Desmoid fibromatosis through the patients' eyes: Time to change the focus and organisation of care?

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Abstract

Purpose: Desmoid Fibromatosis (DF) is a rare, unpredictable disease with no established, evidence-based treatments. Individual management is based on consensus algorithms. This study aimed to examine the specific health-related quality of life challenges faced by DF patients, current experiences and expectations of care.

Methods: Twenty-seven DF patients were purposively sampled from The Royal Marsden Hospital. Two focus groups and thirteen interviews (males 12, females 15; mean age at study 39.5 years) explored health-related quality of life issues and experiences of healthcare. Thematic content was analysed.

Results: Discussions revealed 4 key themes (diagnostic pathway; treatment pathway; living with DF; supportive care). Diagnostic delay resulted from lack of recognition by patients and healthcare professionals. Some patients received an initial diagnosis of cancer, causing significant distress. Treatment decisions were challenging and patients experienced uncertainty among clinicians about optimal therapies. Side-effects of treatment were severe, including fatigue, nausea, anorexia, low libido and depression. Pain was the most debilitating symptom and dependency on painkillers was a significant concern. Functional limitation and restricted mobility frequently affected daily activities. Patients experienced difficulty accomplishing their role in society; relationship problems, caring for children, employment and financial difficulties. Social isolation and lack of understanding were common. The psychological impact of this "life-changing and life-long" condition was profound. All patients requested knowledgeable healthcare professionals, more information, continuity of care and peer support.

Conclusions: DF patients face complex physical, psychological and practical challenges. Comprehensive care services are needed. Increasing awareness may help to improve diagnostic pathways and overall patient experience.

Keywords: desmoid-type fibromatosis; health-related quality of life; pain; functional limitation; supportive care

Introduction

Desmoid-type fibromatosis (DF) is a rare, difficult to manage mesenchymal tumour, which accounts for 3% of soft-tissue tumours[1, 2]. The reported incidence is 2-4 individuals per million per year[3]. DF usually affects patients aged between 15- 60 years, with a peak around 30 years[4]. Approximately 85-90% of cases are associated with mutations in the β-catenin gene (*CTNNB1)*, leading to accumulation of β-catenin protein[5]. In a minority of cases (5-10%), DF can arise in the context of familial adenomatous polyposis (FAP)[5]. Tumours can originate in almost any area of the body, most frequently extremities, trunk, abdominal cavity, abdominal wall, head and neck[5, 6]. Although not by definition 'malignant', the morbidity caused by this disease and its associated treatments is far from 'benign'[7]. The clinical course varies; tumours may progress rapidly, remain stable for many years or undergo spontaneous regression[8, 9]. Furthermore, there is no linear relationship between symptoms and size of DF lesions.

Due to its heterogeneous behaviour there are no established, evidence-based treatment guidelines and decisions are based on consensus recommendations[5, 6]. Treatment options include surgery, radiotherapy, chemotherapy, hormonal agents and non-steroidal anti-inflammatory drugs (NSAIDS)[5, 6]. The marginal clinical benefit of these treatments coupled with potentially serious adverse effects leads to challenging management decisions. Angiogenesis inhibitors, such as pazopanib, have shown activity in a small case-series, however have yet to be validated in larger clinical trials[10]. More recently, the gamma-secretase inhibitor PF-03084014 has shown promising activity with clinically-meaningful symptomatic benefit and durable responses in small numbers of patients[11,12]. The optimum duration of systemic therapy remains undefined and there are issues related to access and reimbursement of novel agents, which inevitably play an important role in the management of specific patients. Over the past decade, most specialist centres have moved away from primary surgery towards a first-line 'watch-and-wait' policy for asymptomatic tumours. In contrast to radical and often mutilating surgery, preservation of function and avoiding disfiguring outcomes has become a priority in the management of DF patients[5, 6].

Although desmoid tumours are incapable of metastasizing (often described as benign disease), patients sometimes undergo treatments similar to cancer patients, and experience symptoms of pain and reduced mobility and/or ability to carry out everyday activities. However, the patient's health-related quality of life (HRQoL) has not been studied extensively. The care for DF patients may be compromised by a lack of insight into the physical, psychosocial and practical challenges they encounter. In order to optimise supportive care services, we must gain better understanding of the DF patient journey. Therefore, the aims of this study were to examine the specific challenges faced by DF patients, current care experiences and expectations of care.

Methods

Sample and procedure

Adult DF patients were recruited from the Royal Marsden Hospital sarcoma outpatient clinic. Patients were eligible if they were (1) aged ≥18 years; (2) diagnosed with histologically proven DF (primary or recurrent disease, any primary location, any management plan including active surveillance); and (3) could communicate in English. Patients with significant cognitive impairment or mental health problems, as determined by the referring HCP, were excluded. We employed purposive sampling to obtain a varied sample with regard to age and tumour localization.

The medical consultant, clinical nurse specialist or physiotherapist offered potential participants an information letter. With their permission, a member of the research team (OH/EY) contacted them to ask if they were willing to participate. Two focus group meetings of 120 minutes were scheduled at the hospital and interviews were conducted until saturation (no new topics appeared) was reached. Patients for the interviews were recruited according to an inductive design, indicating that the results

of the focus groups and previous interviews shaped the recruitment for the next interviews. All participants gave written informed consent prior to the focus group or interview. Participants completed demographic, clinical and HRQoL questions before the focus group or interview. HRQoL was assessed with the EORTC QLQ-C30[11]. This contains five functional scales (physical, role, cognitive, emotional and social functioning), a global health status/QoL scale, three symptoms scales (fatigue, nausea/vomiting, pain) and six single items assessing dyspnea, insomnia, anorexia, constipation, diarrhea, and financial impact. Each item is scored on a four-point Likert-scale, except general QoL, which has a seven-point Likert-scale. Scores were linear transformed to a 0-100 scale[12]. Higher score on functional scales and general QoL scale means better functioning and HRQoL, whereas a higher score on the symptom scales mean more complaints. Although developed for cancer patients, these questions can be appropriate for any respondent, e.g. 'Do you have any trouble taking a long walk' (physical functioning). The study was deemed exempt from full review and approval by a research ethics committee (CCR the Royal Marsden Hospital), but was approved by the service evaluation committee of the Royal Marsden Hospital (SE606).

Focus groups and interviews
Two focus groups and thirteen interviews were conducted by two members of the research team (EY and OH). A semi-structured interview schedule was used (Attachment 5). Topics and questions were based on clinical experience and literature[5, 6].

Data analysis and reporting
The consolidated criteria for reporting qualitative research (COREQ) were followed to ensure accuracy of this qualitative study. The interviews were transcribed smooth verbatim using F4 software. Data analysis was conducted by two coders (OH and EY) via ATLAS.ti 8.0 using thematic analysis [13, 14]. Both coders read the transcripts several times, highlighted sections that were related to the research objectives, independently selected and coded these into key themes and subthemes. Thereafter, the coders discussed their findings, refined the key themes and subthemes and resolved differences until consensus was reached. All quotes were anonymised.

