

Scan Facts

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THE DERMATOLOGICAL FEATURES OF TUBEROUS SCLEROSIS AND THEIR TREATMENT

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Introduction

Tuberous Sclerosis (TS) is a complex genetic disorder characterised by abnormal collections of normal tissues involving many different organs of the body including the skin. It was first recognised as a separate entity in the 19th century. It is also known as Bourneville's disease, Pringle's disease or epiloia. TS is inherited but its effects are very variable even in the same family. 60-70% of cases are thought to be the result of new gene abnormalities. Recent UK studies have shown the incidence to be between 1 in 7,000 and 1 in 10,000. Usually the skin changes or epilepsy develops before the age of 5 years but the disease may remain latent until adult life. Approximately up to 90% of patients have skin changes.

Skin Changes in TS

The commonest skin changes seen in TS are: ovoid or ash leaf-shaped small white patches (hypomelanotic macules), angiofibromas, shagreen patches, forehead plaques and unguinal fibromas.

Hypomelanotic macules (ash leaf macules)

Small white patches on the trunk and limbs (Figure 1) occur before any other skin findings and are usually present in infancy. The commonest skin sign, they are found in up to 90% of patients, are usually multiple, irregularly scattered, sometimes have an ash leaf shape and are 1-3cm in length. The lesions are usually present at birth but are often difficult to see in the newborn without an ultraviolet light (Wood's lamp). Their presence can be helpful in diagnosis of children with infantile spasms but one or two such marks are seen in approximately 8 per 1000 of apparently normal newborn babies. Another pigment abnormality sometimes seen is the confetti lesion, an area with stippled loss of pigment, typically on the extremities.



Figure 1: Ash leaf macule on a leg

Facial Angiofibromas (adenoma sebaceum)

Facial angiofibromas (Figure 2) are considered to be diagnostic of TS but are only found in just over three quarters of patients and often appear several years after the diagnosis has already been established by other

means. They were originally called adenoma sebaceum because they were thought to be derived from sebaceous glands (grease glands) in the skin. However they have been shown in fact to be angiofibromas, composed of blood vessels ('angio') and fibrous tissue ('fibroma').

Angiofibromas usually first appear between 3 and 10 years of age, but sometimes later. They may rarely be present at birth or develop in infancy. They first appear as pin-point sized red spots affecting the nose, cheeks, chin and occasionally ears, which may later slowly enlarge up to 1-5 mm diameter. Significant enlargement of lesions often occurs around puberty (when they can be mistaken for teenage acne) but sometimes occurs into early adult life. Thereafter, they often show very little change. Larger nodules are firm to the touch. They are usually red and sometimes lightly pigmented, but may be pale, particularly on the nose. The nodules often form clusters around the nose and chin and may bleed and occasionally become infected, particularly if caught. Injury to the nodules during shaving can be a particular problem for patients and carers. In addition, prominent lesions may be disfiguring and require treatment primarily for this reason.



Figure 2: Facial angiofibromas

Shagreen patch

A shagreen patch (Figure 3) is an irregularly shaped, irregularly thickened, slightly elevated soft skin coloured patch usually on the lower back, which is made up of excess fibrous tissue. It is found in approximately 40% of TS patients but may not be apparent in young children. Although often quite large, shagreen patches are generally hidden by clothing and rarely cause disfigurement which warrants treatment. Larger or more protuberant lesions may cause discomfort or rub on clothing, and may require treatment for this reason.



Figure 3: Shagreen Patch on the lower back

Forehead plaques

Forehead plaques (Figure 4) are common and vary in size. They are generally situated to one side of the forehead and are raised, firm, red and often pigmented. If large, forehead plaques may be disfiguring, but can often be covered by adjustments in hair style.

Ungual fibromas

Ungual fibromas (Figure 5) are smooth, firm, flesh coloured outgrowths that arise adjacent to or from underneath the nails. They are usually 5-10 mm in length, but can be very large. They are normally considered specific for TS but a single lesion can occasionally occur after trauma. They are seen in 15-20% of TS patients and appear at or after puberty. Treatment may be required if unsightly, or for lesions which catch and are painful.

Café au Lait

Café au lait spots (flat brown marks) may also appear in TS but they are not a diagnostic sign.

Other cutaneous features

Poliosis, a localised patch of white hair, affecting scalp hair or eyelids occurs in 60% of cases of TS. Soft skin tags are sometimes seen around the head and neck, armpits and groin.



Figure 4: Forehead Plaque



Figure 5: Ungual fibromas

Types of Lasers

Lasers produce a powerful beam of pure light, which is absorbed by the skin to produce heat. The type of light, which may be visible or invisible, determines which kind of tissue is damaged. Where lesions have many blood vessels, lasers can be used to lighten and sometimes flatten them by damaging the blood vessels. Where there are few blood vessels, lasers can be used to vaporise lesions. Most lasers used to treat blood vessels in the skin produce a beam of light of blue, green or yellow colour, which

is strongly absorbed by blood. The blood is heated by the laser light, and this damages the blood vessels. The heat, however, can also diffuse outside the blood vessels into adjacent tissue, resulting in damage to neighbouring structures, including the fibrous tissue of angiofibromas (AF). This effect is often helpful in flattening lesions, but may produce scarring. There are several lasers which can have this effect, including the argon laser, the Nd:YAG (KTP) laser and the copper vapour or copper bromide lasers. The pulsed dye laser produces yellow light in much shorter bursts than these lasers, and more specifically damages blood vessels. As a result the risk of scarring is smaller, but the fibrous component of AF is not affected to any great extent.

