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Bourneville Tuberos Sclerosis- Difficulties of the Diagnosis- A Case Report

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The imagery investigations confirmed the existence of the brain hamartomas and of the angiomyolipomas of the kidneys, supporting the diagnosis of Bourneville disease.

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Bourneville Tuberos Sclerosis- Difficulties of the Diagnosis- A Case Report

Mihaela Lungu ^α & Alina Ursoiu ^σ

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I. CASE REPORT

The patient C.A., sixteen years old, female, takes notice of the appearance-approximately two years ago- of some cutaneous lesions on the chin, initially considered by herself to be acne vulgaris. These

lesions were the only signs which determined her to ask for a medical examination, the teen-ager having no other complaints.

In her personal history there are no diseases and in her family history we find 4 healthy brothers and no hereditary pathology. Anamnesis excludes epilepsy and mental retardation.

The cutaneous lesions have a typical aspect of angiofibromas, with a red colour, are situated in the chin region- those with a bigger size, maximum 2 mm, in the nasogenian folds and on the cheeks, where the lesions have small dimensions. These lesions are firm on palpation and they are not accompanied by other symptoms (pain, prurigo). The clinical aspect is of angiofibromas (fig.1).



Fig.1 : Angiofibromas

The general clinic examination, the neurological examination and the examination of the fundus of the eye were normal.

The cutaneous lesions raise the suspicion of Bourneville Tuberos Sclerosis, for this reason indicating imagistic investigations.

On the CT examination of the brain there are found hyperdense formations with the diameter of 2, 4 and 5 mm, subependymally situated in the sidewall of the third ventricle, with a typical aspect of hamartomas (fig.2)

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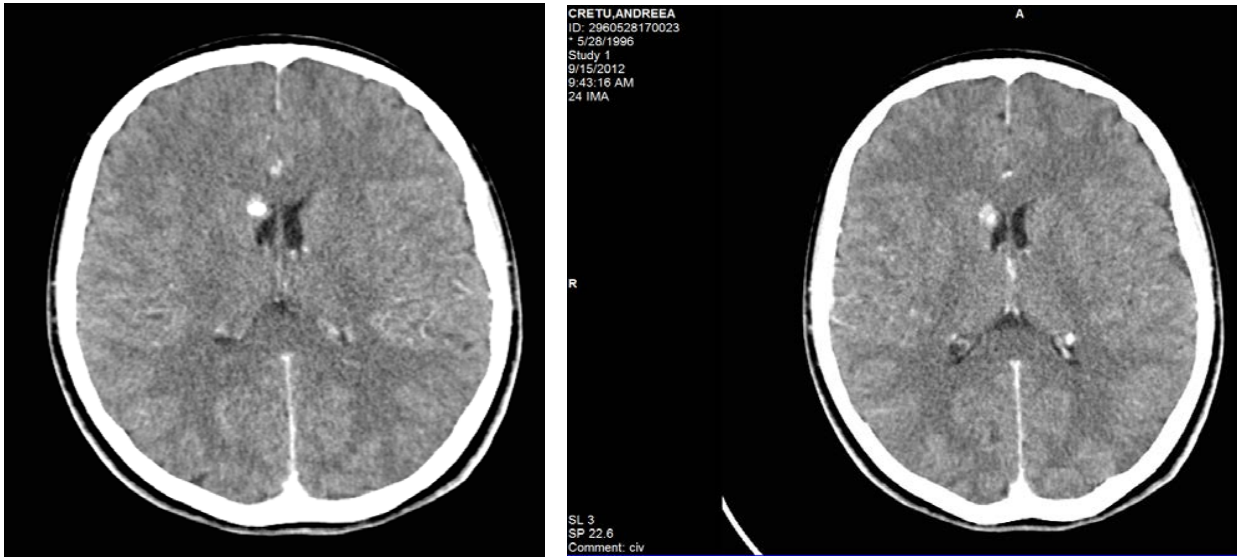


Fig. 2 : Subependymally hamartomas

The abdominal-pelvic CT examination shows multiple formations with a fatty density included, with diameters between 2-6 mm, situated on the cortical zone of the kidneys, on both sides. In the left kidney it can be visualized a pericalyceal superior formation of 18 mm diameter, which highly captures the contrast substance, compatible with an angiomas lesion (fig.3) The thoracic CT and the echographic examinations of the heart exclude lungs or heart lesions.