Results

Participants
Twenty-seven patients participated (14 in focus groups and 13 interviews). Sociodemographic and clinical characteristics are presented in Table 1. DF patients scored low on all functioning scales (physical 74.4; role 55.8; social 52.8; cognitive 70.1; and emotional 56.9) and global QoL scale (56.9) of the EORTC QLQ-C30 and exceptionally high on the symptom scales pain (59.0), insomnia (56.9), fatigue (53.6) and financial difficulties (31.9).

DF patient journey
Analysis of the transcripts resulted in 4 main themes (diagnostic pathway; treatment pathway; living with DF; supportive care) and 12 subthemes (Figure 1).

Diagnostic pathway (Appendix 1)
Most DF patients reported delay in diagnosis. Patients frequently did not recognise the serious nature of their own symptoms. In addition, patients experienced a long diagnostic trajectory within primary and secondary healthcare accompanied by frustration and uncertainty. Patients felt that their symptoms were not taken seriously and were transferred from 'hospital to hospital to hospital' to receive the correct diagnosis of DF. Several patients were initially given a diagnosis of 'cancer' or 'malignant sarcoma' and informed that the anticipated prognosis was bleak. These patients experienced shock and distress due to a cancer diagnosis and the ordeal of sharing this news with their families. Others found that receiving an incorrect diagnosis of cancer resulted in meaning-

making, leading them to reassess the direction of their lives and consider positive lifestyle choices such as applying for a 'dream job' or instigating a 'health kick'.

Treatment pathway (Appendix 2)
DF patients experienced uncertainty among clinicians about the optimal sequence of therapies. Patients found decisions challenging, including potentially debilitating surgery, radiotherapy or cytotoxic chemotherapy without clear evidence of treatment effectiveness. Several patients felt that there was a lack of guidance from healthcare professionals (HCP) about the most appropriate treatment and desired someone with knowledge and experience to make the best decision on their behalf. With the benefit of hindsight, some patients would have made different decisions and may have benefited from talking to others with DF. Treatment-related side effects were common and severe. Patients reported profound fatigue, loss of concentration, daily nausea, vomiting and loss of appetite, lack of libido and depression. Those who received chemotherapy suffered from 'horrendous' side effects including mouth 'blisters', 'vomiting daily' and 'feeling like death'. Radiotherapy induced pulmonary fibrosis caused 'life-changing' breathlessness on minimal exertion.

Living with desmoid-type fibromatosis (Appendix 3)
Pain was the most debilitating symptom mentioned by all patients. Dependency on painkillers was a significant concern for several patients and others found that pain was unresponsive to analgesics. Patients reported functional limitation due to pain and restricted mobility, affecting daily activities such as bathing, dressing and carrying children. Others had to give up hobbies such as horse riding and rock-climbing. The unrelenting nature of pain and its impact on patients' lives led to substantial psychological distress.

In addition to physical limitations, DF patients experienced difficulty accomplishing their role in society. The side effects of treatment and long-term effects of the disease caused relationship problems for several patients. Loss of libido affected sexual relationships and some reported break-down of their relationship or marriage. Patients did not want to feel as though they were a burden on their partner or family and tried to cope alone. DF patients also felt that their parental role was affected including difficulty breastfeeding, playing games with their children and depending on their partner or parent for childcare. Others felt guilty, as they did not want to burden their parents, siblings or children to the detriment of other family members. Suicidal thoughts were expressed by those who found the burden too much to bear (N=3).

DF patients experienced changes to their employment due to their disease and found it frustrating being unable to work and wanting to feel 'normal'. Some patients felt discriminated due to enforced sick leave, which was demoralizing and led to loss of progression within their job role. In addition, financial difficulty resulting from loss of employment and hospital travel expenses, was a significant concern for several patients.

Patients felt that those around them had difficulty understanding their disease, leading to frustration. At times, if not receiving active treatment, others perceived that the disease was not 'serious' and patients described that concern went 'away when it's not malignant'. Social isolation was considerable and many felt 'alone'. Patients also felt socially behind their peers, who had greater confidence due to more experiences.

Patients described the psychological demands of living with a chronic disease that is 'life-changing and life-long'. The uncertain behavior of DF and fear of recurrence were mentioned by most patients. Others felt hyper-vigilant to any changes in pressure, sensation or pain, worrying that this signified deterioration in their disease. Negative body image was common, especially among women who felt 'less feminine' due to lumps or surgical scars. Many patients experienced depression and negative

thoughts due to their disease and treatment, feeling 'down and down and down' and describing that they could not 'be in that state forever'. Mood changes were common among patients.

Supportive care (Appendix 4)
All patients found it frustrating that many HCP lacked knowledge about their rare disease. Many wanted a concise information leaflet that they could present to their friends, family and others. Although treatments were similar to cancer patients, almost all patients felt that there was no specific support available for them and were turned away by cancer charities and organisations. Being treating in a cancer-specific hospital, patients felt uncomfortable, like 'a fraud', as if they would 'waste time' because others may have 'terminal conditions'. Several patients indicated that the information provision by HCP was largely missing or could be improved. Patients were frustrated by lack of continuity and differing opinions offered by clinicians at each hospital appointment. Patients indicated that adequate psychological support was not accessible and felt it would be comforting to meet others with the same condition, whom they could contact for support and advice. They considered that face-to-face contact or online groups would be useful.

Discussion

In the present study we found that DF patients experienced many challenges related to their diagnosis and treatment. The most commonly reported difficulties were delayed diagnosis, treatment uncertainty, treatment-related side-effects, debilitating symptoms, resulting in limitations in physical and psychosocial functioning, and financial challenges.

Most patients described a prolonged diagnostic pathway. Diagnostic delay is a common feature of rare diseases, such as DF, whereby the low prevalence leads to reduced public awareness, and lack of expertise outside specialist centres[15]. Patients' health-seeking behaviours are complex and can be influenced by physical, social and psychological factors[16]. The clinical presentation of DF is variable. Non-specific symptoms (e.g. pain or swelling) and very low prevalence are associated with diagnostic difficulty and error in primary care[17]. Most soft tissue lumps are not 'aggressive' or malignant, and therefore clinical suspicion among HCP may be low[18]. It is not known whether diagnostic 'delay' influences overall outcomes for DF patients, however it is undeniable that a lengthy diagnostic pathway can cause significantly psychological morbidity. Duration of diagnostic delay has been shown to be positively correlated with psychological distress among cancer patients[19]. Several DF patients received an initial diagnosis of 'cancer' which caused significant anxiety. Others found that confrontation with a potentially life-threatening disease provided meaning-making, prompting them to make positive changes to their lives. Regrettably, inaccurate diagnosis is not uncommon; a retrospective review of 320 specimens from DF patients found that up to one third of cases were incorrectly ascribed a diagnosis of DF[20].
Greater awareness of DF is needed among the public and HCP. The analogy of a 'golf-ball' has been used to increase awareness of the potentially serious nature of soft tissue lumps for soft tissue sarcomas[21]. HCP should also have easily accessible, accurate information to improve early diagnosis and provide appropriate support to patients and their families[15].

