ABNORMAL LOCALISATION OF THE RADIAL SPOKE
HEAD PROTEIN 4 AS A CAUSE OF PRIMARY CILIARY
DYSKINESIA- CASE REPORT
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Primary ciliary dyskinesia (PCD) is a rare genetic heterogeneous disease with autosomal recessive inheritance. The estimated prevalence is 1:10 000 to 1:40 000 live-born. To date, more than 40 different genes where identified to be involved. The most common form of PCD is defects in the outer dynein arms and radical spokes. PCD is characterized by chronic upper and lower respiratory tract disease, infertility, neurological manifestations and abnormality in left-right asymmetry. The severity of disease varies between patients. When growing up, symptoms become more pronounced with the presence of destructive changes in the lungs, bronchiectasis, and the development of respiratory failure.

In this report we present a brother and sister, from young, healthy parents, who have recurrent respiratory symptoms and respiratory infections from infancy: chronic cough, recurrent bronchopneumonia and bronchitis. The first medical examinations, hospitalisations and laboratory testing for cause of recurrent infections were done in infancy. The laboratory findings were inconclusive. In May 2018., the boy, aged 11, is underfed (BMI 14.39kg/m2, 5. ct) with clinical problems of chronic cough, recurrent infections and frequent hospital admissions. The girl, aged 9, has the same disease, respectively.

Noninvasive ventilation (NIV), refers to the administration of ventilatory support without the use of intubation and is a widely used and efficient method for treating patients with chronic respiratory insufficiency. Recent studies show that the spectrum of patients who could be candidates for NIV may require further evaluation, because, if started as early as possible, patients could benefit from less complications and a better quality of life.

This retrospective study included 7 patients with chronic respiratory insufficiency due to neurological and respiratory disorders as part of a complex genetic disorder which were followed and treated at Srebrnjak Children’s Hospital and ultimately received NIV as the main therapy of choice for respiratory failure. Laboratory findings and clinical status, as well as incidence of respiratory symptoms and frequency of hospital admissions were analyzed.

All of our patients fulfilled diagnostic criteria for chronic respiratory failure (requiring daily oxygen support, having baseline SaO2 < 88% on room air or pCO2 > 50 with a normal pH value).

Of our 7 patients 2 (28.6%) were female and 5 (71.4%) were male. Only one patient had no confirmed genetic diagnosis while 6 (85.7%) had proven chromosomal aberrations or genetic mutations. The most frequently diagnosed respiratory difficulties were recurrent respiratory infections of the upper airways (N=7, 100%), lower airways with bronchial obstruction (N=5, 71.4%) and recurrent pneumonias (N=2, 73.4%).

Results show that all of the included patients had a better respiratory status (measured by pulse oxymetry and acid-base status) and a significant decrease in the frequency of respiratory infections was also observed.

We conclude that noninvasive ventilation is the therapy of choice for children with chronic respiratory insufficiency. We suggest that NIV should be started as early as possible after the initial diagnostic procedures were performed and the clinical and diagnostic criteria have been met. The indications for NIV should be further extended as to prevent severe complications in patients with chronic diseases that affect respiratory muscle strength and would also be beneficial for their quality of life.