P-149 - DESCRIPTION OF PHENOTYPE, CLINICAL PRESENTATION, NEUROIMAGING AND FOLLOW UP OF A SERIES OF PATIENTS WITH X-LINKED ADRENOLEUKODYSTROPHY

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INTRODUCTION: X-linked adrenoleukodystrophy (X-ALD) is the most common peroxisomal disorder and is caused by mutations of ABCD1(Xq28) gene. The dysfunction of the peroxisomal membrane protein codified leads to very-long-chain fatty acids (VLCFA) accumulation, which confirms diagnosis. The age of onset and morbidity are highly variable and progression is unpredictable. This disorder has many different phenotypes and has a characteristic neuroimaging. OBJECTIVE: To describe phenotype, initial symptoms, neuroimaging, laboratory, management and outcome of a series of patients with X-ADL. METHODS: Retrospective-prospective and descriptive study. Review of clinical records and Magnetic Resonance Imaging (MRI) of patients diagnosed with X-ADL who have been evaluated at the Neuropsychiatry Service of Hospital San Borja Arriaran since 1990. RESULTS: 28 male patients with elevated VLCFA in plasma: 16 with childhood-cerebral form, 4 with Addison disease only, 5 asymptomatic and 3 with adolescent-cerebral form. The average age of symptoms onset was 7.8 years old (3 to 16 years old). Initial clinical presentation: behavioral disorders: 13/28, cognitive impairments: 10/28, adrenal insufficiency: 6/28, hearing loss: 6/28, visual loss: 6/28, seizures: 6/28 and gait disturbance: 3/28. MRI showed enhanced T2-signal in the parieto-occipital region in 17/28 (only one patient asymptomatic), frontal region in 3/28 and normal in 8/28 (4 Addison disease only and 4 asymptomatic). Management: 14 patients received corticosteroid replacement therapy (CRT), 6 received Lorenzo’s oil, one patient had hematopoietic stem cell transplantation (HSCT) in an advanced disease stage with an unfavorable result and 5 patients received fludrocortisone. Thirdteen patients are alive: 4 with Addison disease only, 4 with progressive deterioration and 5 persist asymptomatic, in regular control with MRI. CONCLUSIONS: In our series of patients the most frequent form was the childhood-cerebral X-ADL and most of them initially presented with behavioral disorders and cognitive impairments, being the classic parieto-occipital pattern the most frequent neuroimaging alteration. Half of our patients needed CRT and the only patient who received HSCT died. Diagnostic suspicion must be high in patients that initiate cognitive impairment or behavioral disorders at school age associated with other subtle neurological symptoms, so to perform an early neuroimaging and study of VLCFA, and be able to offer specific treatment at an early stage.