

Understanding desmoid-type fibromatosis



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The bone & soft tissue cancer charity

About this booklet

This booklet is for anyone who has been diagnosed with desmoid-type fibromatosis (DF). It explains what DF is, how it is diagnosed and the treatment options available to you. It is your personal guide with space available to keep all your diagnosis information and details of your next hospital appointments. You can also include contact details for your doctor, clinical nurse specialist and other healthcare professionals caring for you.

You may find it useful to share the information in this booklet with your partner or family members to help them understand about DF. If you have any questions about anything you read in this booklet please contact Sarcoma UK's Support Line. We offer information, support and a signposting service to other organisations that can help.

Treatment may vary depending on which nation of the UK you live in. Please ask your doctor or clinical nurse specialist about the treatment options available in your area.

This booklet was produced by **Sarcoma UK**, the only charity in the UK focusing on all types of sarcoma cancer.

Our mission is to amplify awareness, inspire involvement, and fund ground-breaking research to transform the lives of everyone affected by sarcoma.



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Contents

What is desmoid-type fibromatosis (DF)?	2
How is DF diagnosed?	2
Types of diagnostic scans and tests.....	3
Who should treat me?	4
What treatment is available?	4
What happens after my treatment?	10
What if my DF comes back?	10
What support is available?.....	10

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Questions you might consider asking your doctor or sarcoma clinical nurse specialist?

What is desmoid-type fibromatosis (DF)?

Desmoid-type fibromatosis (DF) is sometimes called Desmoid Tumour or aggressive fibromatosis. It is a rare type of benign (non-cancerous) tumour. People with DF may have more than one tumour located in the same area of the body. DF develops from fibroblasts. These are a type of cell that provides support to the body's tissues. DF can occur anywhere in the body but it is mostly found in the arms, legs and abdomen (tummy). DF can affect anyone but it mostly affects adults, especially women, between 30 and 40 years old.

“DF can affect anyone but it mostly affects young adults, especially women.”

It is difficult to predict how DF will develop. Sometimes they can be slow growing and they have also been known to get smaller without any treatment.

There is a small number of people (5-10%) with DF who have conditions called familial adenomatous polyposis (FAP) and Gardner Syndrome. This is a rare condition that can run in families. People affected by FAP may be diagnosed with DF following routine tests and scans to investigate the progression of their condition, such as a colonoscopy. Usually their DF will be in the abdomen and treatment will be different to those who do not have FAP.

How is DF diagnosed?

People with DF that is not related to having FAP may discover a lump and visit their GP who will refer them to a specialist doctor.

A specialist will do:

- A clinical examination – looking at and feeling the lump
- A scan – taking pictures of the inside of the body using ultrasound, CT or MRI
- A core needle biopsy – taking or testing a tissue sample. Core needle biopsies use a core (meaning hollow) needle to remove the tissue. This is usually done under local anaesthetic.

Types of diagnostic scans and tests

MRI

Magnetic Resonance Imaging (MRI) uses magnets to create an image of the tissues inside the body. This is the most common scan used to diagnose DF.

X-ray

Uses x-radiation to take images of dense tissues inside the body such as bones or tumours.

Ultrasound

A scan that uses soundwaves to create images from within the body.

CT

The Computer Tomography (CT) scan takes a number of x-rays to make 3D images of an affected area.

Colonoscopy

This test uses a thin, flexible tube called a colonoscope to look inside the colon.

Histopathology

Examination of a tissue sample by a pathologist under a microscope to identify disease. The tissue sample can

A biopsy of DF should be done by a specialist sarcoma team who has expertise in diagnosing and treating DF. A diagnosis of DF should be confirmed by an specialist soft tissue pathologist.



“A colonoscopy uses a thin, flexible tube called a colonoscope to look inside the colon.”



Has my tissue sample been sent for mutational analysis?

“The MDTs with the experience to treat DF are usually only found in sarcoma centres.”

be taken during a biopsy or from a tumour removed during surgery. A diagnosis of DF should be confirmed by a specialist soft tissue pathologist.

