

Tuberous Sclerosis

(Optional sheet to be included where recipient of PHR has Tuberous Sclerosis)

The material in this sheet has been adapted from the Therapeutic Guidelines book 'Management Guidelines for People with Developmental and Intellectual Disabilities' and updated from the 2005 version 'Management Guidelines – Developmental Disability' which can be consulted for more detailed information.

Tuberous sclerosis is characterised by a triad of features: skin manifestations, intellectual disability and epilepsy. It is a multi-system genetic disease in which there are tuber-like growths in the brain and other major organs. However there is a wide variation in the spectrum and severity of its effects and in a small proportion of cases characteristic features may not be evident.

Tuberous sclerosis is an autosomal dominant condition, although two thirds of cases are due to new mutations. Virtually all cases arise from mutations in either the TSC1 or TSC2 genes, although other genetic and environmental factors can influence clinical severity. It occurs in fewer than 1 in 6000 children.

A neurologist usually makes the diagnosis with the assistance of brain, renal and cardiac scanning techniques, but the characteristic calcification is often evident on skull X-ray. Examination with a Wood's light may reveal hypomelanotic patches in the patient and a parent.

CHARACTERISTIC FEATURES

Skin Lesions: there are a number of types

- A butterfly-shaped yellow-reddish nodular facial rash appearing after the age of two years. These facial angiofibromas are wart-like lesions which tend to proliferate in puberty and can be confused with acne.
- Ashleaf spots are irregular areas of depigmentation which occur in about 60% of cases. These white patches can usually be detected in the first years of life. A Wood's light will help to identify these especially in fair-skinned individuals and babies.
- Shagreen patches (fibroepithelioma) are thickened yellowish areas of skin on the lower back.
- Subungual or periungual fibroma occurs under and around the fingernails and become more prevalent with age.

Intellectual Disability

About half of affected individuals will have an associated intellectual disability. There is a definite association between the severity of intellectual disability and the severity of epilepsy.

Seizures

Seizures have been reported in up to 80% of childhood cases but may be less frequent in adults. The first evidence of Tuberous Sclerosis may be infantile spasms. Infantile spasms, complex partial and myoclonic seizures are the commonest types encountered. The seizure disorder is often difficult to control but may become less of a problem with age.

Other Health and Behaviour Issues

- Cortical tubers are usually related to the severity of the epilepsy and intellectual disability.
- Subependymal hamartomas are markers for the disease but usually don't have complications. The rare one can undergo malignant degeneration, especially giant cell astrocytoma.
- The renal cystic disease is indistinguishable from autosomal dominant polycystic kidney disease and may present in infancy. Renal angiomyolipomas tend to present after puberty.
- Rhabdomyomata of the heart may cause problems in the perinatal period. Growths can also occur in the eye, bone, lung and liver.
- Sleeping problems are common in children.
- Behavioural difficulties are common and include hyperactivity in 59%, aggressive behaviour in 13% and autism in 50%.
- Dental abnormalities such as multiple enamel pits are commonly found.

KEY RECOMMENDATIONS FOR MANAGEMENT

- Genetic counselling for family. The causative gene mutations can be detected in up to 80% of cases and prenatal diagnosis is an option in future pregnancies for these families.
- Treatment of cardiac failure and arrhythmias which occur in association with rhabdomyomata.
- Management of seizures. Neurosurgery to remove cerebral hamartomas may occasionally help control focal seizures.
- Treatment of cutaneous lesions by laser therapy, dermabrasion and cautery.
- Management of behaviour.
- Renal ultrasound scans every 5 years.
- Regular dental care.

NATIONAL SUPPORT ASSOCIATION

Australian Tuberous Sclerosis Society

Freecall 1300 733 435

Web address: <http://www.atss.org.au>