

High Levels of Alpha Fetoprotein in the Neonatal Period: Which Metabolic Cause?

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Abstract

Patients in the newborn period with high level of Alpha fetoprotein (AF) can be secondary to some metabolic diseases like Citrin deficiency (CD), with cholestasis as the most important sign at this age. Another disease is Tyrosinemia type I (TYR 1), with no weight gain as the most frequent manifestation.

All of these two causes of increase level of AF can be treated, but CD has better prognosis, although TYR 1 under nitisinone (NTBC) therapy can control better the liver function and reduce the risk of liver cancer if we start the treatment before 2 year of age. The growth catch-up is good too with an earlier treatment.

Nowadays, with the neonatal screening program in several countries, we can see less cases of CD and TYR 1. But this doesn't mean that this will not appear in others countries, mainly in those with frequent cases of consanguineous marriage.

In a neonatal patient with high level of AF, we need to check amino acids in plasma, amino acids and organic acids in urine. Those two causes of increase AF levels can be practically excluded only with this study.

Keywords: *Alpha Fetoprotein; Neonatal; Metabolic Disorder*

Background

Alpha fetoprotein (AF) in the neonatal period can increase but will return to normal values with the evolution of the days. When these elevated levels persist and increased even more, there are two metabolic diseases that we need to rule out.

Citrin deficiency (CD), is an autosomal recessive metabolic disorder. Infants with CD can show intrahepatic cholestasis, low birth weight and growth retardation.

Blood tests will confirm the diagnosis with increased levels of direct bilirubin, alkaline phosphatase (AF) and G-Glutamyl transferase (GGT). AFP is very high, mostly more than 100000 ng/ml.

The plasma amino acids with the high levels of methionine, threonine, citrulline and threonine: serine ratio will give us important information about the possible cause of the patient jaundice. The confirmation of CD is with the determination of SLC25A13 gene.

Lactose free MCT rich formula is the main treatment and the prognosis is good in most cases, with normal growth and motor skills.

Tyrosinemia type I (TYR 1), is also an autosomal recessive disorder. The deficiency is on fumarylacetoacetase (FAH).

The symptoms include a history of failure to thrive, lost weight and later polyuria, polydipsia (secondary to Fanconi syndrome) delay motor skills. Later on will develop microcytic hypochromic anemia, hypoglycemia, hypophosphatemia, liver dysfunction and signs of rickets. The AF level is high but much lower than in citrin deficiency. Plasma amino acids revealed high levels of tyrosine, phenylalanine and methionine. In the urine, we can see generalized hyperaminoaciduria, succinylacetoacetic acid and succinyl acetone. The diagnosis can be determined by reduced activity of fumarylacetoacetase in the skin fibroblast or by FAH gene. The patients will have signs of rickets secondary to Fanconi syndrome. Treatment is with NTBC: 1 mg/kg/day, bid, with an amino acid based tyrosine and phenylalanine free powdered infant formula containing essential and non - essential amino acids, carbohydrate, fat, vitamins, minerals, trace elements with long chain polyunsaturated fatty acids and prebiotic fibres.

Conclusion

In the neonatal period, if we find high levels of AF, we need to exclude CD and TYR 1.

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