Patients reported that uncertainty among HCP about the optimal treatment strategy led to anxiety and loss of confidence in HCP. Due to the rarity of DF and lack of known meaningful endpoints, randomised phase 3 clinical trials have not been possible thus far[5]. In keeping with the standard oncological approach, most randomised phase 2 studies evaluate treatment-efficacy based on radiological endpoints and may not include other aspects which are relevant to the DF patient's well-being[22, 23]. Consensus guidelines, based on best available evidence, aim to clarify and unify the approach to the management of DF[5]. An initial watchful waiting period is recommended followed by a multidisciplinary treatment plan for those with clearly progressive disease[5, 24]. Watchful-

waiting is associated with uncertainty, fear, stress and anxiety in patients with cancer and other non-malignant conditions[25].
Patients who received an array of anti-cancer therapies, reported that side-effects were underplayed by HCP. Discrepancy between patient reported outcomes and clinician-assessed toxicity has been well-described in patients treated with anti-cancer therapies[26]. Clinicians frequently underestimate the frequency and severity of side-effects which can lead to inadequate supportive care[27]. Coping with the uncertainty of treatment efficacy combined with the unpredictable natural history was extremely difficult. The treatment of rare diseases such as DF is challenging. Therapeutic options are often expensive and lack investment from pharmaceutical companies due to the small potential market. A number of drugs for DF patients, including gamma-secretase and tyrosine kinase inhibitors have shown promise in early phase clinical trials, and comparative, randomised data are needed[5]. Defining the optimal, clinically meaningful, endpoint of such studies will be challenging. Unfortunately, tyrosine kinase inhibitors that have shown benefit, are not easily accessible for DF patients as they are not licensed or reimbursed in all European countries[5]. In the absence of validated predictive factors an individualised approach is recommended for all DF patients[24]. The risks and benefits of treatment should be carefully considered, integrating tumour location and characteristics with individual patient factors and preferences. Routinely measuring HRQoL in clinical practice and in research trials will lead to a better understanding of treatment efficacy from the patient perspective.
Due to the low prevalence and the lack of expertise DF patients are forced to become "knowledgeable" experts about their own disease. Appropriate information is largely lacking and should be developed. Careful counselling at a specialist center, where HCP have knowledge about DF, is mandatory and should be offered to all patients affected by DF from the time of their diagnosis[5, 6]. Treatment and follow-up at a specialist center will also improve the continuity of care.

Pain was the most debilitating symptom among DF patients. The mechanisms of pain in DF are complex and multifactorial and there is no direct correlation between pain and disease progression[5, 6]. Pain also commonly affects daily functioning in cancer survivors, especially in the first few years after treatment[28]. Numerous recommendations, which have been made to reduce pain in cancer survivors, may also be applicable to DF patients, including pain-screening and pharmacotherapy[29-32]. Due to the chronicity of DF there is concern about the long-term adverse effects of painkillers, risks of misuse, overdose and addiction. In our study some DF patients felt physically and psychologically-dependent on painkillers. As with chronic, nonmalignant pain, multimodal interventions that incorporate nonpharmacological therapies could be integrated into therapy for DF patients, aiming to restore functionality where possible.

This study showed that DF can have a considerable impact on relationships, social roles and functioning. A diagnosis of DF and/or its treatment affected finances and employment, through a reduced ability to work, need to adapt roles in the workplace and sometimes even job loss. Patients also voluntarily changed employment after self-reflection about life's priorities. The ability to work following DF is important for maintaining self-respect, identity, and living conditions[33]. Financial strain due to lost productivity and medical costs, including travelling costs to the hospital can lead to dependence on family members and have an adverse effect on patients' social relationships. Patients do not want burden their partner, family or friends. DF patients sometimes feel isolated and miss out on important social activities. Most people have little familiarity with the physical and psychosocial impact of DF and may not know how support a patient. Taking into account the relatively young patient population, patients should be supported to maintain a 'normal' life[5, 6]. In addition to "curing" the disease, the imperative long-term goal of any treatment strategy for DF should include maintenance or reintegration of a patient to social and work roles as far as possible. Interventions that promote social integration for DF patients (e.g. psychological and social support) and vocational counseling seem indicated.

Improved peer support may have helped DF patients to relate their experience to that of others with the same disease. For example, (online) peer-support groups and disease-specific information portals for patients with other rare disease have been shown to significantly reduce feelings of social isolation, improve knowledge, self-efficacy, problem-solving skills and effective interpersonal interactions[34-36]. The Royal Marsden Hospital is developing a regular clinic specifically for DF patients, which will also enable patients to meet each other at the hospital. In addition, multi-disciplinary support, involving pain specialists, physiotherapists, social workers and psychologists, should be offered to DF patients when indicated. Many patients in our study felt uncomfortable that they had to attend a clinic for patients with cancer and therefore a dedicated monthly multidisciplinary clinic (depending on the local resources) may provide a more suitable environment for their outpatient appointments.

Several limitations of this study should be taken into account. Firstly, most participants received some form of treatment and therefore may not adequately reflect the full range of DF patients. Patients on a watch and wait policy may have less problems and no treatment-related side effects. In addition, data on ethnic background were lacking, there were no Afro-Caribbean and Asian participants. Due to the limited number of patients we could not examine gender differences with statistical probability, however generally female patients had more problems with body image and talked more about what other people thought about their disease, while males reported uncertainty about disease growth or recurrence more often. In addition, our study sample may reflect patients who feel comfortable talking about their patient journey. We relied on patient self-report for the clinical characteristics of their disease. Our results should also be interpreted in the context of financial constraints of the UK National Health Service (NHS): leading to reduced access to certain drugs (e.g. pazopanib).

Further research is needed, preferably an international, population-based study which will integrate patient-reported outcome data with objective clinical examination, radiological findings and potentially molecular characteristics. This will allow greater insight into potential prognostic factors, treatment efficacy from the patients' perspective and enable provision of a more holistic approach to care. Overall, DF patients face complex physical, psychological and practical challenges. Comprehensive services including improving peer-support networks are needed. Increasing awareness of this debilitating disease may also help to improve diagnostic pathways and overall patient experience.

