GENETICS AND GENODERMATOSES

BOURNEVILLE-PRINGLE DISEASE: THE MISLEADING ACNE!!

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Background: Tuberous Sclerosis, also known as Bourneville's disease, is an autosomal dominant syndrome with variable clinical expression. We report a case of a 12-year-old little girl with tuberous sclerosis, treated for 2 years as moderate inflammatory acne by general practitioner.

Observation: It's a young female patient of 12 years old from a consanguineous family, having a history of suspected epileptic seizures in childhood stopped at 5 years old with normal psychomotor development.
Physical examination revealed several small-scattered facial angiofibromas that were histologically determined by skin biopsy. Hypomelanotic macules on her hands and back, shagreen patches on her forehead and left inguinal area. Periungual fibromas were seen in the fifth right toe.
The MRI of brain showed multiple calcified sub-ependymal nodules and cortical tubers. Neurological consult shows: normal status of conscience, without meningeal signs of irritation, cranial nerves without deficit, and no ataxia. Cardiological consult and ECG were normal. Multiple hamartomas or retinal achromic patches were noticed by ophthalmologic evaluation. Radiography of the chest was normal but truncal- abdominal CT had showed small size hepatic and renal angiomyolipoma. So with the above clinical findings, a diagnosis of Tuberous sclerosis was made, and the patient is under regular monitoring.

Key message: STB is a fairly common disease but it's often underdiagnosed , with variable clinical expression dominated by neuropsychic and renal involvement. Therapeutic management must be multidisciplinary.