Case report: Maternal tyrosinemia type 1a under NTBC treatment with tyrosine- and phenylalanine restricted diet in Chile

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Abstract

We report the case of a 17-year-old girl with Tyrosinemia type 1a who carried a planned pregnancy to term while being under 2-(2-nitro-4-trifluoromethylbenzoyl)-1,3-cyclohexanedione (NTBC, nitisinone) treatment and a tyrosine- and phenylalanine-restricted diet. She was on treatment since 2 months of age with poor metabolic control prior to her pregnancy (tyrosine 838 +/- 106 umol/L). NTBC and a low tyrosine and phenylalanine diet were continued during her pregnancy. She unfortunately suffered from urinary tract infection and anemia during her pregnancy, with median plasma tyrosine and phenylalanine levels of 613 +/- 106 umol/L (200-400 umol/L) and 40.2 +/- 8 umol/L (35-90 umol/L), respectively. After 40 weeks of gestation, the patient gave birth to a healthy boy, with no adverse effects related to the use of NTBC. The newborn presented with a transitory elevation of plasma tyrosine levels and normal phenylalanine, methionine, and succinylacetone levels. By 12 months of age, the child was determined to have normal psychomotor development. At 20 months old, he was diagnosed with a mild developmental delay; however, global cognitive evaluation with the Wechsler Intelligence Scale for Children (WISC) test at 5 years old showed normal performance. Here, we discuss one of the few reported cases of nitisinone treatment during pregnancy and demonstrate a lack of teratogenicity and long-term cognitive disabilities.

Keywords

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