**Haddad Syndrome**

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**Introduction**

The aim of this report is to present the case of a 1.5-year-old girl with congenital central hypoventilation syndrome (CCHS-so-called Ondine’s curse) with Hirschsprung’s disease. These two diseases make Haddad Syndrome. The child was born at term, by natural delivery in good general condition -10 points in Apgar score .Post- delivery period was complicated with necessity to apply mechanical ventilation in an unclear mechanism .The child was developing shallow breathing with carbon dioxide retention.

**Case Description**

In the first phase of hospitalization attempts were made to extubate the patient and keep her on CPAP respiratory support or on spontaneous respiration with passive oxygen therapy. It was within a short period (several minutes to 2 days) that cumulation of carbon dioxide developed, and the child did not demonstrate respiratory effort. Re-intubation with mechanical ventilation was required. Based on the clinical picture the suspicion of congenital central hypoventilation syndrome was formulated .At the age of 2 months tracheotomy was performed. At the age of 2.5 months the child developed a sudden and massive infection with enterocolitis and paralytic ileus. Laparotomy was performed with production of ileostomy. On the grounds of intraoperative picture and histopathological examinations of tissue samples , collected during the surgery ,the diagnosis of total colonic aganglionosis was established -Hirschsprung’s Disease The child wasqualified on preliminary basis for aganglionic enterectomy and reconstruction of alimentary canal continuity within planned time .In diagnostic process metabolic disorders, SMA, myasthenia, myogenic and neurogenic causes were ruled out .Karyotype was female. Imaging procedures ( ultrasound, MRI scans of CNS, Echocardiography, and chest X-ray did not reveal any abnormalities which could be the cause of respiratory insufficiency. Based on the results of radiological investigations ( angio-CT scan and gastrointestinal motility scan) ,diagnosis of vascular ring-right subclavian artery was established .During her stay in hospital , results of genetic tests arrived. The 20/26 PHOX2B mutation confirmed congenital central hypoventilation syndrome.

**Discussion**

The 20/26 PHOX2B mutation is better and less symptomatic than the 20/27 mutation. The patient is in relatively good condition, breathing while active on her own via tracheostomy. During sleep she requiresmechanical ventilation. Phoniatric tracheotomy tube is used. The girl was qualified for home mechanical ventilation program and for home nutrition via Broviac catheter, handled by parents, who were trained to use the whole equipment.

The girl’s physical and psychomotoric development progresses and she has an excellent contact with peers .Even better quality of her life after decannulation and noninvasive ventilation is expected. Implanting the diaphragm pacemaker is one of possible solutions to treat her further allowing her to breathe by herself. The question is when is the best time to decanulate her and what type of noninvasive ventilation would be the best (mask or helmet etc). Another uncertainty is if or when diaphragm pacemaker should be used.