

AMÍGDALAS PÉTREAS E PÁPULAS DE MARFIM: COMBINAÇÃO PATOGNOMÔNICA RARA - RELATO DE CASO NEURORRADIOLOGIA

DADOS DO CASO

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ABSTRACT

Lipoid proteinosis (Urbach-Wiethe syndrome) is a rare autosomal recessive genodermatosis caused by ECM1 gene mutation, characterized by progressive deposition of eosinophilic amorphous hyaline substance and generally benign and chronic disease course.

CLINICAL HISTORY

Female, 19y. Since 11 months of age: papules, diffusely infiltrated and thickened skin. Mainly distributed on the back, sacral region, face, eyelid margins, shoulders, and elbows. Alopecia is observed in the occipital region (Figure 1), along with infiltrated skin at the hairline, and a hyperpigmented infiltrated plaque in the axillary region.

IMAGING FINDINGS

In the central nervous system investigation, computed tomography of the head with and without contrast revealed the presence of coarse calcifications in the temporal lobes, specifically in the uncus region, bilaterally (Figures 2,3,4 and 5). These findings may be associated with future neurological sequelae such as ataxia, seizures, and psychosis.

DISCUSSION

Lipoid proteinosis (LP), Urbach-Wiethe syndrome, or

cutaneous-mucosal hyalinosis, is a rare autosomal recessive genodermatosis caused by ECM1 gene mutation characterized by progressive deposition of eosinophilic amorphous hyaline substance in the skin and mucous membranes [1]. Clinically characterized by yellowish or ivory-colored papules, which can form plaques, mainly on the face, neck, friction areas, and eyelid margins [2]. The course of LP is benign and chronic; however, early diagnosis is crucial for initiating treatment due to its impact on the patient's quality of life [3]. The typical findings on exams include dense bilateral calcifications in the mesial temporal lobes and striatum. On computed tomography (CT), these are symmetric hyperdense curvilinear ("comma-shaped") lesions, while on magnetic resonance imaging (MRI), they appear hypointense on T1, T2, and GRE sequences. Involvement of the amygdala is pathognomonic [2].

The patient in question presented frequent dermatological lesions typical of LP, such as skin infiltration and thickening in extensor areas. The extensive alopecia plaque in the occipital region, although less common, was also present. The anatomopathological examination of skin fragments from the scalp and armpit showed findings that favor the diagnosis of lipoidoproteinosis. CT scan of the head showed coarse calcifications in the temporal lobes, specifically in the uncus region, bilaterally.

The presence of papules on the eyelid margins (moniliform

blepharosis) occurs in 50% of cases and is a pathognomonic sign of the disease. The coarse calcifications in the temporal lobes support the diagnosis, highlighting the importance of radiologists' familiarity with the condition to aid in suspicion and early diagnosis/treatment, aiming to minimize the impact of LP on the individual's quality of life.

DIFFERENTIAL DIAGNOSIS

The differential diagnosis of this pathological entity should

be done with erythropoietic protoporphyria, whose lesions are restricted to areas exposed to the sun, with no involvement of mucous membranes. Other differential diagnoses are papular mucinosis, leprosy, amyloidosis and xanthomatosis cutaneous

TEACHING POINTS

Urbach-Wiethe disease (Lipoid proteinosis) is a rare disease that primarily affects the skin, upper respiratory tract, and central nervous system. Involvement of the amygdala is pathognomonic.

REFERENCES

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FIGURES



Figure 1 Occipital alopecia



Figure 2 Head CT Volume Rendering Reconstruction demonstrates gross symmetrical calcifications in the region of the bilateral uncus (white arrows).

