**Introduction**

Adrenoleukodystrophy is an X-linked, inherited metabolic disorder. Here, we present 3 clinical cases of different phenotypes with one mutation in ABCD1 gene in one family.

**Patient 1**

At the age of 9 years, manifestation of neurological symptoms was observed, skin color changed, these symptoms progressed monthly.

MRI of the brain: 13 points on Loes scale.

X-linked adrenoleukodystrophy was confirmed by biochemical (elevation of very long chain fatty acids - VLCFA) and molecular genetic studies (a mutation in exon 1 of the gene ABCD1 (c.871G>A (p.Glu291Lys) in the hemizygous state).

The child was diagnosed with X-linked adrenoleukodystrophy, childhood cerebral form, primary adrenal insufficiency, and hormone replacement therapy was prescribed.

Six months after the manifestation of the disease the child died.

**Patient 2**

At the age of 1 year 4 months the boy stopped sitting down, was constantly sleeping, was sluggish.

Molecular genetic study: an identical mutation in the ABCD1 gene was detected.

During the examination, primary adrenal insufficiency was diagnosed, hormone replacement therapy was prescribed.

Up to 4 years of age during dynamic observation there were no changes in the nervous system, according to MRI of the brain, there were no pathologies. Currently, the boy has X-linked adrenoleukodystrophy: Addison’s disease only.

**Patient 3**

14 years: At the time of the examination, there were no complaints. There have never been clinical signs of adrenal insufficiency and neurological symptoms.

The diagnosis of X-linked adrenoleukodystrophy was confirmed by elevation of VLCFA and the presence of a mutation in the ABCD1 gene, as in younger brothers.

During ACTH stimulation test (with cosyntropin), cortisol was increased up to 1173 nmol/l.

Sex hormones correspond to puberty stage (Tanner 3).

The boy may have an asymptomatic X-ALD phenotype.

**Conclusions**

There is no correlation between the genotype and the phenotype. In this family, the presence of 3 forms of the disease is noted: childhood cerebral form, Addison’s disease only and asymptomatic phenotype. However, progression of the disease is possible; patients require medical follow-up. Factors affecting the development of one form or another are currently unknown.