

Don't Take These Lesions at Face Value

For about 3 years, dozens of lesions have been growing in size and prominence in the middle of this 8-year-old girl's face. Under the presumption that they represent acne, she was treated for that condition, with no benefit. At a loss, her primary care provider refers her to dermatology.

The patient has a history of seizures—beginning early in life—that are under decent management. There is no family history of similar problems, nor of any related disease.

She has been having behavioral problems at school, where she is a year behind her peers. Some of her counselors have suggested she has autism spectrum disorder—a suggestion with which her family strongly disagrees. The patient's behavior in clinic is well within normal limits, as is her general appearance.



EXAMINATION

Distributed in a butterfly-like pattern, the facial lesions cover the middle portion of her face and nose. Each of the 1-to-2-mm papules is identical: firm, round, and pink; none are pustular. There are no comedones on her face.

Further examination reveals 2 to 4 hypopigmented macules scattered around her trunk. On her upper back, there is a solitary, pink, polygonal plaque covered with fine, soft papules. On her trunk, there are 4 café-au-lait macules, each ranging from 1 to 3 cm;



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none are seen on her axillae. Her nails and teeth are free of notable lesions or changes.

Given the findings, the most likely diagnosis is

- Tuberous sclerosis
- Neurofibromatosis type I
- Albright disease
- Addison disease

ANSWER

The correct answer is tuberous sclerosis (TS; choice “a”).

DISCUSSION

TS is a rare, genetically transmitted complex that was originally described by Désiré-Magloire Bourneville in 1880. It once was considered *extremely* rare, with estimates of about 1 in 100,000 births. But the invention and proliferation of MRI technology has helped to establish an incidence closer to 1 in 12,500 births—because imaging detects the characteristic involvement of internal organs, especially the brain, kidneys, and lungs, and helps us connect it to multisystem findings in affected patients.

TS involves defects or mutations of 2 genes: *TSC1*

or *TSC2* (or both); these genes act as tumor growth suppressors by regulation of cell proliferation and differentiation. Two-thirds of TS cases are caused by spontaneous mutation, while one-third are caused by autosomal dominant inheritance with quite variable penetrance.

This results in a wide spectrum of disease, with many cases starting shortly after birth with life-threatening seizures caused by “tubers” (tumors) in the brain, followed later by a wide variety of findings (as exhibited by this patient). These cases have a poor prognosis; others, with incomplete penetrance, are far less severe.

This patient’s variety of skin lesions are characteristic of TS. The facial lesions are benign hamartomatous lesions called *angiofibromas*. The hypopigment-

ed macules on her trunk are classically dart-shaped; these are known as *mountain ash leaf spots*. The leathery, bumpy plaque on her upper back is a *shagreen patch*. And although café-au-lait macules (albeit in greater number and size) are commonly associated with neurofibromatosis, they are often seen in TS as well. Behavioral and academic problems, as well as features similar to those seen in autism, also are commonly observed in patients with TS.

Once TS is diagnosed, the patient is often referred to a genetic specialist for confirmation and counseling. The primary care provider should also be advised of the diagnosis with regard to ongoing management. As for the patient’s skin manifestations, angiofibromas can be removed with laser therapy or dermabrasion. **CR**