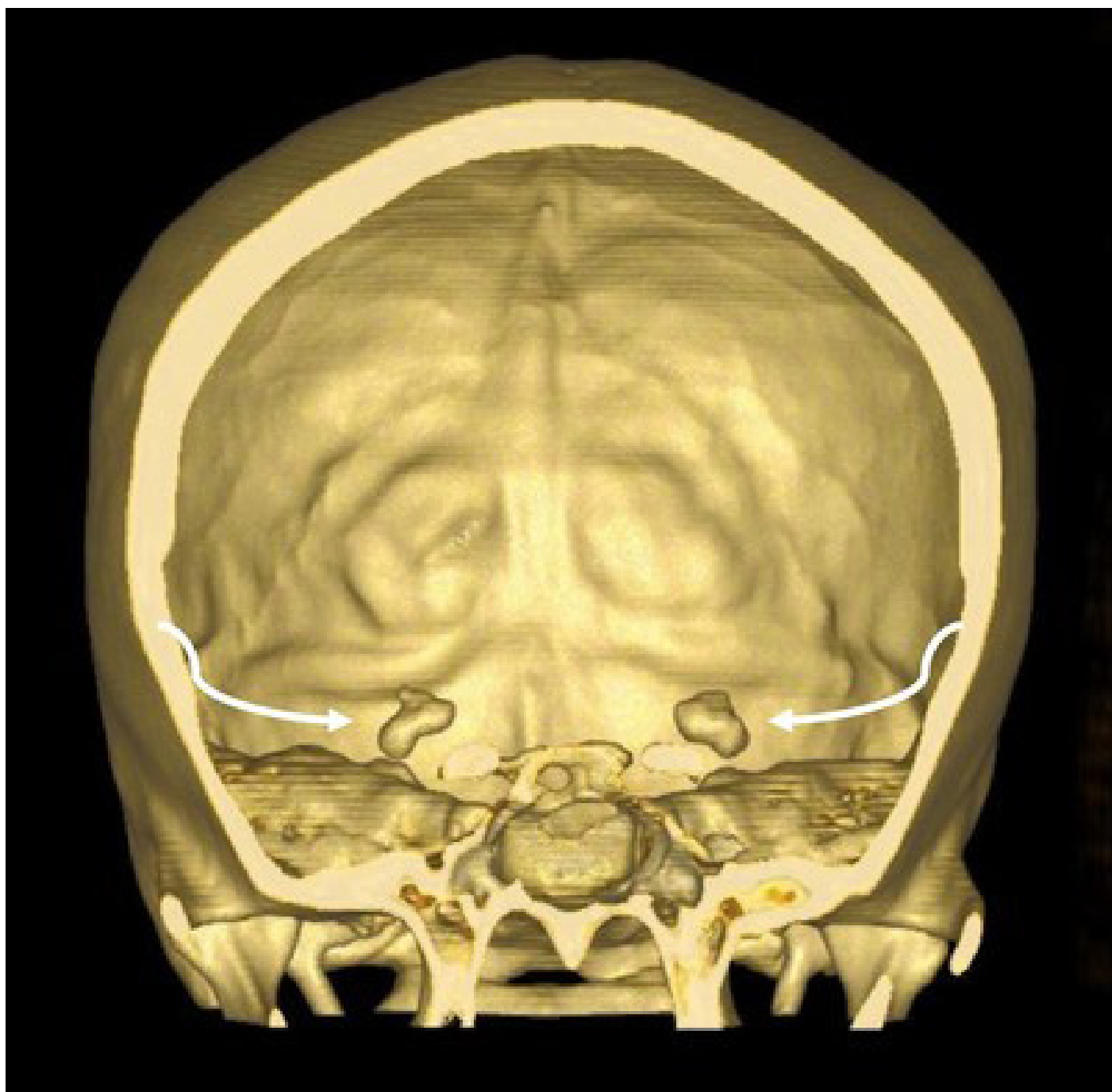


Figure 3 Head CT Volume Rendering Reconstruction demonstrates gross symmetrical calcifications in the region of the bilateral uncus (white arrows).

