups recruited from clinics, and thus may have been high-functioning, resourceful people not representative of the population. Similarly, the sample of Andrews *et al.* [4] was contacted through the Australian Hereditary Bowel Cancer Register, and those who decline to be on the register may gave different interests to the study group in terms of accessing further social support.

Positive family functioning has also been found to serves as an indicator for better adjustment to FAP. [11,14,16] Duncan *et al.* [16] reported family relationships as being both a positive and negative factor associated with a positive test result for FAP. Patients felt that identifying with other gene-positive family members could be both harmful and beneficial, while others indicated they felt more distanced from particular family members. Esplen *et al.* [14] explain this by reasoning that experience with FAP prior to receiving a diagnosis will only be beneficial if such experiences were positive. If a family member coped poorly with their diagnosis, this will negatively impact the newly diagnosed patient's perception of the illness and their own subsequent adjustment. This suggests that more family-orientated support and counselling may be necessary in the management of FAP for adolescents. This is particularly so given that 77% of one study cohort [11] used family members as an additional source of information and support when they felt that their needs had not been met. It should be noted, however, that Duncan *et al.* [16] interviewed only a small group of people seen at the same genetics service, and Esplen *et al.* [14] used a focus group of seven people to extrapolate some of the aspects of the questionnaire used in their study. Their results may therefore have been biased in that participants may have been those with the greatest

number of concerns.

Satisfaction with current genetic services

Few studies evaluate patient satisfaction with FAP-specific genetic services. Thus, for the purposes of this review, studies which evaluated familial cancer clinics regardless of the type of cancer were considered. Six of the thirteen studies evaluated patient satisfaction with current genetic services and the fulfilment of client needs. [11-13,18-20] However, only four sample sets shall be considered as only four cohorts are described amongst these studies. Further, three of these four samples [12,13,18-20] are based on the one clinic, and thus, an element of bias needs to be considered when analysing the results including the sociodemographic and medical characteristics of those particular participants and service providers. Only the study by Andrews *et al.* [11] involves a slightly broader demographic, having recruited participants from four Australian hereditary bowel cancer registries located in four different states. Bias can still not be ruled out, as those who decline to be a part of the registry may have different opinions to the study group.

The main method of data collection employed was questionnaires. Two study cohorts completed both pre- and post-clinic questionnaires, [12,13,19,20] while the other two completed just one questionnaire post-visit. [11,18] Andrews *et al.* [11] carried out a retrospective study while the others were all prospective. This introduces some bias to the levels of satisfaction participants felt, as the memory would be more vivid for participants in prospective studies. As well, there is a chance for recall bias – patients who have had bad experiences may remember them more than those who had good experiences.

A pilot study on experiences of patients and family members in Amsterdam [18] found that a large majority of respondents were satisfied with the care provided by the geneticist. However, only using a post-clinic questionnaire does not allow for comparison of expectations and participant needs before and after clinic attendance. This issue is more clearly addressed in the studies by Pieterse *et al.* [19,20] and Collins *et al.* [12]

Information needs were generally satisfied in the genetic counselling session. In their study, Collins et. al [12] found that upon arrival at the clinic, most participants wanted 'very detailed' information about each topic; post-clinic, most thought they had received 'enough', and very few indicated that they would need to go back for more. Similarly, Pieterse *et al.* [20] found that the most effective method of communication, in terms of meeting the needs of participants, was the provision of medical information. Clarification of a patient's risk perception helped to ease the worries of some, though it was usually those who were older or less educated. It was thought that perhaps younger people have greater knowledge of genetic concepts and thus a higher expectation of the clinic. [13] For FAP patients, it would therefore be important to ensure that sessions were adapted to suit the information needs of these younger people.

It was further added that additional sessions with a genetic counsellor might be helpful. This was supported by the Andrews *et al.* [11] study, which found that those who had received genetic counselling – on average five years ago, and before the age of twenty-three – could recall only approximately two thirds of the medical and risk information presented, and may not have received information during the consultation for future issues such as childbearing. Thus, there is a need for genetic services which occur on an ongoing basis. The 'before and after' style of data collection used in the design of both Pieterse *et al.*'s [19,20] and Collins *et al.*'s [12] studies may have benefited from a follow-up questionnaire some time after to better assess patient satisfaction with time.

Conclusion

As predictive testing for FAP is usually offered during early adolescence, decisions about surgery and other treatment and management options need to be considered when the patient is very young. Surgical management is highly invasive, making it difficult for otherwise young and healthy individuals to weigh up the future risk of cancer against the more immediate impact of surgery. With few available resources for guidance, patients may be underprepared for the situation.

Poor psychological adaptations are a concern, considering the already documented difficulty in psychosocial adjustment for adults to a FAP diagnosis. [4] The fact that marital status has been found to be a significant and independent predictor for better adjustment [15] creates the question of whether young people, who are less likely to have such a relationship at the time of diagnosis, will be able to find this source of support. While there have been studies in young people which look at their support needs, [4,11] these have been retrospective. This means participants may have had difficulty recalling certain details. It would be useful to ascertain where adolescents feel their main sources of support are, and how this impacts their adjustment to the diagnosis, management and treatment of FAP.