Lasers which vaporise the skin do so by using infrared light to rapidly heat the water in the skin. Thin layers of skin are carried away by the steam which is produced (laser ablation or laserabrasion). These lasers can be used to flatten and smooth out lumpy skin. The commonest laser used for this purpose is the carbon dioxide laser which seals blood vessels as it vaporises, thereby reducing bleeding. Benefits over dermabrasion are that the laser is more precise and the lack of bleeding gives good visibility whilst operating. The laser beam seals blood vessels and nerve endings and so is thought to reduce postoperative swelling and pain. The amount of heat damage to the skin is minimal such that healing is rapid and the risk

of scarring minimised. The Erbium:YAG laser works in a similar way. This laser has less heating effect than the carbon dioxide laser and bleeding during and after treatment is more common, but the results are very similar.

Sometimes a test area is performed before treating the whole area. All laser treatments are uncomfortable, and although adults often require no anaesthetic, occasionally local or general anaesthetics are needed. Children often require either a local anaesthetic cream, or general anaesthetic depending on individual tolerance and the type and extent of treatment. Lasers are a potential hazard to the eyes, which are always protected during treatment.

The treatment of facial angiofibromas

Various treatments have been described for AF including curettage, excision, dermabrasion, cryosurgery and electrosurgery.

The preferred treatment for small and flat red lesions is the pulsed dye laser. It is often possible to produce a significant degree of improvement but complete clearing does not occur and several treatments are required. As these lesions are usually seen in younger children, a general anaesthetic is often required. The treatment causes dark bruising in the skin, which lasts for up to 2 weeks. The skin is fragile for a few days after treatment and sometimes blisters and

crusts. Significant scarring is very rare indeed with the pulsed dye laser, and although a little more likely if the areas are injured after treatment, this is rarely a problem, even in the most disabled individuals.

Larger red nodules often respond better to the argon, Nd: YAG (KTP) and related lasers, where flattening as well as lightening are often achieved. As with the pulsed dye laser, significant improvement but not complete clearing can be achieved, and several treatments are generally required. Although bruising does not occur, blistering and/or crusting are more likely but the healing time is similar. The scarring potential is marginally higher with these lasers, but remains low and is generally not significant. The wound care is similar to the pulsed dye laser.

Very large lesions, particularly when pale are better treated by vaporisation, usually with the carbon dioxide laser (Figure 6). Treatment is more painful than the other lasers, especially around the nose, where a general anaesthetic is generally required. Often only one or two treatments are required and results are generally very good with a low risk of scarring, although sometimes the skin will be more pale than normal as a result. The laser leaves a wound like a graze, which oozes for several days before crusting. Wound care is therefore more complicated often requiring dressings, but severe disability is not a contra-indication to treatment.

With all lasers, tanning or darkly

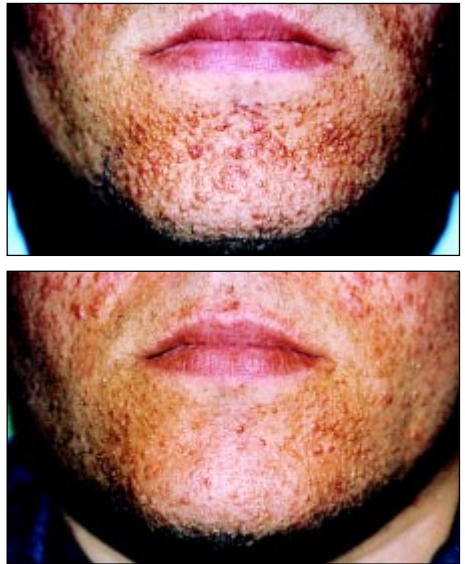


Figure 6: Angiofibromas on the chin before and after Carbon Dioxide laser treatment

pigmented skin is more difficult to treat, and the treated areas sensitive to sunburn. Rigorous sun protection is therefore required several months before and after treatment.

The decision when to treat can be a difficult one. If significant disfigurement or symptoms are present in early childhood, treatment may be carried out, but in the knowledge that further lesions may develop or enlarge, and require treatment later. Treatments later in life are more likely to be permanent, but the need for further treatment later cannot be excluded. Sometimes a range of different lasers may be needed to get the best results. The advantages and disadvantages of laser treatment have to be considered in each individual case.

The treatment of other skin lesions in TS

Shagreen patches can be shaved flat using a sharp knife called a dermatome or flattened by dermabrasion or laserabrasion. Ungual fibromas can be removed by surgical excisions, electro-surgery or laser ablation. The raised patch-like angiofibromas of the forehead or scalp can be flattened by laser vaporisation or may require removal by plastic surgery.

Summary

The skin lesions in TS are well recognised and some, particularly facial angiofibromas and unguinal fibromas, can cause considerable disability. A number of different treatments are available for these lesions, including lasers. Lasers are often preferred particularly for angiofibromas and unguinal fibromas. Lasers are expensive and although a full range of lasers necessary to treat skin lesions of TS are not very widely available, the number of centres able to offer some forms of laser treatment is increasing.

Glossary

lesion	an area of change in appearance or texture on the skin
macule	a flat area of altered skin colour
erythematous	redness
cryosurgery	uses freezing temperatures to remove superficial raised skin lesions
electrosurgery	the use of electricity to cauterise and or cut tissues
curettage	uses a spoon- or ring-shaped cutting instrument (a curette) to remove superficial raised skin lesions
dermabrasion	uses a hand-held high speed abrading machine to make superficial irregularities in surface of the skin more even

Further information on TSC and the work of the TSA can be obtained from: Mrs Diane Sanson, Head of Administration, PO Box 12979, Barnt Green, Birmingham, B45 5AN. Tel/Fax: 0121 445 6970
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