Tuberous Sclerosis in a 15-Year-Old Girl

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Tan-pink acneiform lesions on the face of a 15-year-old girl had not responded to topical acne therapy. A 1 × 0.5-cm, elevated subcutaneous nodule was noted on the right lateral knee. The lesions on her face and knee had been present for 11 years. The family history was noncontributory.

The patient, who did not speak until after her second birthday, had difficulty in comprehending verbal commands and could not read or write. Hearing in the left ear had been impaired since birth. Plain lateral skull films showed several calcifications (C, arrow). Axial CT scans of the brain revealed multiple subependymal periventricular calcified tubers (D, arrow). No other organs were involved. The lateral knee nodule was thought to be a hamartoma; a biopsy was not performed.

Robert P. Blereau, MD, of Morgan City, La, diagnosed tuberous sclerosis. This autosomal dominant disease of variable penetrance features the clinical syndrome of multiple hamartomas of the skin, CNS, kidney, heart, retina, and other organs. At least 1 in 10,000 persons is affected; one third of cases are familial. About 25% of patients present with a triad of epilepsy, angiofibromas (adenoma sebaceum), and mental retardation. The younger the patient's age at clinical presentation, the greater the likelihood of retardation.

Adenoma sebaceum of the face is the most common skin manifestation; it typically appears at age 2 or 3 years. The lesions, which may resemble acne, are angiofibromas that rarely involve the sebaceous glands.

Other cutaneous lesions associated with tuberous sclerosis are:

- Hypopigmented macules. Among the earliest signs of the disease are white macules and white tufts of hair with no underlying skin pigmentation. As many as 90% of patients have hypomelanotic macules that are present at birth or develop in infancy. A Wood lamp examination can accentuate these ash-leaf-shaped lesions, which may be difficult to discern on white skin.
- Shagreen patches. These soft, skin-colored to yellow, pigskin-textured plaques frequently occur during early childhood. The usually solitary lesions are commonly found in the lumbosacral area.
- Subungual and periungual fibromas of the digits. These lesions arise from the stratum of the fingers and toes; they occur in about half of all adolescents with the disease.

Brain lesions occur in more than 90% of patients; they are associated with mental retardation in fewer than 50% of those affected. Cortical tubers may be noted on radiographs and CT and MRI scans shortly after birth. A brain tuber occasionally may differentiate into a malignant astrocytoma. Generally, the higher the number of brain tubers, the greater the degree of retardation. Cardiac rhabdomyomas, which are found in about half of affected children, may lead to congestive heart failure and arrhythmias. Among the other systemic manifestations of tuberous sclerosis are...
hamartomas of the kidneys or polycystic renal disease, which can cause hematuria, pain and, occasionally, renal failure. Angiomyolipomas may cause generalized cystic or fibrotic changes in the lungs. Retinal hamartomas are common but rarely cause vision problems. Periosteal bone formation and phalangeal pseudocysts are sometimes noted on x-ray films.

Diagnosis of this disease is based on the presence of the clinical syndrome, typical skin lesions, and brain calcifications. MRI is the preferred imaging study; however, plain skull films can be elucidating. CT scans can exclude calcified subependymal nodules. Calcifications may be seen on imaging studies before skin lesions appear.

Baseline studies include renal ultrasonography, echocardiography, and chest roentgenography. The differential diagnosis includes acne; warts, especially on the digits; neurofibromatosis; nevoid basal cell carcinoma syndrome; and multiple trichoepitheliomas.

Treatment options for cosmetic removal of facial lesions are excision, electrosurgery, cryosurgery, dermabrasion, and laser therapy. Anticonvulsants are used to control seizures. Mental retardation is treated supportively. Increased intracranial signs and symptoms warrant immediate testing and neurosurgical intervention.

Malignancies are rare; renal cell carcinoma and aggressive astrocytomas occur most commonly. Life expectancy is normal except in those with mental retardation. Heart and kidney problems can contribute to morbidity and mortality.

REFERENCES:

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