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Table 1: Sociodemographic and clinical characteristics of study participants

Sociodemographic characteristics	
Age time diagnosis – mean (SD; range)	34.1 (15.5; 11-70)
Age at time study - mean (SD; range)	39.5 (13.7; 23-74)
Sex N (%)	
Male	12 (44.4%)
Female	15 (55.6%)
Ethnic background N (%)	
White	21 (77.8%)
Mixed	2 (7.4%)
Black or black British	2 (7.4%)
Chinese	1 (3.7%)
Other	1 (3.7%)
Relationship status N (%)	
Single	5 (18.5%)
Dating/in a relationship	8 (29.6%)
Married	10 (37.0%)
Living common	3 (11.1%)
Separated	1 (3.7%)
Caring responsibilities children under 18 years N (%)	
Yes	9 (33.3%)
No	18 (66.7%)
Highest formal education N(%)	
Less than compulsory school	1 (3.7%)
Compulsory school	1 (3.7%)
Further education	9 (33.3%)
Higher education – undergraduate	7 (25.9%)
Higher education – postgraduate	7 (25.9%)
Professional qualification	1 (3.7%)
Other	1 (7.1%)
Employment status N (%)	
Employed full-time	14 (51.9%)
Employed part-time	5 (18.5%)
Looking after home or family	2 (7.4%)
On temporary medical leave / disability	3 (11.1%)
Unemployed	2 (7.4%)
Missing	1 (3.7%)
Clinical characteristics (self-report)	
Location of desmoid N (%)	
Abdominal wall	3 (11.1%)
Intra-abdominal	3 (11.1%)
Retroperitoneal/pelvic	3 (11.1%)
Extremity/girdles/chest wall	11 (40.7%)
Head and neck / intrathoracic	3 (11.1%)
Shoulder/scapula	4 (14.8%)
Treatment received N (%)	
Watch and wait policy	9 (33.3%)
Surgery	12 (44.4%)
Radiotherapy	6 (22.2%)
Chemotherapy	11 (40.7%)
NSAIDS	4 (14.8%)
Hormonal treatment	10 (37.0%)
TKI	2 (7.4%)
Pain management	10 (37.0%)
Physiotherapy	6 (22.2%)
Occupational therapy	2 (7.4%)
Recurrent disease N (%)	
Yes	11 (40.7%)
No	16 (59.3%)
Comorbid disease N (%)	
None	9 (33.3%)
One	15 (55.6%)

Two or more	3 (11.1%)
Health-related quality of life functioning scores 0-100 – Mean (SD)[a]	
Global quality of life	56.9 (22.9)
Physical functioning	74.4 (24.4)
Role functioning	55.8 (37.8)
Social functioning	52.8 (40.4)
Cognitive functioning	70.1 (29.5)
Emotional functioning	56.9 (27.2)
Health-related quality of life symptom scores 0-100 – Mean (SD)[b]	
Fatigue	53.6 (36.2)
Nausea	13.2 (19.6)
Pain	59.0 (39.6)
Dyspnea	19.4 (32.5)
Insomnia	56.9 (42.2)
Appetite	14.5 (22.1)
Constipation	16.7 (31.1)
Diarrhea	16.7 (29.5)
Financial difficulties	31.9 (33.3)

[a] Higher scores indicate better functioning; [b] Higher scores indicate more symptoms

Figure 1: Schematic representation of main findings

Theme 1: Diagnostic pathway

- Subtheme 1: Delay in diagnosis
  - *Category 1:* Patient delay
  - *Category 2:* Doctors' or health care system delay - symptoms not taken seriously
- Subtheme 2: Wrong diagnosis
  - *Category 1:* Negative experience
  - *Category 2:* Positive experience – meaning making

Theme 2: Treatment pathway

- Subtheme 1: Treatment decision making
- Subtheme 2: Direct treatment side effects

Theme 3: Living with desmoid-type fibromatosis

- Subtheme 1: Physical functioning
  - *Category 1:* Pain and discomfort
  - *Category 2:* Dependency on pain-killers
  - *Category 3:* Functional limitations because of pain
  - *Category 4:* Mobility
- Subtheme 2: Role limitations
  - *Category 1:* Marital functioning
  - *Category 2:* Family functioning
  - *Category 3:* Job/finances
- Subtheme 3: Social functioning
  - *Category 1:* Lack of understanding
  - *Category 2:* Social isolation
- Subtheme 4: Emotional functioning
  - *Category 1:* Living with a chronic disease
  - *Category 2:* Uncertainty – tumor behavior - recurrence
  - *Category 3:* Mood
  - *Category 4:* Body image

Theme 4: Supportive care

- Subtheme 1: Finding your way in the health care system with a rare disease
- Subtheme 2: Information provision
- Subtheme 3: Continuity of care
- Subtheme 4: Peer support

Appendix 1: Quotes Theme 1 Diagnostic pathway

Subtheme 1: Delay in diagnosis
Category 1: Patient delay
It was the size of the thing growing. That's how I really realised there's something wrong. It sounds trivial but my trousers wouldn't fit. And I hadn't got any clothes that would go round this lump. We didn't know what it was. I thought I was getting fat. So, you do what you can to try and lose weight. (Female, 71)
I went to the GP because I start looking a lump in my chest. I was working as a beautician at the beginning so I thought 'cause my work, this happens. It's a muscle that is developing there. So I thought, okay. So I was dealing six months with the same thought that it is the muscle. (Female, 32)
So when I eventually went to my GP 'cause I'd just watched this thing grow for years. (Female, 28)
So, originally I just had a small lump in my arm that I did nothing about for a couple of months and then my wife told me to go to the doctor's. (Male, 40)
Category 2: Doctors' or health care system delay - symptoms not taken seriously
So it took a long, long time to diagnose…. The diagnosis took a year. And I was shipped from hospital to hospital to hospital. (Female, 25)
The obvious thing that was distressing was the diagnosis took forever at a regional hospital. And they hadn't a clue what they were looking at really. The anxiety that went along with that was quite difficult to bear. (Female, 71)
But going back to one of the points that you guys made as well, about the poo-pooing what it was, I actually felt the first lump when I was 14, in the back of my leg. And the doctor went, "Oh, it's a cyst, don't worry about it." A year later it'd got to the size of an egg and it was only the fact that my mum took me to the A&E that they dealt with it. (Male, 45)
And I went to the doctors and they said, oh, it's growing pains. I was 18 years of age. They were like, well, maybe it's just your natural growth. It's just like, you get made to think you're making it up. And then they sent me for X-rays, but didn't show anything. And it was when I was about 20, just before my 21st birthday, my dad took a picture of me in a tight top. He said, no, there is something there. Take that picture to the GP. I showed the GP and she went, oh, I'll refer you if you want to. We have to give you an appointment within 16 weeks. In 16 weeks you could be dead. So I actually had to pay to see the same consultant private. But it is the best 150 pounds I have ever spent. (Female, 32)
On MRI they didn't see that it was Fibromatosis, they said it was muscle wastage. They said "That's fine, don't worry about it, just massage it." So, I was massaging it for about a year as much as I could and then it [pain] became totally unbearable and then I had the referral to The Royal Marsden Hospital and received the diagnosis of Fibromatosis. (Male, 40)
Subtheme 2: Wrong diagnosis
Category 1: Negative experience
So just went to the GP and they sent me for some scans and, in fact, phoned me up and told me it was cancer, Sarcoma, so actually said I had cancer. So originally I was under the impression that it was a more serious condition than Fibromatosis in the comparable sense. So obviously that first diagnosis of having cancer was quite a shock to obviously tell your family and everything and go through all of that. (Male, 40)
And she [consultant] said we think you've got a malignant sarcoma. And the treatment we're going to have to give you, your baby probably won't survive. And then she said the survival rate for you afterwards isn't necessarily that great. But then I got referred The Royal Marsden and they took biopsies. I saw Dr. Hayes and he said... We were talking through stuff and then he said... I mentioned that I was pregnant and he went, oh, you're pregnant. He said, well, it's almost definitely fibromatosis then. And that completely changed everything. It was all quite a bit of a whirlwind experience. (Female, 28)
Category 2: Positive experience – meaning making
Because, obviously, when I was first diagnosed, I thought it was sarcoma. So I thought I was going to die. And that really made me reassess my life. Realised I didn't want to die whilst doing my current job. So I, basically, applied for my dream job. And I'm moving on towards the career that I want. So it gave me quite a healthy kick really. So that has been the positive element. (Female, 38)