85 – 90% of DF tumours have mutations that are only found in DF. An investigation called mutational analysis looks for these mutations in DF tumours. This can help to accurately identify a DF tumour.

Who should treat me?

DF patients are treated under an oncology team. Oncology is the branch of medicine that treats cancer. DF is not a cancer but oncologists (doctors who specialise in the treatment of cancer) have the experience and skills to look after DF patients. Your treatment plan will need to be managed by a team of experts from a wide range of health care professions called a multidisciplinary team (MDT). The MDTs with the experience to treat DF are usually only found in sarcoma centres. Your MDT will include your clinical nurse specialist, surgeon and oncologists with experience of treating DF. See page 12 for more information on your MDT.

What treatment is available?

Treatment for DF depends on the area of the body that is affected, the size of the tumour and how fast it is growing. It also depends on how close your tumour is to important structures in your body such as a major blood vessel or an organ. Your doctor should explain your treatment options to you and why your MDT has made their decisions. DF tumours can be unpredictable, they can stabilise (stop growing) and even regress (shrink). No one knows why this happens. If a DF tumour stabilises or shrinks then you may not need treatment.

In the past, treatment for DF has relied on surgery. Now you are more likely be under a Watchful Waiting programme. See the box (opposite) for more details on how experts came to this decision.

Watchful Waiting

Watchful Waiting is usually the first approach to treat people who are newly diagnosed with DF. This is sometimes known as a 'watch and wait' policy, 'active monitoring' or 'active observation'. Watchful Waiting ensures effective treatment can be held in reserve for when you need it. You will be constantly monitored to ensure treatment is given at the earliest appropriate time.

People diagnosed with DF are usually on a watch and wait programme for 1 -2 years. You may be on a watch and wait programme for a shorter or longer time depending on your individual circumstances. If your condition stabilises or regresses watch and wait will continue. If your condition progresses then other treatment options will be considered for you.

If you are on a watch and wait programme you may be worried that your DF is not being treated immediately. However, there are advantages to watch and wait programme:

- Treatment options for DF come with side effects. Although these treatments can be very helpful, it is important to only use them if you are having symptoms that affect your quality of life or your tumour has grown greatly.

In 2017 a consensus paper was written with involvement of doctors and nurses from across Europe who treat DF. This paper is an updated version of the 2015 consensus paper. Previously different countries had treated DF patients in different ways and there is currently little research into what are the best treatment options. The group found that it is best to have understanding of the growth of the tumour before deciding if any treatment is needed. This can be achieved by reviewing a patient with DF under a Watchful Waiting programme.

“People with DF are usually on a watch and wait programme for 1-2 years.”

If you are having symptoms that are affecting your quality of life you should be offered symptom management. Ask your GP for help. They may ask your local pain management team or symptom control team for further advice. You can also speak to your clinical nurse specialist if you are worried about any symptoms you are experiencing.

- Some treatments cannot be given more than once, for example, radiotherapy. Radiotherapy can be helpful to improve your symptoms but it may not shrink the tumour.
- It is possible that having repeated surgery may cause the tumour to be more aggressive in its recurrence and growth.

Monitoring

If you are on a Watchful Waiting Programme you will be monitored regularly by your specialist team. Usually every three months. At each appointment you will be checked for signs that you may need further treatment. This will be done by:

- Giving you a chance to discuss your symptoms
- A clinical examination - looking at and feeling the tumour
- You may also have a scan, such as an ultrasound or MRI

The reasons you may need further treatment could include:

- You are experiencing symptoms that are affecting your quality of life. For example, increasing pain.
- Your tumour is growing quickly
- Your tumour has grown and is getting close to important structures in the body like nerves, a major blood vessel or an organ.

