



# ADRENAL INSUFFICIENCY DUE TO X-LINKED ADRENOLEUKODYSTROPHY DIAGNOSED IN LATE ADULTHOOD

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## INTRODUCTION

X-linked adrenoleukodystrophy (X-ALD) is a rare condition presented mainly in males during childhood and early adulthood. It represents almost 10% of primary adrenal insufficiency (PAI) or Addison's disease cases.

## CLINICAL CASE

A 67-year-old male was diagnosed at the age of 61 with PAI during testing for mesenteric panniculitis while admitted in Internal Medicine service. He was treated with hydrocortisone and referred to outpatient endocrinology clinic. The patient did not attend and restarted follow-up 6 years later.

His past medical history included dyslipidemia, anxiety and parkinson-like syndrome since 2012. He was taking hydrocortisone 20-5-5mg, with no mineralocorticoid. He had normal blood pressure with no significant changes between supine and orthostatic position. Blood tests showed normal levels of serum sodium, potassium and renin. Aldosterone was in the lower limit of normality (table 1). 21-hydroxylase antibodies were negative and the abdominal CT scan showed normal adrenal glands (Fig. 1). Due to the normality of these two tests, plasma levels of very long-chain fatty acids (VLCFA) were assessed; they were elevated (table 2).

Genetic study demonstrated a mutation in the ABCD1 gene: p.E292K(c.874G>A). Skin biopsy revealed high levels of VLCFA in fibroblasts and low levels of ALD- protein. MRI of the brain had no alterations in the white matter (Fig.2). DATscan exposed changes in the nigrostriatal pathway suggestive of Parkinson's disease. He satisfied the Addison-only phenotype.

The patient had a 24 year old son and a 30 year old daughter who wanted children. She presented high levels of plasma VLCFA and the genetic study revealed the same mutation as her father. She was referred to genetic counseling.

Table 1. Laboratory findings

	Value	Normal Values
Renin	7,5 uU/ml	4,2 - 59,7
Aldosterone	3,7 ng/dl	3,7 - 21
Sodium	140 mEq/L	136 - 145
Potassium	3,9 mEq/L	3,6 - 5,3

Table 2. Long chain fatty acids

	Value	Normal Values
Behemic acid (C22:0)	70,3 umol/L	51,1 - 113,4
Lignoceric acid (C24:0)	<b>109,5 umol/L</b>	44,3 - 92,4
Hexacosanoic acid (C26:0)	<b>4,7 umol/L</b>	0,22 - 0,88
Ratio sC24/C22	<b>1,56</b>	0,55 - 0,89
Ratio sC26/C22	<b>0,067</b>	0,004 - 0,021

Figure 1. Abdominal CT scan

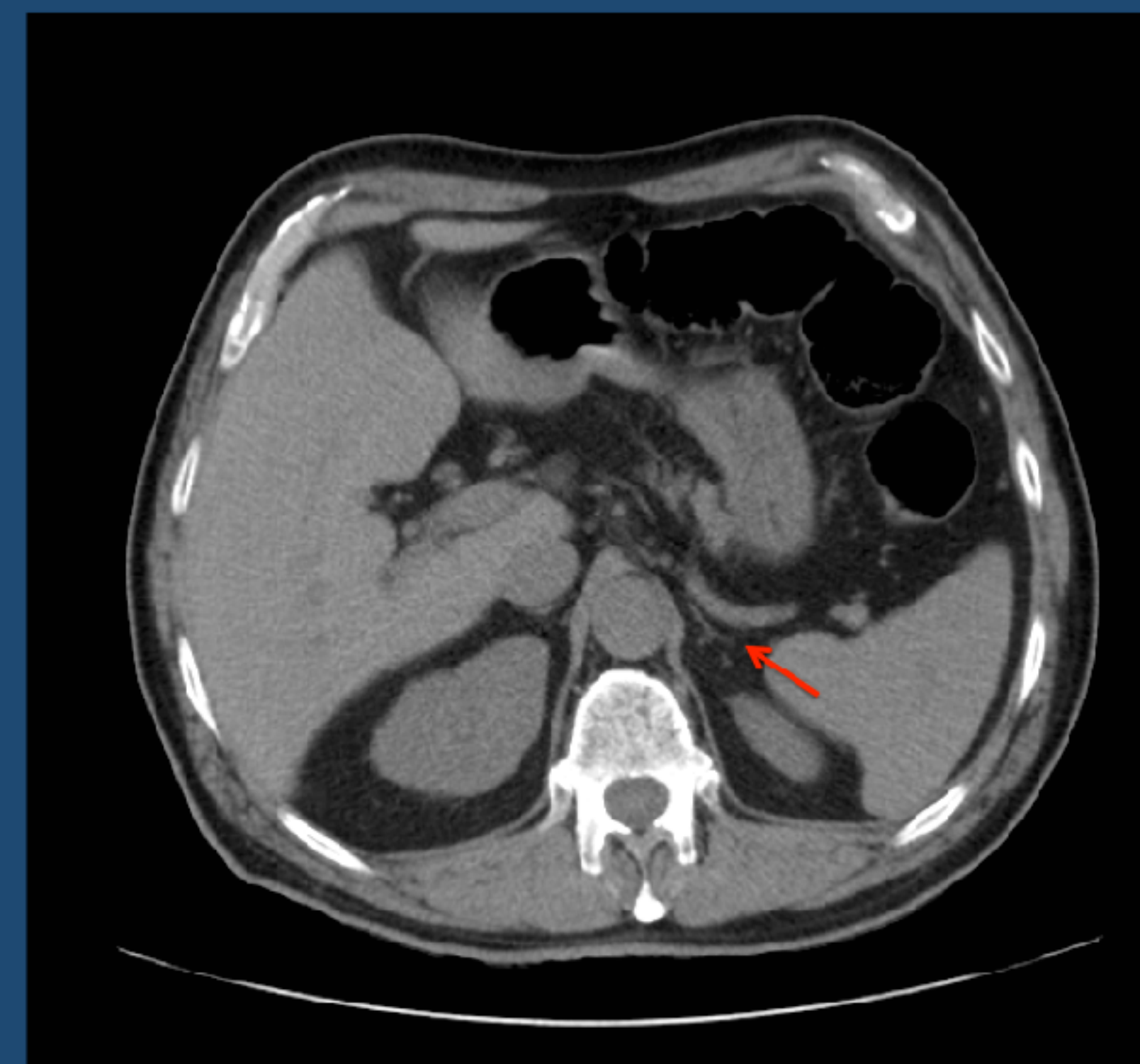


Figure 2. Brain MRI



## CONCLUSION

To consider X-ALD in males diagnosed with PAI at any age because of its implications, one very important being the transmission to offspring.

