sos desmoid

sos desmoid - Support group for patients with aggressive fibromatosis / desmoid tumours

Living with Desmoid Tumour: Knowledge, Pathways, Support

Desmoid Tumour: Understanding & Treatment

What are desmoid tumours?

Desmoid tumours – also known as “aggressive fibromatosis” – are rare tumours of connective tissue with locally aggressive growth. Although they do not form metastases, their growth can displace or damage surrounding tissues. This often leads to pain, restricted movement, or functional impairments.

In rare cases, multiple tumours may occur at the same time (multifocal). There is also an increased risk of recurrence. Care should always be provided in an experienced sarcoma centre.

Who is affected?

In Germany, approximately 200–400 people are diagnosed each year. Frequently affected are:

- Young adults between 20 and 40 years
- Women more often than men, sometimes in connection with pregnancy
- People with familial adenomatous polyposis (FAP)

Why do desmoid tumours develop?

Desmoid tumours arise due to genetic changes (mutations) that permanently activate cell growth. Most commonly, mutations occur in the CTNNB1 gene, and in FAP in the APC gene. Injuries or surgeries are suspected as possible triggers, but not as the sole cause.

Typical symptoms

Symptoms depend on the tumour location:

Arms or legs: palpable lumps, pain, restricted movement

Abdomen: abdominal pain, digestive problems

Head/neck: palpable lumps, difficulty swallowing and/or breathing, pain

How is a desmoid tumour diagnosed?

- Imaging: MRI with contrast is generally recommended. For abdominal tumours or if MRI is not possible, CT may be used as an alternative.
- A tissue sample (biopsy) is required for a definitive diagnosis and should ideally be performed at an experienced centre..
- Additional genetic tests can support the diagnosis.

IMPORTANT

The diagnosis should always be made or confirmed in a sarcoma centre or by experienced desmoid experts.

Desmoid Tumour: Understanding & Treatment

Does it need immediate treatment?

In the past, the rule was: “The tumour must be removed.”

Today we know that surgery is not always the first choice, provided the tumour is not threatening vital structures.

Experts therefore often recommend initial careful monitoring – known as active surveillance.

What treatment options are available?

Active Surveillance
Regular MRI scans as long as the tumour remains stable and symptoms do not worsen.

Drug therapies
- Targeted therapies (e.g., imatinib, sorafenib, pazopanib, or nirogacestat)
- Low-dose chemotherapy in specific situations (combination of methotrexate + vinorelbine or vinblastine, liposomal doxorubicin)

Surgery
Surgery is only useful in selected cases, for example in severe symptoms or impending loss of function. Recurrences are possible, even after complete tumour removal.

Local treatments
Targeted destruction of tumour tissue, e.g.:
- Cold (cryoablation))
- Ultrasound waves (HIFU)

Radiation therapy
Radiotherapy is used only in very rare exceptional cases, for example when other treatments are not an option.

What is nirogacestat (Ogsiveo®)?

Since 2025, a medication specifically developed for desmoid tumours has become available for the first time. Nirogacestat blocks important growth signals in tumour cells. In many patients, tumour growth can be slowed or stopped, and in some cases the tumour shrinks. Many patients also report reduced pain and improved quality of life.

What is happening in research?

Further new drugs are currently being tested in clinical trials to improve treatment options.

IMPORTANT

Every treatment decision should be made individually within an interdisciplinary team at an experienced centre. A second opinion may be helpful.

Medical content advice:
Prof. Dr. Bernd Kasper, Sarcoma Centre Mannheim
Many thanks for the professional support..

Desmoid Tumour: Living & Coping

A desmoid tumour changes life

The course of the disease is often unpredictable. This uncertainty can be very distressing for many patients.

Physical burden

- **Pain** is common and can limit daily life if the tumour presses on nerves, muscles, or organs..
- **Restricted mobility** occurs when muscles or joints are affected. Everyday activities or sports may become difficult.
- **Tumours in the abdomen** can cause digestive problems and, in rare cases, serious complications.

Emotional burden

The diagnosis often triggers fear and uncertainty. Many patients feel alone because the disease is rare and often invisible to others.

Support in everyday life

- **Symptom diary:** Record pain and limitations to better assess changes together with your medical team.
- **Physiotherapy:** Maintains mobility and can relieve symptoms..
- **Pain management:** Consultation and treatment in specialized clinics can help control pain effectively.
- **Psychological support:** Discussions with psycho-oncologists can reduce anxiety, worries, and uncertainty.

IMPORTANT

Take an active role in your treatment – in close cooperation with your treating specialists. Being well informed and making decisions together can provide strength and confidence.

Desmoid Tumour: Help & Support

The diagnosis of “desmoid tumour” is life-changing for many patients and affects more than just the body. The disease can impact professional, family, and social life. It is therefore all the more important to find and accept appropriate support early.

Medical support

Seek care at a sarcoma centre. Different specialists work together there in an interdisciplinary way. These centres have special expertise and develop an individualized treatment strategy together with you.

Psychological support

Uncertainty and anxiety are normal. Psycho-oncological counselling can help you find ways to cope with emotional stress and regain stability.

Patient support group & exchange

Connecting with other patients is particularly valuable for many – realizing “I am not alone.” Here you will find understanding for issues such as pain, fear of follow-up examinations, or dealing with authorities.

Social & legal support

- Hospital social services can help with questions about rehabilitation, sick pay, pensions, or disability status.
  However:
- Because desmoid tumours are rare, they are often not correctly classified within the healthcare and social system. Many applications fail not due to lack of need, but because the specific burden of the disease is not understood. Our specially developed “medical-social statement” for patients clearly and comprehensibly describes the impact of the disease. It can help improve your chances of a fair decision – for example when applying for rehabilitation, aids, disability status, or psycho-oncological support.

IMPORTANT

Seeking and accepting help is not a sign of weakness, but of strength. Get in touch with us!

Desmoid Tumour:
Community & Association

sos-desmoid Association – stronger together

We are the organization in Germany for patients and relatives affected by a desmoid tumour diagnosis..

Our mission:
To inform • To support • To connect

We work closely with leading desmoid experts, the German Sarcoma Foundation, and other patient organizations nationally and internationally.

What we do

- Provide clear and understandable information in print materials, online media, and at events
- Enable exchange – in online groups, meetings, or by phone
- Connect patients with desmoid experts and sarcoma centres
- Advocate for the patient voice within the healthcare system

Why networking matters

Many patients report:
The first contact with other patients is a turning point. Suddenly there are people who understand the same concerns.

What community means

- Understanding and belonging – finally someone who listens and truly understands.
- Realizing that it is possible to find ways to live with the disease
- Practical tips – from managing pain in daily life to dealing with authorities
- Knowledge and expertise – through collaboration with experts and the German Sarcoma Foundation

IMPORTANT

Our association can support you and help guide you on your personal path with a desmoid tumour diagnosis..

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Visit our website!

You will find up-to-date information, practical tips for daily life, and ways to connect with our community.

Further helpful information:
www.sarkome.de
www.sarcoma-patients.org

Support our work! Your donation helps us to:

- Create understandable information materials
- Organize support groups and meetings
- Promote scientific and health policy advocacy

Every contribution counts –
together we can achieve more.

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