Dermatofibrosarcoma Protuberans

What is dermatofibrosarcoma protuberans (DFSP)?

DFSP is an uncommon skin tumour arising in the deeper layer of the skin (the dermis). It grows slowly but has a tendency to recur after excision. Luckily, it rarely spreads to other sites beyond the skin.

What causes DFSP?

The cause is unknown, but an injury to the affected skin may be a predisposing factor. Recent advances show tumour cells carry abnormal chromosomes within the tumour cells—t(17;22)(q22;q13)—resulting in the fusion gene COL1A1-PDGFB. This encodes a protein that causes the tumour to grow.

Who is at risk of DFSP?

DFSP is rare, and affects less than 1 person in every 100,000 inhabitants per year, It usually presents in early or middle adult life between 20 and 59 years of age, but all ages can be affected. The tumour is rare in children. Males are affected slightly more frequently than females. There does not appear to be any racial predilection

What are the signs and symptoms of DFSP?

DFSP usually presents as a painless thickened area of skin (plaque) and/or nodule that feels rubbery or firm to touch and is fixed to the underlying skin. It may be red-brown or skin coloured. It usually grows very slowly over months to years.

Rarely it presents as a soft depressed area of skin making the diagnosis even more difficult. DFSP may range in size from 0.5 to 25 cm in diameter. Fifty to sixty percent of tumours arise on the trunk often in the shoulder and chest area. The remaining 35% of tumours are found on the limbs and 10 to 15% on the head and neck region.

DFSP is often diagnosed when it enters a more rapid growth phase giving rise to larger lesions. Neglected tumours may reach large proportions.

How is DFSP diagnosed?

The absence of symptoms often leads to a delay in diagnosis. Redness and pain only occur in 15% of cases. It is often mistaken for other skin conditions particularly in the early stages. Skin biopsy is needed to confirm the diagnosis.

It has a characteristic appearance under the microscope with densely arranged spindle shaped cells. It may be difficult to assess complete removal due to extensions widely in the skin and deeper structures. It is important to identify fibrosarcomatous DFSP, a more aggressive tumour, that requires more aggressive treatment.

DFSP's chromosomal abnormalities can be detected using reverse transcription polymerase chain reaction (RT-PCR) or fluorescence in situ hybrydization (FISH).

In most cases no other investigations are necessary. However, if there is suspicion of metastasis or there is fibrosarcomatous transfomation, lymph node ultrasound, chest X-ray and pelvic ultrasound scan may be arranged.

What is the treatment for DFSP?

Treatment of DFSP and DFSP with fibrosarcomatous transformation consists of wide excision of the lesion including deep fascia, with 1–3 cm margin of normal skin. This may take more than one surgical procedure to ensure complete removal of the tumour.

Mohs micrographic surgery, which is a special surgical technique to control tumour margins, is sometimes used to check that all the abnormal cells have been excised. Recurrence after Mohs surgery is reported to be around 1%.

Radiotherapy is sometimes used in addition to surgery if the tumour cannot be completely removed by surgery. Chemotherapy is ineffective.

The tyrosine kinase inhibitor imatinib mesylate is used to treat rare cases of local advanced inoperable or metastatic dermatofibroscarcoma characterised by COL1A1-PDGFB fusion gene, with 50% response rates.

What is the prognosis?

Follow-up with clinical examination of the site of the DFSP is recommended every 6 months for 5 years, and then annually.

The tumour only metastasises (spread beyond the skin) in 5% of cases. It spreads in 1% via lymphatic vessels to the regional lymph glands and in 4% via the blood stream, most commonly to the lung followed by the brain, bone and heart.

Local recurrences arise in 11–20% of cases, usually within 3 years of initial surgery, so follow-up is important. The recurrences are treated surgically as described for the original primary tumour.