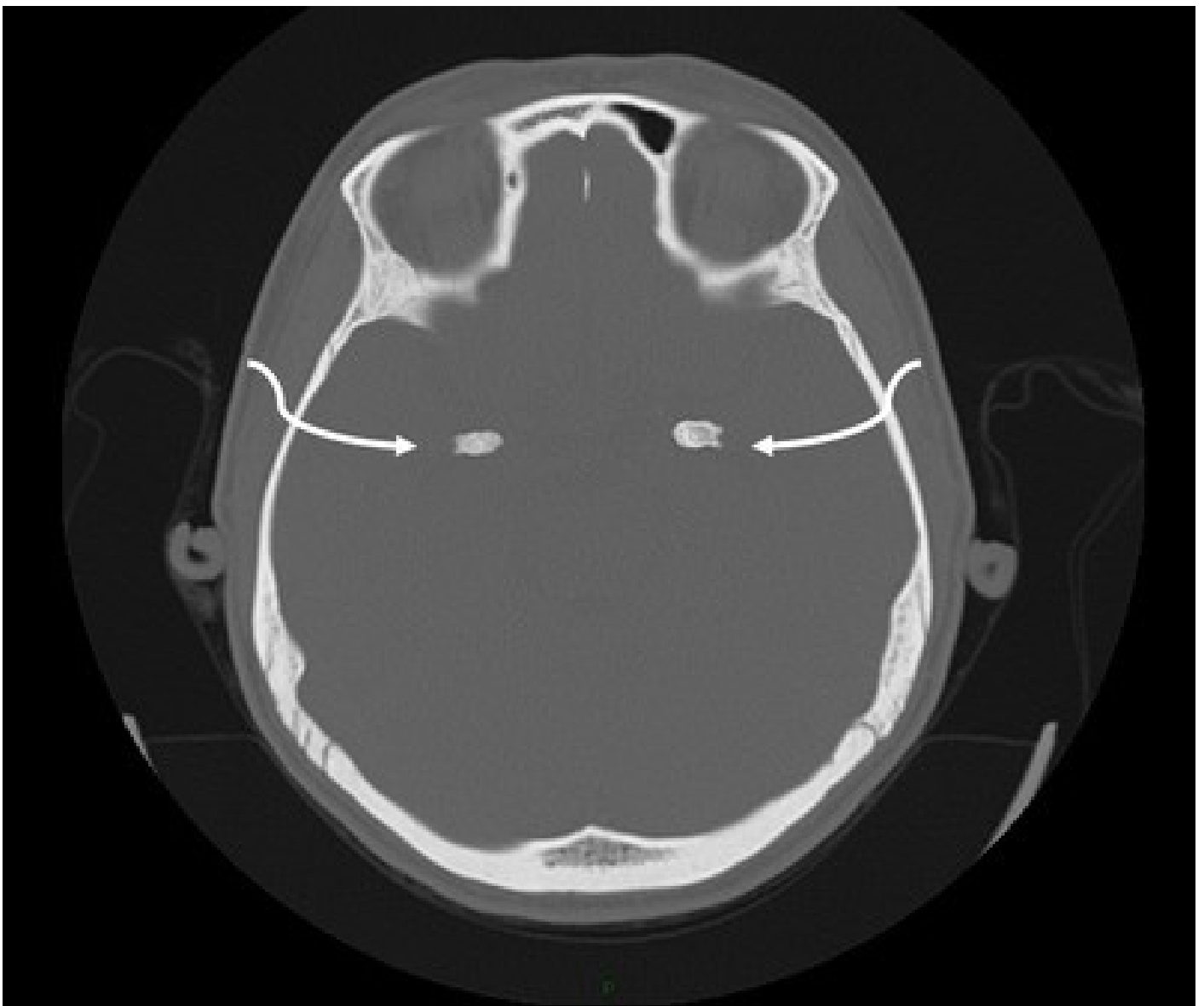


Figure 4 Skull computed tomography in bone window (B), showing symmetric coarse calcification in the bilateral uncus region (white arrows).