DF and pregnancy

During pregnancy the rate of growth of the DF may change, sometimes the tumour can grow and then may settle down

after pregnancy. Any changes with your tumour can be safely managed and less than half of pregnant women with DF need treatment to manage these changes. DF does not increase the risk of anything going wrong during pregnancy but if you are concerned speak to your doctor or clinical nurse specialist who should be able to reassure you. If you are considering getting pregnant please speak to your specialist doctor or clinical nurse specialist first.

Surgery

It is sometimes possible for your surgeon to remove the tumour. Many patients are keen to have their tumour removed. In the past, surgery used to include taking the tumour out along with a wide area of normal tissue too. This is known as taking a wide margin. Now it is thought that removing a large amount of tissue around the tumour does not always make a difference to whether it will regrow, so surgeons now aim to just remove the tumour.

Sometimes after surgery for DF, the part of the body where the tumour was removed does not work properly or you are left with cosmetic changes to your appearance. Surgery also does not guarantee the tumour will not return. This is why, for most DF patients, surgery will only be offered after a period of Watchful Waiting and after their case has been discussed by an MDT.

Chemotherapy

Chemotherapy is a treatment that uses drugs to destroy tumour cells. Chemotherapy can help switch off the tumour and make it dormant. This may help with symptoms such

The treatment you receive will be based on your individual circumstances. Your MDT will discuss your case and decide on what treatment is best for you. You may receive any of the following treatments depending on your specific circumstances.

“Chemotherapy can help switch off the tumour and make it dormant.”

as pain. The tumour may get smaller with chemotherapy treatment or stop growing. Chemotherapy can cause side effects so the decision to use it needs to be balanced with how your symptoms are affecting you and how the tumour is growing. The decision to use chemotherapy is on a case by case basis. It is more likely to be used if your DF is progressing quickly and if your symptoms cannot be managed another way.



Ask your clinical nurse specialist for more information on TKIs

Tyrosine Kinase Inhibitors

Tyrosine Kinase Inhibitors (TKIs) are a newer type of treatment called a targeted therapy. They work by blocking growth signals inside the tumour cells. The main drug used to treat DF this way is called Imatinib. Special funding is needed to make imatinib available for DF patients. You can ask your clinical nurse specialist for more information on this type of treatment.

Anti-inflammatory drugs

Nonsteroidal anti-inflammatory drugs (NSAIDs) can be used to treat DF. They have an impact on the nature of the tumour and can reduce any pain or swelling you may be experiencing.



You may experience side effects from hormone therapy. Ask your clinical nurse specialist about side effects if you are offered this treatment.

Hormone treatment

Hormones are substances produced naturally in the body that control the activity of cells and organs. Hormonal therapies use drugs to interfere with the way hormones are made or how they work in the body. Tamoxifen is an antioestrogen drug used to treat breast cancer. It has been found to work in treated DF either using this drug alone or alongside nonsteroidal anti-inflammatory drugs. It is commonly used as the first medical treatment

for DF, particularly if the DF is in the abdominal wall.

Radiotherapy

Radiotherapy uses high-energy radiation beams to destroy tumour cells. It can be used after surgery or as a treatment on its own. It is used to improve symptoms and to try and get the tumour to switch itself off and get smaller. DF patients are usually fit and healthy and radiotherapy can have significant long term side effects, so the decision to use radiotherapy needs to be balanced with how significant the symptoms are. It is rarely used to treat children and younger adults.

Isolated Limb Perfusion

Isolated Limb Perfusion (ILP) is a treatment where chemotherapy drugs are given directly into an arm or leg to treat a tumour. It is used as an alternative to surgery to treat DF in the arm or leg where surgery will cause damage to the arm or leg or make it not work properly. Not all specialist centres offer this treatment. If ILP is considered appropriate for your case, you might need to be referred to a specialist centre with expertise in this procedure.

Cryoablation

Cryoablation is increasingly being used to treat DF. It uses extreme cold to freeze the tumour. A needle called a cryoprobe is put through your skin directly into the tumour. A gas is then pumped through the cryoprobe to freeze the tumour. It can be used to treat tumours that are small and easy to get to through the skin. This technique is not available in every specialist centre. If you are eligible for this treatment your clinician may refer you to a specialist centre with expertise in delivering cryoablation treatment.

