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Doronina A., Nadrah Mossa, Korovina O., Ovsyannikov D.

Tyrosinemia type 1: a case report

Peoples` Friendship University of Russia

Background. Tyrosinemia type 1(TT1) is an autosomal recessive disorder caused by deficiency of the enzyme fumarylacetoacetate hydrolase (FAH), which catabolizes fumyralacetoacetate into fumarate and acetoacetate [1]. The pathophysiology of the disease is explained by the subsequent accumulation of tyrosine and its metabolite succinylacetone in the liver, kidney and peripheral nerves, leading to dysfunction of these organs [1,2]. TT1 usually presents in early infancy with failure to thrive, vomiting, jaundice, hepatomegaly, elevated liver enzymes and bleeding tendency [3].

Case presentation. We report a case of a 2-month-old girl with tyrosinemia type 1 presented with dyspnea, fever, diarrea and hepatosplenomegalia. Examination revealed a sick-looking infant with signs of severe jaundice, hepatosplenomegaly, elevated liver enzymes and ProTime INR test. X-ray findings revealed a left-side pneumonia, echocardiographic findinds: separation of pericardium, ultrasound findings showed a severe liver fibrosis, hepatosplenomegalia. Tandem mass-spectrometry (TMS) revealed an increase of methionine (152,4 mikromoll), tyrosine (626,7mikromoll), valin (379,9 mikromoll), leucinisoleucine (693,2 mikromoll). Chromatography indicated a singnificant increase of succinylacetone (195 mMM). DNA analysis identified a homozygous mutation.

Conclusions. Despite the intensive therapy of Sol. Albumini, Verospironi, antibiotics, $O_{2,}$ Pulmikort, the exitus lethalis was diagnosed. This report demonstrates the value of screening for the early diagnosis of TT1 to prevent associated mortality.

References

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Key words: tyrosinemia, hepatosplenomegalia, succinylacetone

