



Successful treatment of dumping syndrome in infant with Haddad syndrome

Authors: Jugovac Vita¹, Šuk Tuđman Franciska Dora¹, Kosanović Pia¹, Grizelj Ruza², Dumić Kubat Katja² (mentor)

¹ School of Medicine, University of Zagreb, Zagreb, Croatia

² Pediatric Department, The University hospital Centre Zagreb, Zagreb, Croatia

Background:

Haddad syndrome (HS) is a rare neurocristopathy caused by mutations in the PHOX2b gene. Two main symptoms include congenital central hypoventilation syndrome (CCHS) and Hirschsprung disease (HD). Impaired glucose tolerance is one of the rarely reported and possibly overlooked symptoms of HS.

Case presentation:

Our patient is a 4-month-old boy born at term from a second uncomplicated pregnancy from healthy, young, and unrelated parents. He was transferred to the newborn intensive care unit on the first day of birth due to frequent apnoea. Multiple unsuccessful attempts of extubation and signs of hypoventilation during sleep lead to diagnosis of CCHS. The boy also developed signs of HD in first weeks of life. Diagnosis of HS was confirmed by genetic testing. At the age of four months frequent asymptomatic hypoglycaemias were observed despite adequate peroral intake. The lowest measured blood glucose (BG) was 2.1 mmol/L, with insulin levels of 2.6 mU/L. Readings from continuous subcutaneous glucose monitoring revealed dumping syndrome pattern with hyperglycaemia in first hour, followed by hypoglycaemia in a second hour postprandially. Various combinations of milks and frequency of feedings were attempted to prevent dumping syndrome but none of the were successful. Finally, at the age of 6 months, to retard the digestion of carbohydrates in the small intestine, an alpha-glucosidase inhibitor (Acarbose) was introduced in treatment which led to normal glucose tolerance without side effects.

Conclusions:

Haddad syndrome is a rare condition that needs lifetime care of a multidisciplinary team. Given its rarity, it is usually diagnosed late. Impaired glucose tolerance is an overlooked symptom of unexplained aetiology that can easily be missed. Acarbose is safe, tolerable, and efficient in treatment of impaired glucose tolerance in patients with HS.

Keywords:

Alpha-glucosidase inhibitor, Congenital central hypoventilation syndrome, Haddad syndrome, Hirschsprung disease