



"Most men's awareness doesn't extend past their dinner plates." - Scott Westerfeld

May is Tuberos Sclerosis Awareness Month

Tuberous sclerosis

Tuberous sclerosis, also called tuberous sclerosis complex, is a rare genetic disease that causes noncancerous (benign) tumors to grow in many parts of the body such as the skin, brain/nervous system, kidneys, and heart. The signs and symptoms of tuberous sclerosis vary widely, depending on where the tumors develop and how severely a person is affected.

Tuberous sclerosis is often detected during infancy or childhood. Some people with tuberous sclerosis have such mild signs and symptoms that the condition is not diagnosed until adulthood, or it goes undiagnosed. Others experience serious disabilities.

Although there is no cure for tuberous sclerosis, treatments are being studied. The course or severity of the disease can't be predicted, but with appropriate care, many people who have tuberous sclerosis lead full, productive lives.

Causes:

- ✚ Tuberous sclerosis is inherited. Changes (mutations) in two genes, TSC1 and TSC2, are responsible for most cases of this disorder.
- ✚ Only one parent needs to pass on the mutation for the child to get the disease. However, most cases are due to new mutations, so there usually is no family history of tuberous sclerosis.

Symptoms:

- ✚ **Skin abnormalities:** Some people with tuberous sclerosis have patches of light-colored skin, or they may develop small, harmless areas of thickened skin. There may be red patches on the face containing many blood vessels (adenoma sebaceum) and raised patches of skin with an orange-peel texture (shagreen spots), often on the back.
- ✚ **Nail defects:** Rough growths under or around the fingernails and toenails.
- ✚ **Tooth abnormalities:** Pitted tooth enamel.
- ✚ **Tongue lesions:** Rubbery noncancerous tumors on or around the tongue.
- ✚ **Seizures:** Lesions in the brain may be associated with seizures, which can be the first symptom of tuberous sclerosis. In small children, a common type of seizure called infantile spasm shows up as repetitive spasms of the head and legs.

- ✂ **Developmental delays:** Tuberous sclerosis can be associated with intellectual disability, learning disabilities or developmental delays.
- ✂ **Behavior problems:** Common behavior problems may include hyperactivity, raging outbursts, aggression, repetitive behaviors, or social and emotional withdrawal.
- ✂ **Communication and social interaction problems:** Some children with tuberous sclerosis have trouble with communication and social interaction. Some may have autism spectrum disorder.
- ✂ **Kidney problems:** Most people with tuberous sclerosis develop lesions on their kidneys, and they may develop more lesions as they age, sometimes damaging kidney function.
- ✂ **Heart problems:** These lesions, if present, are usually largest at birth and shrink as the child gets older.
- ✂ **Lung problems:** Lesions that develop in the lungs (pulmonary leiomyomas) may cause coughing or shortness of breath, especially with physical activity or exercise.
- ✂ **Eye abnormalities:** Lesions can appear as white patches on the light-sensitive tissue at the back of the eye (retina). These do not always interfere with vision.

Call your health care provider if:

- ✂ Either side of your family has a history of tuberous sclerosis.
- ✂ You notice symptoms of tuberous sclerosis in your child.
- ✂ Call a genetic specialist if your child is diagnosed with cardiac rhabdomyoma. Tuberous sclerosis is the leading cause of this tumor.

Coping and support:

If your child has been diagnosed with tuberous sclerosis, you and your family will face a number of challenges and uncertainties. For parents, the behavior issues that can accompany tuberous sclerosis may be the most challenging. Common problems such as raging outbursts, aggression, repetitive behaviors, or social and emotional withdrawal can be extremely hard to cope with. Remember that the behavior is not your fault — and it is not your child's fault, either. Let your child's doctor know if these problems develop. The earlier you and your child get help to develop learning skills to manage these problems, the more likely your child is to do well in the long term.

Prevention:

- ✂ Genetic counseling is recommended for couples who have a family history of tuberous sclerosis and who want to have children.
- ✂ Prenatal diagnosis is available for families who are known to have a history of this condition.
- ✂ However, tuberous sclerosis often appears as a new DNA mutation, and these cases are not preventable.

Reference Links:

- ✂ <http://www.ncbi.nlm.nih.gov/pubmedhealth/PMH0001791/>
- ✂ <http://www.mayoclinic.org/diseases-conditions/tuberous-sclerosis/basics/coping-support/con-20032953>
- ✂ en.wikipedia.org/wiki/Tuberous_sclerosis
- ✂ emedicine.medscape.com/article/1177711-overview

For any enquiry or assistance please contact: wellness@medicaretpa.co.in

Disclaimer: No information contained here should be relied on in making health decisions. Always check with your doctor or other health care provider.