

Tuberous Sclerosis

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Definition

Tuberous sclerosis is a genetic disorder that causes benign tumors to form in many organs including the brain, eyes, skin, heart, kidneys and lungs. It is characterized by some of the following abnormalities:

1. lesions in the cortex and white matter with seizures (93 per cent) and mental retardation (62 per cent);
2. retina or optic nerve hamartomas (53 per cent)
3. skin fibrous-angiomatic lesions (83 percent)
4. cyst-like areas in phalanges (back) (66 percent) showing evidence of sclerosis
5. renal (kidney and surrounding area) angiomyolipomata (45 to 81 per cent)
6. pit-shaped enamel defects on the teeth

Introduction

Tuberous sclerosis (also referred to as tuberous sclerosis complex (TSC) to distinguish it from Tourette's syndrome) is a genetic disease that affects multiple organs. Because of better testing methods, the estimates of those who have the disease have risen dramatically in the last few years. It is now believed that the condition occurs in approximately 1:6000 to 8000 live births and affects approximately 50,000 individuals in the U.S. and more than 1 million worldwide. Males and females seem to be equally affected. While it affects all races, it appears to be uncommon among blacks. It is believed to be inherited through a dominant trait in the 9th or 16th chromosome, with about 86 per cent representing fresh mutations from unaffected parents. It may be diagnosed anytime from birth to adulthood. Over 50 per cent of people with TS have normal intelligence and lead normal lives. The life expectancy for the majority of people with TS is normal even for those with seizures and learning disabilities.

The disorder can present itself from birth through adulthood, and there are a wide variety of manifestations and severity. For example, all patients with skin lesions do not develop mental deficiency or seizures or both. White macules (spots) are present at birth or early infancy in nearly all cases and can be seen by the use of a Wood lamp. Facial lesions appear in 50 per cent of children by age 5. The lesions vary in color from flesh to pink to yellow to brown and occur on the nose, cheeks and elsewhere including around and between the nails of the fingers and toes. They are classified as: "thumb-print" spots, "lance-ovate" or ash leaf spots where one end has a sharp tip and the other is rounded, and confetti spots which are tiny. At first the lesions across the nose and cheeks often look like a red pin point facial rash, but they later develop into bumps and the redness fades. Collagen accumulates in the skin at the nape of the neck and lower back and causes slightly elevated yellowish brown patches that have the texture of orange peel.

The seizures, which usually begin in early childhood, may first be myoclonic and later develop into grand mal seizures and are sometimes difficult to control. The earlier the seizures begin the more likely the occurrence of mental retardation. One in 2 people with TS will show signs of learning difficulty; the degree of disability can vary greatly. By the time a child with TS is two, it is usually clear if he or she will have learning problems. Autistic and hyperactive behavior patterns may occur.

Occasional abnormalities include rhabdomyomata and angiomatica of heart, cystic changes in lung, hamartomata of liver and pancreas, and hypertension.

Diagnosis

The essential feature of dementia is the development of multiple cognitive (mental) deficits. Forgetfulness is usually the first sign but to make the diagnosis of dementia other mental changes that affect the ability to function must also be present. These may include changes in thinking, decision-making, judgment, language, orientation to time and place, mood, behavior or personality. Changes usually begin slowly and worsen over time. After a complete history and physical examination doctors will usually perform blood tests to rule out treatable causes of decreased mental functioning like thyroid disease, vitamin deficiency, electrolyte imbalances, infections, or drug toxicity. When possible a mental status examination will be performed. Neuropsychological testing may also be needed. The doctor may order a CT or MRI scan to rule out brain tumor, subdural hematoma, stroke or hydrocephalus. Sometimes a spinal tap to examine cerebral spinal fluid will be done, and sometimes an EEG will be ordered. The only definitive way to diagnosis Alzheimer's disease is by autopsy when the characteristic loss of nerve cells, abnormal amyloid plaques and neurofibrillary tangles are seen under the microscope.

Prevention and Treatment

No preventative measures are known.

There is no known cure, but there is treatment available for several of its presenting symptoms. Seizures can be treated with anti-seizure drugs. If it becomes apparent that a child is not keeping up with peers in development, individual learning issues need to be addressed. A child may need help with speech and communication issues. Social workers, occupational therapists and health visitors are also good sources of information and aid. The advice of a dermatologist should be sought for the facial rash. Dermabrasion or laser therapy can be used to eliminate facial lesions and cosmetic surgery is sometimes an option. Intracranial hypertension caused by a benign tumor may need a shunting procedure or removal of the tumor. Progressive cystic renal involvement can be addressed by surgical decompression. The Tuberous Sclerosis Association is also a good source of information and support.

Emergency Situations – What can go wrong?

- **Status epilepticus** – prolonged seizure activity such as a seizure that lasts for more than 10 minutes or several seizures that occur one after another for 20-30 minutes.
- **Injury** – including bruising, concussion, fractures or even drowning if the seizure occurs during a bath.
- **Trouble breathing** – individual's lips may turn blue.

What to Do:

1. Clear the area around the individual, stay with him/her to prevent injury, DO NOT put anything in his/her mouth.
2. Try to write down what happened before, during and after the seizure and how long the seizure lasts.
3. Notify agency nurse/supervisor as soon as possible.
4. Call 911 if the seizure lasts longer than 5 minutes, if individual is injured or if he/she stops breathing.
5. Begin rescue breathing if you are certified to do so.

Conclusion

Tuberous sclerosis complex is a genetic disorder that is characterized by skin and eye lesions, seizures and mental retardation. Not everyone who has the lesions has seizures or mental retardation; in fact, 50 percent of persons with TSC live normal lives. Males and females are equally affected, and it appears to be more rare in African Americans. TSC can present from birth to adulthood; as soon as a family member is diagnosed, all immediate family members should be examined for the complex. There is no known cure. Treatment includes seizure control (which can be

difficult); early intervention services (when a child has developmental delays); cosmetic treatment; and investigation and removal of malignant tubers.

References

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