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DYSKINESIAS IN A PATIENT WITH CEREBROTENDINOUS XANTHOMATOSIS

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PRESENTATION

History: Male, 48 years old, seen at a neurology outpatient clinic, attendant reports total aphasia. Since childhood he refers to retarded neuropsychomotor development, besides low weight and seizures. Sister nas similar clinical picture.

PHYSICAL EXAMINATION

Presents global ataxia, does not assume orthostasis, axial component, dysmetria and decomposition of the upper limbs bilaterally, asymmetrical to the left. Extended arms maneuver: high amplitude and low frequency tremor, associated with ataxia. He has dyskinetic movement of head, trunk and limbs, axial predominance in the neck region and spontaneous nystagmus. He has nodules in joints, suggestive of tendinous xanthomas. Symmetrical reflexes bilaterally in upper limbs and exalted in bilateral patellar, bilateral flexor cutaneous-plantar reflex, unresectable glabellar.

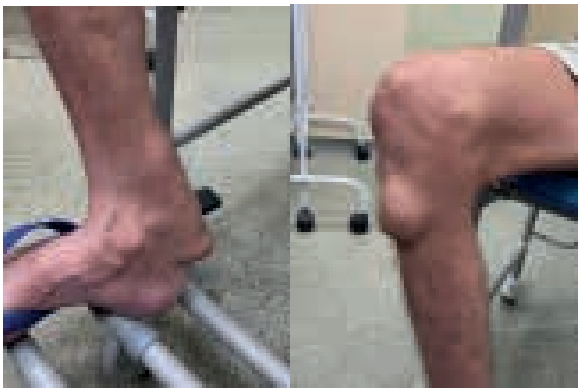

Figure 1. Photographs of the patient, showing expansive lesions.

COMPLEMENTARY EXAMS

Magnetic resonance imaging (MRI) of the skull showed intra-axial, non-expansive, bilateral, symmetrical lesions in the white matter of the cerebellar hemispheres, associated with hyperintense areas on T2/ FLAIR in brainstem and basal nuclei, suggestive of cerebrotendinous xanthomatosis (CTX) when associated with clinical. Patient does not use levodopa.

DISCUSSION

The (CTX) is a rare autosomal recessive disease caused by mutations in the CYP27A1 gene, leading to absence of the mitochondrial sterol 27-hydroxylase, responsible for cholesterol metabolization in the bile synthesis pathway (NIE E CHEN, et al, 2014). Thus, there is accumulation of cholesterol in tissues, including the brain, leading to progressive neurological dysfunction marked by dementia, epilepsy, hyperreflexia, spasticity, movement disorders such as parkinsonism, dyskinesias, and cerebellar ataxia. In other tissues it causes tendon xanthomas, atherosclerosis and diarrhea (Pilo-de-la-Fuente, Jimenez-Escrig, et al , 2011). Cerebellar ataxia and tremor are justified by lesions in the dentate nuclei, extending into the surrounding white matter of the cerebellar hemispheres. Trunk-members dyskinesia can be explained by neurodegenerative theory, neurotoxic mechanisms, such as the production of lipid peroxidation, with excess cholesterol in CTX and free radical formation, leading to a higher rate of dopamine neuron loss due to lipid accumulation in the substantia nigra (ZAND E LI, et al, 2021). Furthermore, CTX is a genetic disease, it can be associated with mutations in the CYP2D6, the dopamine D2 and D3 receptor genes, and the serotonin 2A and 2C receptor genes that cause dyskinesia (MA E REN, et al, 2021).

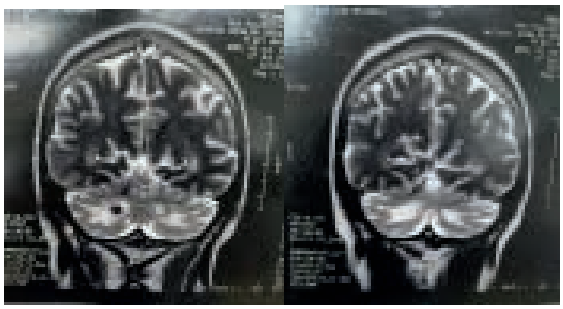

Figure 2. MRI imagens ou the patient in the case.

FINAL COMENTS

CTX with dyskinetic and ataxic movements that are unusual in the presentation of this disease.

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