

Neurofibromatosis (NF) and Tuberous Sclerosis Complex (TSC)

What are neurofibromatosis (NF) and tuberous sclerosis complex (TSC)?

- There are many types of neurocutaneous syndromes (syndromes that include skin findings). The most common is neurofibromatosis (NF). There are 7 types of NF. Although they are all different, they all have skin and neurologic findings. Neurofibromatosis type 1 (NF1) is the most common type, and, while it is inherited, half of the cases are new mutations.
- Tuberous sclerosis complex (TSC) is less common but can involve seizures, lesions on the face, and learning difficulties. Half of children with TSC have normal intelligence and a third have profound intellectual disabilities.

How common are they?

Neurofibromatosis type 1 occurs at approximately 1 in 3,500 births in the United States; TS, at approximately 1 in 6,000.

What are some common characteristics of children who have NF or TSC or of these conditions as children present with them?

- **Neurofibromatosis type 1:** The findings and the course of the condition vary greatly. Children with NF1 are often born with many light-brown birthmarks, called *café au lait spots*, or the spots may form over time. Their shins may be curved. Over time, these children develop small tumors under the skin that can range from pea to grape size. Half the children also have large tumors that can vary in size and can be disfiguring. Many of the tumors are benign, but they have an increased risk of malignant changes. Cancerous lesions are more common in adolescents and adults with NF1. When tumors grow in the brain, they can trigger seizures; seizures occur in 6% to 7% of children with NF1. When tumors grow in another organ, such as the kidney or heart, they can cause damage and blockage. Headaches are common and must be distinguished from brain tumors, but they respond well to medication. Hearing loss occurs in 10% of children with NF1. Different learning disabilities are common (50%), but intelligence is usually normal. Children with NF1 tend to have large heads but are frequently short in stature.



Child with café au lait spots

MICHAEL J. LYONS, MD, FAAP

- **Tuberous sclerosis complex:** Seizures are common, and 80% to 90% of children with TSC have a seizure disorder. Autism spectrum and behavior disorders are also common, but reported incidence varies widely. The most common skin finding is an ash leaf spot, which is a light-colored oval lesion. As these children age, acne-like lesions can develop on their faces.

Neurofibromatosis (NF) and Tuberous Sclerosis Complex (TSC) *(continued)*

Who might be on the treatment team?

- Pediatrician/primary care provider in the medical home
- Pediatric subspecialists, including neurologists, geneticists, and developmental-behavioral specialists
- Pediatric surgeon (as needed)
- Speech-language and physical therapists

What adaptations may be needed?

Medications

- Children with NF or TSC may need seizure medication. No medications are available to stop the growth of the tumors.
- All staff who will be administering medication should have medication administration training (see Chapter 6).

Physical Environment and Other Considerations

- A behavioral health aide may be needed.
- Sunscreen is especially important, to prevent damage to the skin.
- Children with NF1 can usually have a normal diet and normal play activities.

What should be considered an emergency?

- Seizure
- Blood in the urine
- Appearing pale or having an irregular pulse
- Irregular eye appearance

What types of training or policies are advised?

- Seizure precautions
- Training to administer medications to stop seizures (diazepam)

What are some resources?

Children's Tumor Foundation: www.ctf.org, 1-800-323-7938