Appendix 2: Quotes Theme 2 Treatment pathway

Subtheme 1: Treatment decision making
The choice of surgery or radiotherapy, obviously they talk about the ladder of starting with Tamoxifen and various types of chemo, radiotherapy, surgery. Surgery is always seen as the last result, or the end game kind of thing. But obviously the risks of radiotherapy for a younger person and obviously the amount of times you can probably have it in your life, versus potentially the debilitating outcome of surgery, was a very difficult decision to make…. So, we were in a bit of a, "What do we do?" It was very much posed as, "You decide," and I was like, "I don't know what I'm supposed to do." So we were in this position of looking for some guidance of what we should do at that stage. (Male, 40)
Because when I started on the treatment ladder I was told, "We'll start at the bottom rung, we'll move up through the treatment." With hindsight, and I remember thinking it at the time as well, when I was first diagnosed the lump was very, very small. By the time last year, when I had the operation, it was a very big tumour. There is obviously a huge risk of operating and it returning and stuff, but to operate on something that was that big at the time….. it would have been a much smaller one than ultimately spending three or so years of my life having pretty horrible treatment and still ending up having an operation which was obviously a much bigger operation. Just my story looking back, I potentially might have made different decisions at different times and being able to speak to people around potentially what would happen if I was going to do this drug for so long or whatever, would have been really valuable. (Male, 36)
But then it was going through that process of how are we going to treat it. I was saying, just let me get through my 50th birthday. Let me have that at least. And then, having the chemotherapy, which then didn't shrink the tumour and really didn't have that much effect on it. (Female, 59)
They don't want to frighten people. And you think you've got to take it and all the rest of it but then you don't realise that's why you feel like you feel. (Female, 71)
Because I was 23, 24 and just had a baby and everything. I didn't really want to make my own decisions. I didn't feel like I had the right experience behind me or knowledge behind me. And I was quite happy for someone to sit there and say, we think this is the best option for you at the moment. (Female, 28)
There is a lack of consistency in treatment recommendations between different consultants and different teams (pain management team, sarcoma team). I chose to assess the situation myself and since no clear and consistent course of treatment plan was offered that I felt secure and comfortable with, I decided to withdraw from all medication. (Female, 30)
Subtheme 2: Direct treatment side effects
It's when you're in the middle of it [Tamoxifen], two years down the line or whatever it is and you're feeling like death. (Female, 71)
I was being sick daily because of complications with my medication. So I had a big period of sickness, I was nauseous and so on, and I lost a lot of weight last year, obviously not feeling, having much of an appetite. (Female, 38)
The year I had the chemotherapy, that was probably the worst year of my life. I don't think I'd wish that Caelyx chemotherapy on my worst enemy. I was horrendously ill. I didn't quite have appendicitis but I was pretty bad and the side effects were horrendous eczema all over my body and they took me off after ten doses. (Male, 45)
Started on Tamoxifen which is an evil drug. Like these gentlemen here, again, lack of libido, nausea. You don't feel yourself and, yes, it wasn't nice for that period of time…. The side effects of Pazopanib, it made me very sick and fatigued. So I was knackered all the time. Interestingly, it turned my hair white….. From the Caelyx I was very tired, very fatigued, but I think what made it worse was the increase in painkillers I was taking. So, I was upping the amount of Pregabalin, morphine, to a very, very high dosage, and that also really didn't help to my attention levels, concentration, fatigue was awful. So I wasn't sleeping, it was all kind of a Catch 22 circular effect. (Male, 40)
My side-effects were really bad. I had hives all over. It was bleeding. (Female, 28)

