

Repeated syncopes caused by sinus nodal arrest requiring pacemaker implantation in patient with late-onset familial form of congenital central hypoventilation syndrome

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Introduction. Congenital central hypoventilation syndrome (CCHS) is a rare genetic disorder resulting from mutations in the PHOX2B gene characterized by hypoventilation secondary to missing responses to both hypercapnia and hypoxia, that is thought to be caused by neural crest migration abnormalities. Baseline bradycardia, transient asystole or vasovagal syncope have been described in these patients. We present a rare case of girl with late-onset familial CCHS who experienced four syncopes caused by sinus nodal arrest with pauses of up to 20 requiring pacemaker implantation.

Patients and Methods. A 4-year-old girl, previously being hospitalised 5 times for respiratory failure from 6 weeks of age. She experienced two severe cyanotic attacks related to viral pneumonia at the age of 6 weeks and 4 months. At the age of one year, she was diagnosed with severe pulmonary hypertension resulting in acute right ventricle failure. Primary pulmonary, cardiac, neuromuscular and metabolic disease was excluded. Overnight videopolysomnography revealed non-apnoeic oxygen desaturation. Only then, CCHS became the highly probable cause, but this time the parents refused tracheostomy and mechanical ventilatory support. After further upper respiratory tract infection at the age of two years, she remained dependent on a ventilator while asleep.

At the age of three years and nine months, she experienced four syncopes caused by sinus nodal arrest with pauses of up to 20s. Since permanent epicardial pacemaker implantation was performed, no syncopes occurred during a further follow-up.

Afterwards, she failed extubation and conversion to overnight non-invasive ventilatory support after pneumonia. Tracheostomy was done, and she was discharged home using a portable positive pressure ventilator during sleep. Her father was retrospectively found out to suffer from severe headache and excessive daytime sleepiness.

Results. Molecular genetic evaluation of PHOX2B gene was performed in both girl and her father, and casual polyalanine repeat expansion mutation c.741_755dup15 in exon 3 was found in heterozygous form.

Conclusions. Patients with CCHS demonstrate affected autonomic function including heart rate and blood pressure control probably being caused by absence of normal maturation of carotic body and visceral sensory ganglia as a part of dysregulation of autonomic nervous system.