The conclusions of the imagistic investigations are: brain hamartomas and angiomyolipomas of both kidneys.

These lesions, typically associated with the cutaneous angiofibromas, allow the diagnosis of Bourneville Tuberos Sclerosis.

The differential diagnosis was made with other forms of facomatosis (neurofibromatosis) and with other dermatological diseases (acne vulgaris, trichoe-piteliomas, syringomas).

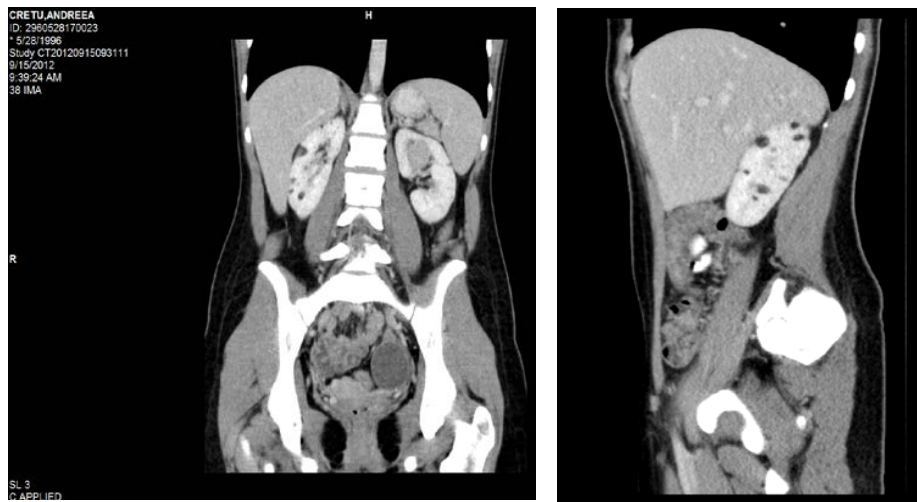


Fig. 3 : Angiomyolipomas of the kidneys

II. DISCUSSIONS

The typical cutaneous lesions (angiofibromas) raised the suspicion of Bourneville disease, later confirmed by the imagistic investigations.

A „forme fruste” of Bourneville Tuberos Sclerosis was confirmed, in a sporadic case, without epilepsy or mental retardation, with typical cutaneous lesions and multiple tumors of the brain and the kidneys.

The case is a sporadic one, the patient having 4 healthy brothers (needed to be investigated). In the Bourneville disease, the heredity is proved in 14-50 percent of the cases.

The forme fruste of Bourneville disease is mentioned in the neurological literature to be frequent.

The therapy focuses on the neurological observation (risk of refractory epilepsy), genetic advise, professional orientation.

The treatment of the angiofibromas is limited to more or less invasive methods: cryotherapy, surgery, laser phototherapy, methods which are frequently followed by the reappearance of the lesions and complications (keloid scars). A very recently case study proposes for therapy the application of an immunosuppressive agent, of a new generation, with good results, but there are necessary randomised studies to confirm the safety of a long time treatment.

Bourneville Tuberous Sclerosis is an inherited disease of an autosomal dominant type, with a high level of new mutations, characterized by multifocal tumors, malformations and typical cutaneous lesions. The locus for the genetic mutations are 9q34 and 16p34.

It is clinically characterized by the triad: refractory epilepsy, mental retardation (sometimes severe) and angiofibromas of the face (typical for 70-90 percent of the patients, along with other types of cutaneous lesions). In the brain, there are hamartomas and calcified tubers nodules (cortical brain stones), which determine refractory epilepsy and hydrocephalus. The neurons have three to four normal size. There are also lesions which involve other types of cells: fibroblasts, cardiac myoblasts, angioblasts, developed in an excessive number and size. These are the causes of the appearance of angioleiomyomas of the kidneys, liver, tests, adrenal gland, rhabdomyomas of the heart, retinal glial nodules. It is presupposed that some inhibitory growth factors are blocked at a certain stage of the embryonic life, fact which leads to the hyperplasia and hypertrophy of the well-differentiated cells.

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