Appendix 3: Quotes Theme 3 Living with desmoid-type fibromatosis

Subtheme 1: Physical functioning
Category 1: Pain
And I sat here with my consultant and I just burst... I went, I cannot deal with this pain any more. (Female, 28)
I suffered from a lot, lot, lot of pain. (Female, 25)
Yes. Well, the pain is probably now the worst thing about it. It seems not to respond to anything, the pain. (Male, 40)
So, it felt like a burn. It was like a bad bruise that someone was always pressing on and then as it grew bigger it pressed on the nerves and I'd get the stinging, stabbing pains. But the pain I had, it was really sensitive so even just wearing clothes and stuff it would hurt. (Male, 40)
That stinging pain. I've got to points where I'd quite happily take a serrated kitchen knife and cut my own leg off, it was that bad. (Male, 45)
Category 2: Dependency on pain-killers
I've been taking Tramadol for seven years and my body is totally hooked on them. You are like cold turkey, if it's not in your system it's horrendous, I can only describe it as suddenly someone just chucking the worst type of flu at you. You ache all over, you can't move, you've a raging headache, you just feel like death. I asked the doctor to come off them and he put me on Gabapentin. But I just couldn't even drive on the Gabapentin, you're just so spaced out. (Male, 45)
And you become more and more dependent and then you become more and more tolerant (Male, 36)
Category 3: Functional limitations because of pain
I think the main thing that affects me is probably the pain element and just general discomfort because of where it is. So, really, this sounds really stupid. It's stupid things like I can't take a bath anymore 'cause I can't lean back on the tumour site. I can't sit on the sofa comfortably without propping myself up so I don't put any pressure. I've got one sleeping position I can sleep in. Just ridiculous things like that really. (Female, 38)
Yes, it was, very much. I just want to act and try and be treated as normal. But certainly the fatigue and the way it affects that, so actually, I suppose, even though I was increasing the painkillers the pain was getting worse and worse and worse and that also tired me out, made me really irritable, short temper, very moody because you're just living with pain 24/7. (Male, 40)
The pressure thing and the sensitivity is always there. I have to sleep on this side at night now and I have to move my arm behind, I have to put my shoulder back so my arm is on my chest because otherwise then I feel the pressure. (Male, 40)
Feel in my armpits. So it's that movement. And if I'm lifting heavy things I get the stabbing pains in some of the... Had to stop horse riding 'cause of where they are and just the force of movement and things. Makes it [pain] worse. So, yes, another thing that I love that I can't do now. (Female, 32)
Well, it's everything. I can't sit properly. It never goes away. (Female, 28)
It's just this constant awareness that I must watch walking through the doors or if anybody is on this side that they don't touch me. Sometimes, the pain just comes on its own. (Male, 40)
When it was in the bad way I was constantly shielding myself in case you bump into a door, it's excruciating, escalating, on the tube and everything, constantly in that protective way. Had a young daughter at the time and she come to jump at me and I would just be defensive all the time because the pain would be so bad. So that really, the pain did change the way I behaved in terms of just walking around, commuting or dealing with people. (Male, 36)
Category 4: Mobility
I used to do a lot of indoor rock climbing. And anything, now, that involves any kind of extension of my shoulders is very difficult. So it's cut off some of my past-times and things that I used to do in my spare time. (Female, 38)
I find that sometimes I get stuck in clothes. I've been actually in a changing room before and not been able to get it off. 'Cause you think, I should be able to do that. Well, I can't… Had to stop horse riding 'cause of where they are and just the force of movement and things. Makes it worse. So, yes, another thing that I love that I can't do now. (Female, 32)
I can't carry my shopping bag, in my left arm so I get frustrated. I struggle to lift and can't carry my granddaughter. (Female, 53)
He put his hand out to shake me and I couldn't lift my hand. (Female, 28)
I find it humiliating to ask for help with basic activities as in that age people are expected to be fully independent and able to do everything themselves. (Female, 23)
Subtheme 2: Role limitations
Category 1: Marital functioning
Well, on a scale of 1 to 10 of cocking my life up, probably at about 9.9 realistically. My marriage broke up because of all the medication and the side effects, if you guys know what the side effects of taking the Tamoxifen are and how it makes you feel. That's what drove my marriage down the drain, which was a 20-year marriage. (Male, 40)
It [lack of libido, don't feeling yourself because of Tamoxifen] affected the relationship with my wife and everything. Luckily she was strong enough and we got through it together but it was my no means easy. (Male, 45)
And you feel like a burden your partner. Because it's quite... when you're very down. You're maybe crying a lot or getting very depressed about things. I live with my boyfriend and it's almost relentless for him. So when I was off for periods being sick and he was the first person, the only person I saw every day. And he'd come back from work and I would just be... Just unburden on him. And I think that has an effect on relationships. Because then you feel like, oh, I'm being a burden. I need to stop. I need to cope. Because I can't... (Female, 38)

Stress has taken a toll on relationship. I am more irritable with my fiancée, fatigue is affecting sex life. (Male, 40)
Category 2: Family functioning
'Cause when he was a baby, his cot, my tumour would hit the side of the cot when I'd to try and get him out and that. So it was quite difficult. I could never breastfeed him. I didn't do any of that. (Female, 28)
Had a young daughter at the time and she come to jump at me and I would just be defensive all the time because the pain would be so bad. (Male, 36)
I've found that with my son as well. When he's doing imagination games and stuff, he takes his little teddy bears for blood tests and things like that. And he's like, oh, we're going to hospital. And all of his... And then I feel, oh my God, his life has just been my hospital. I do feel awful sometimes that that's his…(Female, 28)
But, ridiculously, I felt like a burden, 'cause my mum would go on up to hospital with me. She'd spend more time with me. So he'd [brother] be left alone. So, at some points, I did attempt to take my life, 'cause I just felt like a burden. I felt guilty. I thought I'm not letting my brother have a childhood or the same treatment as me. (Female, 25)
We are moving with them [parents] now because of my thing. So they can help me with my son. (Female, 32)
Category 3: Job/finances
My job has been changed for me. I can't classroom teach any more. (Female, 59)
I changed jobs recently to a part time position as I had a restaurant before and it was too much for me so sold it. (Female, 53)
Not able to find a new job because pain in hindering. (Male, 40)
I was enforced sick leave. So they wouldn't let me come back. I was, just on statutory sick pay. Which was less than half of what I was earning. So, yes, it was 17emoralizing. 'Cause all I wanted to do was be back at work. And that's what takes my mind off it. Now. Is being at work and being active. And so to be told, no, you've got to stay at home. And I missed out on becoming deputy manager as well because of it. And they actually said to me, well, if you hadn't been off sick, then you would have been deputy by now. (Female, 32)
Financially horrendous, lost my house, everything, because of just not being able to work. I was running quite a successful IT company and just the pain and living with the medication, and what it does to you across the day, it's just been really difficult. (Male, 45)
But what impacted me the most was... Because the space between having surgery and radiotherapy, I wasn't given a car to bring me up here. So I was having to travel two hours a day, every single day, on the train. To great expense. I think it cost me a thousand pounds to travel up here for six weeks. (Female, 28)
The biggest impact with mine was financial. So you think, every single time I was coming up here.... It's 50 pounds travel. (Female, 25)
My financial situation has changed because I had to swop from full-time to part-time job because I was not able to work as hard as before. (Female, 23)
I would like to change jobs but feel I am more secure in existing employment. (Male, 35)
I work extra hours to cover hospital travel costs. (Male, 40)
<u>Subtheme 3:</u> Social functioning
Category 1: Lack of understanding
I was 11 and 12. For me to not understand. My family to not understand. But then have the pressure of my friends saying, why are you missing school? Why are you not here? And then not giving me that support from that age is difficult. 'Cause you're all bonding. You're getting to secondary school and I was out a lot of that. (Female, 25)
But also going to work. I'm a teacher. And people saying to me... One of the comments was really great. Your chemotherapy can't be very strong 'cause your hair hasn't fallen out. That was fabulous. Another one, while we were having a teacher training day and I had all this toxicity in my mouth. Couldn't eat 'cause it was just so painful. It was full of blisters. And they prepared this starter. Actually I said, I'm not going. Because the food was literally falling out of my mouth. And I was told I wasn't a team player. (Female, 59)
I'm not really receiving any active treatment. We're just waiting to see if it grows. That in terms of other people's perception of it, I think, unless you're sort of laid on a bed getting chemotherapy, or you're off work, or blah, blah, everyone thinks you're fine. And it feels like you're just suffering a bit from hypochondria. Like it's not really a thing. So everybody's concern goes away when it's not malignant. But I think they don't understand that it's a daily thing that you're living with and physically and psychologically it's really tough. And it's quite difficult to convey that to people, I think. (Female, 38)
Socializing has become difficult because of the pain and uncomfortableness. (Female, 28)
Constant fight with people explaining what it is I have. (Female,28)
Category 2: Social isolation
Emotional support and support from friends and colleagues. People stop asking you about it. And sometimes you just want to talk about it. And sometimes you don't. But sometimes you do. I think, for me, last year, I became quite shut in. I missed out on a whole ton of social occasions. I didn't go to my work party. I didn't go to a whole load of family occasions. Because I felt so disturbed by the whole diagnosis. (Female, 38)
I feel quite isolated. It's nice to know other patients here are feeling the same. You feel so alone. (Female, 32)
I can see that in my daily life now. Where my friends are a lot more confident than me. And they can speak better than me. They've got a lot more experience behind them. With social scenarios. (Female, 25)
<u>Subtheme 4:</u> Emotional functioning