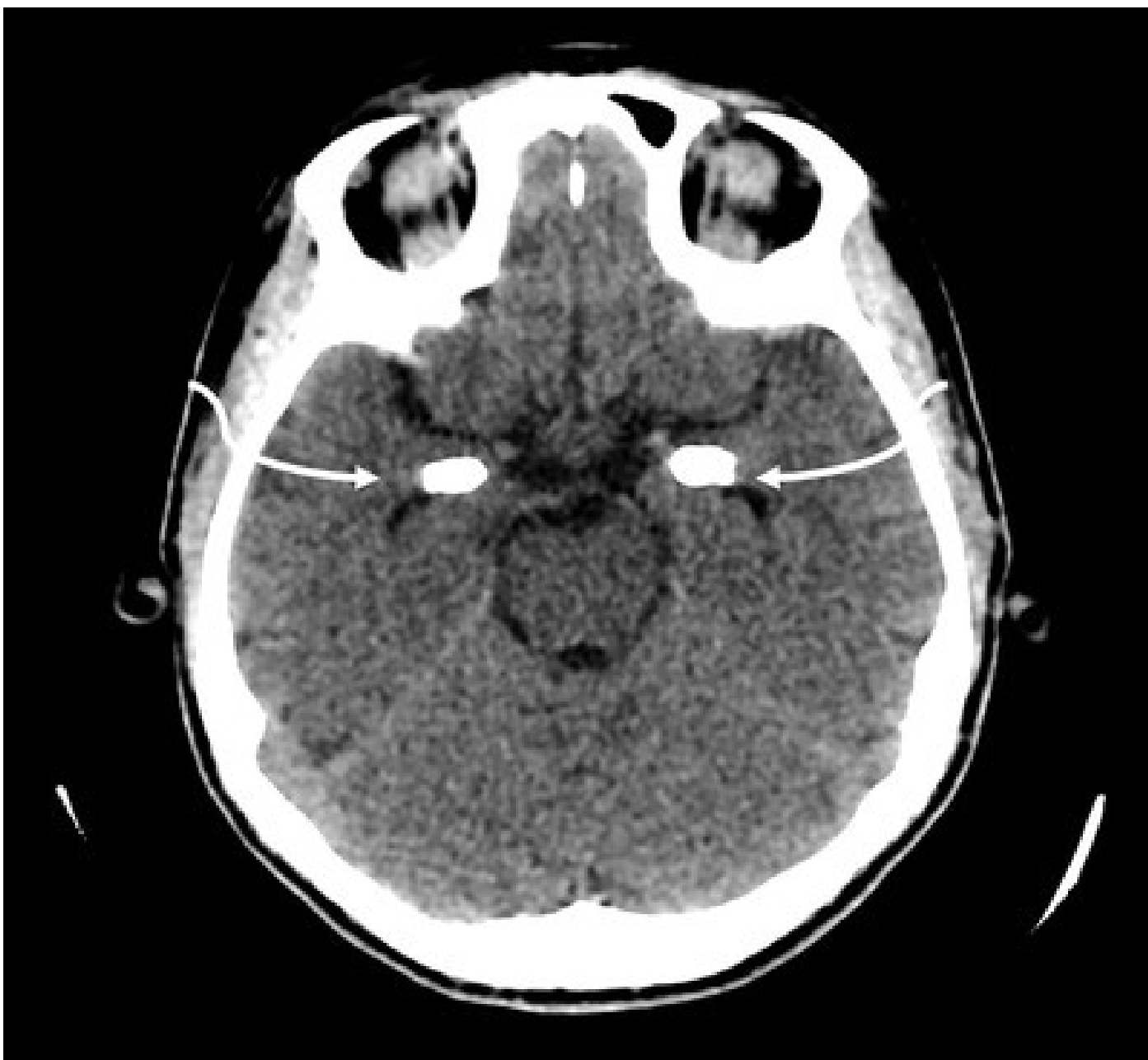


Figure 5 Skull computed tomography in soft tissue window without contrast showing symmetric coarse calcification in the bilateral uncus region (white arrows).