



Skin changes in Tuberous Sclerosis

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Outline

mTOR inhibitors eg Sirolimus

Vascular laser

Ablative laser

Tuberous Sclerosis

Genetic diagnostic criteria
The identification of either a <i>TSC1</i> or <i>TSC2</i> pathogenic mutation in DNA from normal tissue is sufficient to make a definite diagnosis of TSC. A pathogenic mutation is defined as a mutation that clearly inactivates the function of the TSC1 or TSC2 proteins (eg, nonsense mutation), prevents protein synthesis (eg, large deletion), or is a missense mutation whose effect on protein function has been established by functional assessment.
Note that 10 to 25% of patients with TSC have no mutation identified by conventional genetic testing, and a normal result does not exclude TSC, or have any effect on the use of clinical diagnostic criteria to diagnose TSC.
Clinical diagnostic criteria
Major clinical features:
Hypomelanotic macules (≥ 3 , at least 5 mm diameter)
Angiofibromas (≥ 3) or fibrous cephalic plaque
Ungual fibromas (≥ 2)
Shagreen patch (connective tissue nevus)
Multiple retinal hamartomas
Cortical dysplasias (includes tubers and cerebral white matter radial migration lines)
Subependymal nodules
Subependymal giant cell astrocytoma
Cardiac rhabdomyoma
Lymphangiomyomatosis (LAM)*
Angiomyolipomas (≥ 2)*
Minor clinical features:
"Confetti" skin lesions (1 to 2 mm hypomelanotic macules)
Dental enamel pits (≥ 3)
Intraoral fibromas (≥ 2)
Retinal achromic patch
Multiple renal cysts
Nonrenal hamartomas
Definite TSC:
Two major clinical features, or one major and two or more minor clinical features
Possible TSC:
Either one major clinical feature or two or more minor clinical features



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Fig 1. Tuberous sclerosis complex. Clinical appearance of fibrous cephalic plaques (FCPs) and FCP-like lesions smaller than 1 cm. **A**, FCP on the forehead of a lightly pigmented individual. **B**, Two lesions on the forehead of a darkly pigmented individual. **C**, Scalp FCP exhibiting decreased hair follicle density. **D**, Lesion smaller than 1 cm on the forehead, with onset at birth before the development of multiple facial angiofibromas.



“Red comets”



Sirolimus (aka rapamycin)

- Strengths vary from 0.1 -1% ointment
- Price varies a lot; check TSA
- Side effects are rare
 - pimples
 - slower wound healing
 - allergy
 - irritation
 - small amount of absorption by body

Taking sirolimus/everolimus orally also works



Availability of Topical Sirolimus

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Topical Sirolimus is a treatment for facial angiofibromas that are common among people with TSC (Tuberous Sclerosis Complex). A [clinical trial](#) to test the effectiveness and safety of topical Sirolimus cream at 0.5% and 1% is currently being conducted, and the results are hoped to lead to the approval of topical Sirolimus by the Therapeutic Goods Administration (TGA).

In the meantime, many people in Australia living with TSC are using topical Sirolimus for 'off-label' use, which means it is not officially approved for this purpose. This page provides general information only and does not constitute medical advice. Patients are advised to consult their doctors for more information about the suitability of this medicine for their individual case.

There are two steps involved in obtaining the cream:

- First, consult a dermatologist who may prescribe topical Sirolimus;
- Second, fill the prescription at a pharmacy that is able to make the cream.

Vascular laser

- Helpful for anything that's red
- 595nm Pulsed dye laser
- 1064nm Nd:YAG laser
- 532nm KTP laser
- Intense pulsed light (but no Medicare)



Vascular laser

- Bruising
- Patterning (honeycombing)
- Angiofibromas usually come back – ongoing maintenance
- Scarring & infection very rare

Ablative laser

“destructive lasers”

- Erbium:YAG laser
- Carbon dioxide laser

- Also older fashioned treatments like diathermy (electrocutation)



14100

Group

T1 - Miscellaneous Therapeutic Procedures

Subgroup

12 - Dermatology

Laser photocoagulation using laser radiation in the treatment of vascular abnormalities of the head or neck, including any associated consultation, if:

- (a) the abnormality is visible from 3 metres; and
- (b) photographic evidence demonstrating the need for this service is documented in the patient notes;

to a maximum of 4 sessions (including any sessions to which this item or any of items 14106 to 14118 apply) in any 12 month period

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Group

T8 - Surgical Operations

Subgroup

1 - General

Angiofibromas, trichoepitheliomas or other severely disfiguring tumours of the face or neck (excluding melanocytic naevi, sebaceous hyperplasia, dermatosis papulosa nigra, Campbell De Morgan angiomas and seborrheic or viral warts), suitable for laser ablation as confirmed by the opinion of a specialist in the specialty of dermatology—removal of, by carbon dioxide laser or erbium laser ablation, including associated resurfacing (10 or more tumours)



dr.deshan

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Questions?

<https://tsa.org.au/tsa-news/availability-of-topical-sirolimus/>

@dr.deshan

Referrals to **Liverpool Hospital Dermatology** can be made via these 3 options below

Email: SWSLHD-LivDR@health.nsw.gov.au

Fax: (02) 8738-4639

Post: Dermatology Department, 45 - 47 Goulburn Street, Level -1, Suite -7, Liverpool NSW 2170