Category 1: Living with a chronic disease
But what we're all living with has been life-changing and is lifelong. And has knock-on complications for you. (Female, 25)
Mine has become more about the fact that it's just been my life and no matter what, the professor told me, "No matter what we do, you'll be living with this for the rest of your life. It's that simple." He said, "It's come back again. Even if we take it away, it will probably come back again." (Male, 45)
I guess my psychological demeanour just generally, knowing that I've now this probably chronic condition. (Male, 34)
But when they told me that it's coming back, I say, oh, wow, this is a strong. This is not that easy as I thought. And then another one [tumour] in less than a month. I say, oh my God, what is going to happen? What is the next thing? This is never...Never ending. (Female, 32)
Category 2: Uncertainty around tumor behavior and fear of recurrence
Sometimes it feels bigger, sometimes it feels smaller. Yesterday, it actually felt smaller. This morning, I looked at it again and it went all the way around the arm again. I don't understand how it can change in size that quickly or is it my imagination or am I going mad? (Male, 40)
But then without having any sort of pressure on it at all, you can just go through big twinges of just that kind of burn sensation and no stabbing pains. It just seems really weird. Then that throws you psychologically because you're like, "Well, is it growing?" Or if you haven't got that, "Is it not growing? Is it reducing in size?"(Male, 40)
Category 3: Mood
And I was so depressed. I was just going down and down and down and down. But you don't really realise 'cause it goes very slowly. And then that I really struggled with. Because I knew it was my drugs that made me like it. But I couldn't... I need a solution that doesn't involve drugs. 'Cause this is my life forever. And I can't be in that state forever. (Female, 28)
When I was on Tramadol I got very constipated and when I came off it very very depressed and emotional and I was not prepared for that. (Female, 53)
The mood swings, I have the mood swings. (Male, 40)
Even though I was increasing the painkillers the pain was getting worse and worse and worse and that also tired me out, made me really irritable, short temper, very moody. (Male, 40)
I get very tired on the medication I'm on (Naproxen and Tamoxifen) which makes me irritable and stressed.(Female, 53)
Category 4: Body image
It's also the feeling very conscious that I've got a big lump on my shoulder which, as a woman. And I don't feel comfortable having it exposed. So I wouldn't wear a vest top in summer and things like that. And I think that probably has a bit of a knock-on effect with my general body image and my sex life with my partner and things like that. So, I think, last year I was hit really hard by it. I found it really difficult. (Female, 38)
It changes the way you look as a woman. Because you have to cover it all the time, cover it, cover it…(Female, 32)
But I understand what you're saying about when you're dressing and how you look. Because I'd gone from being a person who was always little tops, whatever. But now I've got this massive... Well, it looks like a route map of Great Britain, really. It runs all the way down here. All the way across here. And it's upsetting. It just changes your life, yes. Dramatically. (Female, 59)

Appendix 4: Quotes Theme 4 Supportive care

Subtheme 1: Finding your way in the health care system with a rare disease
About being diagnosed with the thing and understanding what it means. What it is. It's so terribly difficult to find out. It took me weeks on Google. But not even the word 'desmoid'... What does that... Nobody knows what it means. I know a lot of doctors. They say "What?" And then you get onto the whatever it is of rare diseases and so I think that was a huge problem for me. What is this thing? (Female, 71)
I think it is because it's such an unknown thing. I've been into hospital while I've had this and had to explain what it is I've got. Because automatically look at you. Oh my God, you've got a tumour. And when you tell them they go, oh. I don't want to have to explain it every time I come in. And in the end, I actually had a print-out, a sheet, with me. I've had this. And I just used to say to them. (Female, 28)
And I think probably in all of our cases, we're probably the only people that our GPs have seen with this condition. And it's so rare. He had to Google once I got my diagnosis. (Female, 28)
I think one of the worst situations was dealing with …..[one of the cancer patient support organisations], of being a patient who's dealing with all the side effects and treatment of cancer and they were like, "You're not a cancer patient so we're not going to help you." It put my wife, you know, it made her cry because they just wouldn't help. We were like, "Yes, they must deal with obviously a lot worse situations and there are patients out there in a lot worse positions," but it was very much, for us, we were all dealing with exactly the same symptoms, pain, the output of what the treatment gives you and everything. But because it's not cancer they were like, "We're not going to help you." (Male, 40)
I think, personally, find it psychologically quite difficult coming to a cancer hospital and being surrounded by people who probably have terminal conditions. And coming in with my little desmoid. And you feel a bit like a fraud. So I do find in my consultations, I'm really aware that they've probably seen somebody who's dying before and after me. And you don't want to waste time. And you tend to downplay things. So, personally, from my point of view, being surrounded by other desmoid sufferers, would help a lot. Like you say, in the waiting room situation, and if you saw the same faces, that would also be quite helpful from my perspective. (Female, 25)
Subtheme 2: Information provision
And I was seen by an orthopaedic surgeon who after my... I had an appointment after my diagnosis. And during that appointment he asked me what I wanted to do about... And I had literally no information about the condition other than what I could glean off the internet, which was not very much at all. And, at the time, I just wanted it out. So I said, well, I want it removed. So he said, okay, fine. Book you in for surgery. So I left and he said, oh, it'll be between six and 12 weeks for the surgery. And when I left the hospital I thought, this feels really strange, 'cause he hasn't told me what the surgery would involve. He hasn't told me about recurrence which I now know is a problem. So at that time I didn't know. He didn't explain that, basically, it's almost these tumours have like tentacles. So you have to remove a huge margin. In some cases on the shoulder, they've had to remove most of the shoulder muscle. That I could be quite disabled afterwards. It could recur. All this kind of thing. He never went through anything with it. (Female, 25)
'Cause I was laying on the radiotherapy bed and they said to me, right, you are only 26, at this stage. We think that it's going to make you infertile because it's going to obliterate both of your ovaries. They said, do you still want to go ahead? Because, obviously, it's that and it's not cancerous. But if you don't it's not to going to kill the fibral tissue. So it could reoccur. And at that time I wasn't... didn't want children. So I said, right, go ahead. I said, if I have to, I'll adopt. If I want children. So I done that. My last radiotherapy session was April Fool's Day, year 2010. Three months later, I fell pregnant with my son. (Female, 28)
Just a single information sheet about fibromatosis. There isn't one. (Female, 59)
A leaflet with information would be really helpful as I had never heard of it before diagnosis. I was told by the doctor to Google it! I would be better to have a hand out with description of what it is and tips on how to help it e.g. keep out of the sun, use ice and any other tips that would help. Maybe a picture of what it looks like. (Female, 53)
Subtheme 3: Continuity of care
The only difficulty I found here, was that every time we came in, I always saw a different person each time. I know it's all part of the multi-discipline team and everything like that, but it'll be with Dr A and then Dr B and then someone else. It was never one point of contact, but it always felt that you always had to explain again your circumstances because they'd have your notes but they might not have been dealing with it. That to me was probably the most frustrating thing because, even though obviously it was apparent that your case was discussed and people did know your situation, probably not fully but it was always like, "Who are we going to see today?", "Do they really know my full story and background?"And with that it became more of a risk of having different recommendations given to you. It would be, "Oh, in my opinion you should do this," or, "We're thinking of doing chemotherapy," or, "Actually, we're thinking of keeping you on the Tamoxifen for another three months," whatever. (Male, 40)
When you get into this complicated situation, you don't know what's causing what and why you're feeling like you are. And a session with a professional counsellor who's knowledgeable about the whole drug scene would be really good time spent. (Female, 71)
Subtheme 4: Peer support
I am seriously overwhelmed to see all of you with the same disease as me. I live nearly 11 years with this disease and thought I was the only one. (Female, 32)
For me personally, me being out and to know that there are other people that are living with this condition, and being

