Adrenoleukodystrophy (ALD) is a rare X-linked disease caused by a mutation of the peroxisomal ABCD1 gene. It is a progressive condition with a variable clinical spectrum that includes primary adrenal insufficiency, axonal demyelination and the accumulation of high levels of very long chain fatty acids (VLCFA) in the plasma and tissues.

**RESULTS**

- Six cases of ALD were included.
- The mean age at first symptoms of ALD was 4 years and 3 months old (Range: 16 days - 8 years old)
- Parental consanguinity was noted in one case and a family history of ALD was reported for 4 children.
- For four patients, X-linked ALD started as primary adrenal insufficiency, manifesting with skin hyperpigmentation and vomiting (Figure 1).
- For two patients, neurological signs were the first symptoms of the disease (Table 1).

**AIM**

The aim of this study was to describe the clinical, biological, radiological and genetic features of Adrenoleukodystrophy (ALD) in children.

**METHOD**

- We performed a retrospective study of all cases of X-linked ALD who were diagnosed in the pediatrics department of the university hospital of Sfax between 2004 and 2020.
- Specific data related to epidemiology, phenotype and diagnosis of patients with X-linked adrenoleukodystrophy were collected and analysed.

**CONCLUSIONS**

- Brain magnetic resonance imaging (MRI) showed signs of leukodystrophy in 2 cases (Figure 2).
- The plasma very long chain fatty acids (VLCFA) levels were significantly increased for five children (Table 2).
- Genetic testing identified the mutation of ABCD1 gene in 4 cases
- All children developed adrenal insufficiency during the course of the disease (Table 3) and 2 children progressed to a cerebral phenotype (Figure 3).

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