“Cryoablation is increasingly being used to treat DF.”

What happens after my treatment?

After any treatment you receive, you will have regular follow up appointments. These appointments are to check your health and to check the site of your DF. They are also a good opportunity to talk to your doctor or nurse about any concerns you may have. At a follow up appointment you will usually have:

- A chance to discuss symptoms
- A physical examination to look for any signs of your DF returning
- You may have a scan such an ultrasound or MRI

What if my DF comes back?

Sometimes DF can come back after it is first treated. This is called a recurrence. Sometimes your DF can be stable for a long time and then start growing again. If you do have a recurrence or your DF starts growing again your doctors will discuss your care in an MDT meeting and consider the best treatment options for you.

What support is available?

Sarcoma UK Support Line

Our Support Line is here for everyone affected by sarcoma – we support DF patients too as you will be treated under a sarcoma MDT.

- Our Support Line is confidential



supportline@sarcoma.org.uk

- We believe no question is a silly question
- We lend a listening ear
- We can point you in the right direction

Symptom management

This is care that aims to help you live as well as possible with the effects of DF. It could include social, psychological or spiritual support. It may also include dietary services, physiotherapy and occupational therapy. Your clinical nurse specialist may be able to suggest useful services to you and your GP can help you access services in your area.

Peer support

There is a UK support page on Facebook for anyone affected by DF called Desmoid United UK.



This is a closed group where DF patients can share their stories and offer and receive support. To join, search for “Desmoid United UK” in the search bar.

The Desmoid Tumor Research Foundation is an American organisation that offers information and support to DF patients. They also fund research into improved treatments and aim to find a cure.

Other sources of support

- British Pain Society
Resources for people living with pain
britishpainsociety.org
- NHS Pain Support
Tips and advice on how to manage pain
nhs.uk/Livewell/Pain



My diagnosis details

Date of diagnosis

Details of diagnosis

Treatment plan

My multi-disciplinary team (MDT)

You can use this space to keep the contact details of your team members who will be involved in your treatment. You may not see all the health professionals listed below.

Key Worker/Clinical Nurse Specialist (CNS)

Offers support and advice, and acts as an important point of contact for patients when they have a concern.

Name

Direct telephone

Email

Notes

Oncologist

A doctor who specialises in cancer treatments other than surgery, for example, chemotherapy or radiotherapy. An oncologist will usually take the lead in the treatment of people with DF.

Name

Direct telephone

Email

Notes

Surgeon

Treats DF by removing the tumour.

Name

Direct telephone

Email

Notes

Radiologist

A doctor who specialises in diagnosing medical conditions through images, for example, x-rays.

Notes

Radiographer

A clinical technician who takes images, such as x-rays, and may also give radiotherapy as a treatment.

Notes

Pathologist

A doctor who identifies diseases by studying tissue samples.

Notes

Other professionals

You can record the contact details of other professionals you come into contact with in the space below. These may include a dietician, psychologist or social worker.

Name

Contact details and notes

<i>Name</i>	<i>Contact details and notes</i>

Appointment diary

You can keep track of your hospital appointments by recording them in the space below.

<i>Date</i>	<i>Hospital</i>	<i>Appointment/Treatment</i>

This booklet has been produced by the Support & Information Team at Sarcoma UK. It has been reviewed by Sarcoma UK's Information Panel which includes healthcare professionals and people affected by desmoid-type fibromatosis.

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Sarcoma UK makes every reasonable effort to ensure that the information we provide is up-to-date, accurate and unbiased. We hope this booklet adds to the medical advice you have received and helps you make informed decisions about your care and treatment. Please speak to a member of your care team if you are worried about any medical issues.

Sarcoma UK does not necessarily endorse the services provided by the organisations listed in our publications.



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