

Images in medicine

Tuberous sclerosis: a case report



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An 11-year-old girl, from a consanguineous family. With history of suspected akinetic seizures and normal intelligence. Physical examination revealed several small-scattered facial angiofibromas that were histologically determined by skin biopsy. Hypomelanotic macules, shagreen patches on her back. Periungual fibromas were not seen. CT brain showed multiple calcified sub ependymal nodules and cortical tubers. Cardiological consult and ECG were normal. Neurological consult shows: normal status of conscience, without meningeal signs of irritation, cranial nerves without deficit, and no ataxia. No hamartomas or retinal achromic patches was noticed by ophthalmologic evaluation. Radiography of the chest, truncal CT was without abnormality. So with the above clinical findings, a diagnosis of Tuberous sclerosis was made, and the patient is under regular monitoring. Tuberous Sclerosis, also known as Bourneville's disease, is an autosomal dominant syndrome with variable clinical expression. The dermatologist plays an essential role in the history of the disease, since skin manifestations represent the most prevalent clinical features, enabling early diagnosis and intervention in its natural course.



Figure 1: (A) several facial angiofibromas; (B) hypomelanotic macules and shagreen patches; (C,D) CT brain with multiple calcified sub-ependymal nodules and cortical tubers