In addition, family functioning has been found to have an impact on the adjustment of adolescents with FAP, particularly when a family member has had a positive or negative experience with the condition. It may be useful to explore the relationships between the affected adolescent and other family members, and whether family-orientated support and counselling may be beneficial.

Considering that diagnosis and surgery for FAP typically occurs in late adolescence or early adulthood, it is disappointing to note that very few papers focus specifically on FAP in a young cohort. Satisfaction with current genetic services has been assessed primarily through studies with a mean participant age of forty. [12,13,19,20] Young people are believed to have greater knowledge of genetic concepts and therefore a higher expectation of clinics, [12] along with a need to focus on future potential issues such as childbearing. Therefore, their experiences of genetic services may differ greatly to that of an older population. This means it is extremely important to determine if adolescents feel their information and support needs are being met in a medical consultation, what additional resources would be of use and whether they feel follow-up sessions would be appropriate, particularly to address post-operative concerns such as impact on sexual functioning and body image.

Correspondence

R Chan: rchan@nd.edu.au

References

[1.]Charames GS, Ramyar L, Mitri A, Berk T, Cheng H, Jung J, *et al.* A large novel deletion in the APC promoter region causes gene silencing and leads to classical familial adenomatous polyposis in a Manitoba Mennonite kindred. Hum Genet 2008;124(5):535-41.
[2] Hegde MR, Roa BB. Detecting mutations in the APC gene in familial adenomatous polyposis (FAP). Curr Protoc Hum Genet 2006;10(10.8).
[3] Codori AM. Genetic testing for hereditary colorectal cancer in children: long-term psychological effects. Am J Med Genet 2003;116:117-28.
[4] Andrews L, Mireskandari S, Jessen J, Thewes B, Soloman M, Macrae M, *et al.* Impact of familial adenomatous polyposis on young adults: quality of life outcomes. Dis Colon Rectum 2007;50:1306-15.
[5] The Centre for Genetics Education, Family Cancer Services, NSW Health [online]. 2007 [cited 2007 Aug 18]; Available from: URL:www.genetics.edu.au
[6] Neuman H, Cabral BS, Charlson ME, Temple LK. Is internet information adequate to faciliate surgical decision-making in familial adenomatous polyposis. Dis Colon Rectum 2006;50:2135-41.
[7] Parks A, Nicholls R. Proctocolectomy without ileostomy for ulcerative colitis. Br Med J 1978;2:85-8.
[8] Ambroze W, Orangio G, Lucas G. Surgical options for Familial Adenomatous Polyposis. Semin Surg Oncol 1995;11:423-7.
[9] Notter J. Preparing for loop ileostomy surgery: womens accounts from a qualitative study. Int J Nurs Stud 2006;43(2):147-59.
[10] Osterfield N, Kadmon M, Brechtel A, Keller M. Preoperative and postoperative quality of life in patients with Familial Adenomatous Polyposis. Dis Colon Rectum 2008;51:1324-30.
[11] Andrews L, Mireskandari S, Jessen J, Thewes B, Soloman M, Macrae M, *et al.* Impact of familial adenomatous polyposis on young adults: attitudes toward genetic testing, support, and information needs. Genet Med 2006;8(11):697-703.

[12] Collins V, Halliday J, Warren R, Williamson R. Assessment of education and counselling offered by a familial colorecal cancer clinic. Clin Genet 2000;57:48-55.
[13] Collins, V., Halliday, J., Warren, R., Williamson, R. Cancer worries, risk perceptions and associations with interest in DNA testing and clinic satisfaction in a familial colorectal cancer clinic. Clin Genet 2000;58:460-8.
[14] Esplen M, Berk T, Butler K, Gallinger S, Cohen Z, Trinkhaus M. Quality of life in adults diagnosed with familial adenomatous polyposis and desmoid tumor. Dis Colon Rectum 2004;47(5):687-95.
[15] Carlsson A, Bjorvatn C, Engebretsen L, Berglund G, Natvig G. Psychosocial factors associated with quality of life among individuals attending genetic counseling for hereditary cancer. J Genet Couns 2004;13(5):425-45.
[16] Duncan R, Gillam L, Savulescu J, Williamson R, Rogers JG, Delatycki MB. "You're one of us now": Young people describe their experiences of predictive genetic testing for Huntington disease (HD) and familial adenmatous polyposis (FAP). Am J Med Genet 2008;148C:47-55.
[17] Brookings J, Bolton B. Confirmatory factor analysis of the interpersonal support evaluation list. Am J Community Psychol 1988;16(1):137-47.
[18] Bleiker E, Aaronson N, Menko F, Hahn D, Van Asperen C, Rutgers E *et al.* Genetic counselling for hereditary cancer: A pilot study on experiences of patients and family members. Patient Education and Counselling 1997;32:107-16.
[19] Pieterse A, Van Dulmen A, Beemer F, Bensing J, Ausems M. Communication in cancer genetic counselling: does it reflect counselees previsit needs and preferences?. Br J Cancer 2005;92:1671-8.
[20] Pieterse A, Van Dulmen A, Beemer F, Bensing J, Ausems M. Cancer genetic counseling: communication and counselees post-visit satisfaction, cognitions, anxiety, and needs fulfillment. J Genet Couns 2007;16(1):85-96.