
New guide for follow-up of tuberous sclerosis

FROM THE SPECIALTIES

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Patients with tuberous sclerosis complex (TSC) are followed up by multiple medical specialties throughout their lives. The revised guide can be used by paediatricians, general practitioners and other specialists.

Information on tuberous sclerosis should be easily accessible and up to date. Because it is a rare disease, limited knowledge among healthcare providers can lead to inadequate follow-up and treatment. A revised guide for the follow-up of tuberous sclerosis is now available, along with a clinical procedure for treatment with mTOR inhibitors [\(1, 2\)](#).

Tuberous sclerosis complex (TSC) is a multisystem genetic disorder affecting approximately 1 in 6,000–10,000 live births [\(3\)](#). The condition may be diagnosed in the fetus, in childhood or in adulthood, depending on symptoms and findings. It is estimated that 5–9 children are born with the diagnosis annually in Norway. The disease is caused by pathogenic variants in either *TSC1* or *TSC2*, leading to overactivation of the mechanistic target of rapamycin (mTOR) signalling pathway and the development of tumours in multiple organs. The brain, kidneys, heart, lungs and skin are most frequently affected [\(4\)](#). Drug resistant epilepsy is common [\(5\)](#), and tumours, particularly in the brain, kidneys and lungs, can lead to life-threatening complications [\(3, 4\)](#).

Systematic follow-up

Patients should be followed systematically by paediatricians in childhood and by multiple specialists in adulthood in order to monitor disease progression across organ systems [\(3\)](#). Early treatment of epilepsy is important and can improve cognitive development [\(3\)](#). There is no curative treatment, but immunomodulatory therapy with the mTOR inhibitor everolimus can slow the growth of certain tumours [\(3, 6, 7\)](#). Everolimus is generally well tolerated but can cause serious adverse effects [\(6, 7\)](#).

The Norwegian Centre for Rare Diseases at Oslo University Hospital has revised the Norwegian guide for follow-up and treatment of tuberous sclerosis [\(1\)](#) and devised a new clinical procedure for treatment with everolimus [\(2\)](#).

Systemic treatment with everolimus is, in some cases, recommended for drug-resistant epilepsy and for subependymal giant cell astrocytomas when surgical intervention is not feasible, or for renal angiomyolipomas with a risk of complications [\(3\)](#).

Patients may also benefit from topical formulations containing mTOR inhibitors (6). mTOR inhibitors may be considered in patients with lymphangioliomyomatosis or cardiac rhabdomyomas (1).

Clinicians can refer patients to the tuberous sclerosis network (8) for advice on medical follow-up and treatment. The network comprises specialists from all relevant disciplines. In Norway, management of the disease is not centralised, unlike in many other countries (4), and the network therefore plays an important role in ensuring equal access to high-quality care nationwide.

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