Tyrosinemia type I: long-term outcome in a patient treated with doses of NTBC lower than recommended

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Sir,

The paper by El-Karakasy et al. “NTBC therapy for tyrosinemia type 1: how much is enough?” raises the question if doses of nitisinone (NTBC) lower than recommended could be effective in the treatment of tyrosinemia type 1 [1]. The authors report that the duration of follow-up, which does not exceed 27 months, represents a limitation of their study. We follow a 15-year-old male patient with tyrosinemia type 1 diagnosed at 6 months of age and promptly enrolled in an NTBC trial [2]. He started treatment with the recommended average dose of 1 mg/kg/day divided in two doses. After 2 months of treatment, plasma and urinary succinylacetone were undetectable. An improvement in hepatic function was noted within 18 months and serum alpha-fetoprotein progressively decreased and normalized after 24 months. Before starting NTBC therapy, the hepatic ultrasonography showed multiple cirrhotic nodules that disappeared after 48 months of treatment. Our patient thrived well but corneal opacities occurred, when tyrosine levels exceeded 700 $\mu\text{mol/l}$ because of dietary noncompliance. After 114 months of treatment, based on clinical and biochemical follow-up, we progressively reduced the NTBC dose to the

actual dose of 0.55 mg/kg/day. The NTBC serum concentration always remained above the recommended level of 30 $\mu\text{mol/l}$ and urinary succinylacetone remained undetectable.

Until now, the patient is in good condition with normal growth and absence of biochemical and ultrasound evidence of liver injury. Our case suggests that doses of NTBC, lower than recommended, could be effective in the long-term outcome.

Conflict of interest None.

References

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