Introduction:
Tuberous Sclerosis (TS) is a rare genetic condition affecting multiple organ systems. A variety of signs and symptoms develop secondary to the formation of benign tumors (hamartomas). These tumors can cause a spectrum of conditions such as developmental delay, epilepsy, arrhythmias, kidney failure, lung collapse, vision loss, and behavioral problems.

A thorough evaluation is required to determine the impact of these tumors and frequent screenings to assess organ function.

Most cases of TS are determined to be de novo mutation. The disease can also be inherited in an autosomal dominant pattern.

Objective:
The goal of this report is to highlight a unique presentation of this disease, the importance of a broad differential, and the need to further investigate changes in seizure patterns.

Case Description:
Initial Presentation:
An 18-month-old male with a history of complex febrile seizures presented with a 5-month history of staring spells and increasing seizure activity over one week associated with the inability to use the left arm. Initial neuro workup for complex febrile seizures was negative. However, given changing seizure quality and lack of fevers, further workup was indicated. Differential at that time included partial complex seizures, epilepsy disorder, and intracranial mass.

Physical Examination:
- (+) Developmentally appropriate male. Normal muscle bulk and tone. No focal deficits, patient at neurological baseline.
- (+) One hypopigmented macule on lower left abdomen.

Approach:
Diagnostic Criteria for Tuberous Sclerosis

The detection of either TSC1 or TSC2 pathogenic mutation in DNA is sufficient for the definitive diagnosis of Tuberous Sclerosis Complex.

Definitive clinical diagnosis: 2 major features or 1 major with 2+ minor features.
Possible clinical diagnosis: 1 major features or 2+ minor features.

Major Features:
a. Hypomelanotic macules (3+, at least 5mm diameter)
b. Angiofibromas (3+) or fibrous cephalic plaque
c. Ungual fibromas (2+)
d. Shagreen Patch
e. Multiple retinal hamartomas
f. Cortical dysplasias
g. Subependymal nodules
h. Subependymal giant cell astrocytoma
i. Cardiac rhabdomyoma
j. Lymphangiomyomatosis (LAM)
k. Angiomylipomas (2+)

Minor Features:
a. "Confetti" skin lesions
b. Dental enamel pits (3+)
c. Intraoral fibromas (2+)
d. Retinal achoric patch
e. Multiple renal cysts
f. Nonrenal hamartomas

Significant Imaging:
MRI Brain: Multiple findings suggestive of tuberous sclerosis, including cortical tubers, subependymal hamartomas, and possible developing subependymal giant cell astrocytoma.
Echocardiogram: Multiple nonobstructive intracardiac masses, likely rhabdomyomas. Normal cardiac function.
Renal US: Normal sonographic appearance of the kidneys. No cysts or angiomylipomas.

Figure 1. Hypomelanotic macule commonly seen in Tuberous Sclerosis, also commonly known as "Ash-leaf Spots". Patient's presenting with multiple should prompt suspicion for TS. Can be present at birth or early infancy.

Figure 2. Echocardiogram of the patient's heart showed four rhabdomyomas found in the right ventricle and one in the left ventricle. Patient with normal cardiac function.

Treatment:
Treatment of TS consists of close monitoring of the organs commonly affected by hamartomas including:
- MRI and US of the brain, heart and kidneys
- BUN/Cr and Vitamin D to monitor kidney function
- EKG for arrhythmias
- Skin and eye exams
- Blood pressure monitoring

The rest of treatment is targeted at symptoms management. Such as antiepileptic medication, surgical removal of tumors, BP mediations, and extra educational support.

Conclusions:
- Changes in seizure pattern warrant further work up, imaging if not completed before, brain MRI is an important modality for further evaluation as in this case it was diagnostic.
- Skin examination is also important as it can guide clinical suspicion especially in younger age groups.
- Close monitoring is required for patients with TS as multiple organs can be affected at varying degrees.

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