able to potentially have contact with these people, would be really useful and comforting. And also I think we probably all feel like we're out there on our own a little bit and it's nice to know that you've got possibly some other people that you know you can count on your support when maybe times are a little bit tough. (Male, 45)
But again, mirroring what the guys were saying here, being able to speak with people who are dealing with the same disease, it's a chronic illness, and so little is known about it, but it's really valuable to hear their stories. You feel unique in some respects and it's good to hear what other people are going through and you're not really the only one. And being able to potentially help people who are at earlier stages in the treatment to hearing from people who are further down the line, is mentally valuable. (Male, 40)
An online support or chat group or something would really validate your experience. It validates what you've been through. (Female, 25)

Attachment 5. Semi-structured interview template

1.Introductory remarks to explain the nature and purpose of the study.

Good afternoon/evening. Thank you for taking the time to join our discussion on the supportive care needs of patients with desmoid fibromatosis. My name is ____MODERATOR____, and I'm __ASST MODERATOR____.

In particular, we hope to learn more about the physical, psychosocial and practical challenges you face as a desmoid fibromatosis patient and on how your experience has been of the healthcare system: What kinds of things have been helpful to you and what kinds of things you might have wanted or needed but did not receive.

Before we begin, let me suggest some things to make our discussion more productive. Because we'll be recording for an accurate record, it is important that you speak up and that you only speak one at a time. We don't want to miss any of your comments. We'll only use first names here. No reports will link what you say to your name. In this way, we will maintain your confidentiality. In addition, we ask that you also respect the confidentiality of everyone here. Please don't repeat who said what when you leave this room. During the two hours we'll be here, I will ask you questions, and I will listen to what you have to say. I will not participate in the discussion. So please, feel free to respond to each other and to speak directly to others in the group. We want to hear from all of you. We're interested in both majority and minority viewpoints, common and uncommon experiences. So I may sometimes act as a traffic cop by encouraging someone who has been quiet to talk, or by asking someone to hold off for a few minutes.

Does anyone have a problem with audio recording of this focus group? Okay, then we will turn on the recorder and start now.

2. Introduction participants

First of all, can you briefly introduce yourself (first name, age, tumour localisation, treatment received)

3. Main open-ended questions <u>CHALLENGES</u>

-When we think about your health, what would you say the term quality of life means to you?
* What factors would you say contribute to *your* quality of life?
* Which factor would you say is / are the most important to you?

-With that in mind, in what ways has your desmoid fibromatosis diagnosis affected your quality of life?
-With that in mind, in what ways has your treatment for desmoid fibromatosis diagnosis affected your quality of life?
-With that in mind, in what ways has the watch and wait policy affected your quality of life?

PROBE AS NECESSARY:

e.g. in these areas, how has it affected you …
-*Physically:* e.g. symptoms ….. how?
-*Functionally*: e.g. ability to get about / wash / dress …. How?
-*Psychologically:* e.g. feeling worried/ stressed / tense ….. How?
-*Socially:* e.g. impact on relationships, social life …….How
- Other: (Financial/ Spiritual)

-Could you tell me more about that and how you deal(t) with challenges/problems?

-What would you say has been the most important / biggest factor(s) influencing your quality of life since your diagnosis?

-What is the meaning of symptoms for you in relation to disease activity (e.g. fear of progression) and response to treatment?

4. Main open-ended questions <u>(UNMET) CARE NEEDS</u>

-I would like to ask you to talk about the various needs you have as a desmoid fibromatosis patient. -What has your experience been with receiving healthcare services/supportive care for your desmoid fibromatosis?

PROBE AS NECESSARY:

What was good about that care you received till now? Why?
What was bad about the care you received till now? Why?
What did you miss? Why?
What is your experience with referral to supportive care and use of supportive care options?

-I'll now ask you to talk about your needs for a number of different areas. I would be interested to learn for each area whether you have needs in this area and whether or not your needs have been addressed (what is/was helpful and what is/was missing).

Informational needs
Psychological/Emotional needs positive and negative
Physical needs
Risk behaviour needs
Practical needs
Social needs
Spiritual needs
Health care needs

5. Main open-ended questions <u>EXPECTATIONS AND PREFERECES</u>

-What are your expectations of care for patients with desmoid fibromatosis?

-What are your preferences for care for patients with desmoid fibromatosis?

PROBE AS NECESSARY:

If you could design the perfect expert center for desmoid fibromatosis patients what would it look like for patients that are undergoing treatment? What would it look like patients in watch and wait phase? Please be as creative as possible as we are interested in your ideas.

What kind of programs would be helpful to you in dealing with the challenges of desmoid fibromatosis?

What formats are you most interested in? Would you prefer groups, individual, face-to-face, online, or a combination?

6. Summarise the issues raised during the interview and identify which issues are the most important.

-We have discussed a number of things. These are…MODERATOR SUMMARISES POINTS…..

-Out of everything we have talked about today, what are your top three needs and how could they be better met?

- Is there anything else about needs related to desmoid fibromatosis that you can think of that we have not already discussed?

Thank you again for taking the time to participate in this discussion.