Cerebrotendinous Xanthomatosis with Lung Involvement and Acquired Ichthyosis

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Abstract

We report a case of a 28-year-old man with xanthomas over the extensors of the limbs, cataract, chronic diarrhea and various neurological disabilities. There was also an ichthyosis, which predominate over the lower limbs. Chest CT-scan showed bilateral apical opacities with cystic images in the culmen. Brain MRI revealed bilateral and symmetric T2 and FLAIR hyper-signals sequences in the region of dentate nucleus and adjacent cerebellar and periventricular white matter. We have reported the first case of CTX in Senegal, which is remarkable by an acquired ichthyosis and excavated lung lesions simulating a tuberculosis.

Keywords: Cerebrotendinous xanthomatosis; Cholestenol; Ichthyosis

Introduction

The cerebrotendinous xanthomatosis (CTX) is a genetic condition resulting from a deficiency of the mitochondrial sterol 27-sterol hydroxylase enzyme, leading mostly to cholestanol accumulation in various tissues [1]. Those depositions are preferentially located in the crystalline lens, tendons and brain, explaining the classical triad of symptoms consisting of juvenile cataracts, xanthomas and various neurological disabilities [2].

However, lung involvement has rarely been described as a presentation of this entity [3]. On the skin, this disease manifests mainly by tendon xanthomas. To our knowledge, the presentation of acquired ichthyosis has not been reported yet.

We report the case of a CTX with a lung involvement and an acquired ichthyosis.

Case Presentation

A 28-year-old man, born of a second-degree consanguineous marriage was admitted for nodular lesions over the extensors of the limbs, which has been gradually progressing for the past 8 years.

The history revealed the existence of a psychomotor retardation with delay in walking at the age of 5, recurrent chronic diarrhea since early childhood and at the age of 18, a gait difficulty and abnormal movements associated with dizziness. There was also a notion of cataract surgery in his sister.

The dermatological examination revealed firm, painless, mobile, confluent and yellowish nodules of xanthomatous aspect, located bilaterally over elbows, knees, Achilles’ tendons and over the 2nd, the 3rd, and the 4th fingers (Figure 1). There was also an ichthyosis, which predominantly affected the lower limbs (Figure 2). The neurological examination revealed a cerebellar ataxia, a pyramidal syndrome and a cognitive dysfunction. The ophthalmological examination showed crystalline opacities, suggestive of bilateral cortico-nuclear cataract. Standard laboratory investigations, among which serum cholesterol and triglycerides were normal. The tests for serum cholestenol, urinary bile alcohols and the research of a mutation in the CYP27A1 gene were not available in our hospitals. Toxoplasmosis and HIV serologies were negative. Histopathological examination of a skin nodule was consistent with typical xanthoma.

The chest x-ray showed alveolar opacities in the left upper lobe and a right apical infiltrate. Chest CT-scan showed bilateral apical opacities with cystic images in the culmen (Figure 3). Magnetic resonance imaging (MRI) of brain revealed bilateral and symmetric T2 and FLAIR hyper-signals sequences in the region of dentate nucleus and adjacent cerebellar and periventricular white matter (Figure 4). In the absence of chenodeoxycholic acid, oral ursodesoxycholic acid was...
Contingent lipid droplets of needle shaped clefts in septa and alveolar spaces has been reported in patients with CTX [3,6]. An increase in cholestanol level in the bronchoalveolar liquid has also been demonstrated [3]. As in our patient, those pulmonary lesions were often clinically asymptomatic [3]. The radiological images of the lung classically reported in the literature were as diffuse nodular, miliary, infiltrated opacities and pseudo-tumoral pleural opacities [3]. The findings in our patient were remarkable by its nodular appearance and excavated images simulating those of lung tuberculosis. The cystic aspect with gas-fluid level has been reported only in one case [1]. On the lung, lesions could be specifically secondary to the formation of foreign-body granulomas, explaining then these radiological images [1].

On the skin, manifestations of CTX are dominated by tendon xanthomas. The presentation of acquired ichthyosis, as noted in our case, has not yet been reported to our knowledge. In CTX, the ichthyosis could be related to the disorder of the lipid metabolism as it is in some congenital ichthyosis with cholesterol metabolism dysfunction [7].

The CTX is one among the rare treatable genetic conditions [8]. The poor prognosis of this condition has been improved since 1984 with the introduction of chenodeoxycholic acid [9]. In lack of this molecule, cholic acid, which is however less effective, has been also suggested [10].

**Conclusion**

We have reported the first case of CTX in Senegal, which is remarkable by an acquired ichthyosis and excavated lung lesions simulating tuberculosis.

**Conflict of Interests**

None

**References**