the meantime, genetic testing proved homozygosity for the pathogenic variant of the SPAG1 gene.

The treatment of PCD is challenging because of low evidence-based recommendations. However, this case report shows that with regular monitoring and therapy adjustments depending on the circumstances, it is possible to maintain the airways state and pulmonary function at a satisfactory level even when the profound chronic lung changes exist.

416 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, and auscultator findings of crackles. We done the laboratory examination and tried to found out the underlining cause. The laboratory findings reveal no increase in inflammatory tests, negative microbiological samples, with the exclusion of humoral and cellular immunodeficiency's, sarcoidosis, aspergillosis, tuberculosis, heart disease and gastroesophageal reflux. We observed allergic sensitization, obstructive changes in pulmonary function test and bronchial hyperreactivity. High resolution computed tomography (HRTC) of the chest showed bronchiectasis in the lower lobe of the left and middle lobe of the right lung. Pulmonary function tests demonstrated obstructive ventilation functional impairment. Diagnostic bronchoscopy with mucosal sampling was performed. The immunofluorescence test showed RSPH9 abnormal localization in the ciliary axonemes. This finding confirms the diagnosis of PCD. At the same time, we follow up the boy's younger sister, at the age of 8, with the similar clinical symptoms. Pulmonary function tests showed obstructive-restriction functional impairment, while the HRCT scan of the chest revealed atelectasis and bronchiectasis in the segments of the lower pulmonary lobe to the right and V segments to the left. According to the PID diagnosis in older brother, we presumed that girl has the same disease, respectively.

Primary ciliary dyskinesia is a rare disorder with no etiologic treatment.

Early diagnosis is important for introduction of early pulmonary rehabilitation and management of exacerbations in order to preserve the pulmonary parenchyma for as long as possible

417 THE EFFECT OF NIV ON THE CLINICAL OUTCOME IN SEVERAL DIFFERENT PROFILES OF PEDIATRIC PATIENTS WITH CHRONIC RESPIRATORY FAILURE

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Noninvasive ventilation (NIV), refers to the administration of ventilatory support without the use of intubation andis 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 respiratry failure. Laboratory findings and clinical status,as well as incidence of respiratory symptoms and freaquency of hospital admissions were analized.

All of our patients fulfilled diagnostic criteria for chronic respiratory failure (requiring dayly 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 aberations 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 signifficant 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.

418 CYSTIC ADENOMATOID LUNG MALFORMATION – CAUSE OF RECURRENT PNEUMONIA

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A ten-year-old boy was referred to the Clinic for further treatment after frequent bronchopneumonia in the last 4 years. Each time on the X-ray, basal right, the shadow on the lungs was verified. After antibiotic therapy, the control X-ray always showed incomplete regression of the shadow. The boy was born with esophageal atresia with tracheoesophageal fistula and was successfully operated on the eighth day of life.

Post-operative dilatation of esophagus has been successfully performed twice. The boy was in a good general health condition, except he was often tired after the activity. With the auscultation of the lungs, basal right, crepitation could be heard. CT of the chest, in the area of the posterobasal segment of the right lower lung lobe, showed a cluster of cystic air forms with condensed pulmonary parenchyma. The described change corresponded to developmental lung anomaly – cystic adenomatoid lung malformation. Right thoracotomy and lobectomy of the right lower lung lobe were performed. By surgery, the boy had no recurrent bronchopneumonia. Approximately one-third of CPAMs are diagnosed after the neonatal period. These lesions typically are CPAM types 1, 2 or 4, and tend to be smaller than CPAMs that present with respiratory symptoms at birth. A common presentation in older children is recurrent pneumonia. Other presenting complaints include cough, dyspnea and/or cyanosis. Findings on physical examination include decreased breath sounds over the lesion, hyperresonance and chest wall asymmetry with a bulge on the affected side.

419 SUCCESSFUL TREATMENT OF PRIMARY SPONTANEOUS PNEUMOTHORAX WITH VIDEO-ASSISTED THORACOSCOPIC PLEURECTOMY

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Pneumothorax is defined as a collection of air that is located between the visceral and parietal pleura. Spontaneous pneumothorax occurs in the absence of any identified trauma. It is subdivided into primary and secondary types. The annual incidence of primary spontaneous pneumothorax (PSP) in the general population is estimated to be 5 to 10 per 100000. The peak incidence occurs between 16 and 24 years of age. We present the case of a sixteen-year-old boy who manifested on several occasions with right-sided spontaneous pneumothorax. The boy was actively involved in sports. Until spontaneous pneumothorax occurred, he had never been ill. He did not take medication. He had no allergies. A total of three times the right chest was drained. At each arrival, pneumothorax manifested with abrupt pain. The X-ray confirmed the diagnosis each time. After a third spontaneous pneumothorax, a CT of the chest was done. CT showed small apical blebs. The trachea was neatly positioned, neatly branching. The right main bronchus seemed wider than the left. The vascular structures were of a neat appearance. The heart was neatly positioned, with a morphologically neat appearance. Surgical treatment was initiated. After collapse of the right lung, an incision was made in the third intercostal space to the right in the anterior axillary line. An angle camera was installed. Upon placement of the camera, the trocar is placed in the seventh intercostal space in the median axillary line. After identifying the apical blebs changes with a linear endostapler, a resection is performed. A partial parietal pleurectomy is then performed. Negative pressure drainage is set. In prophylaxis, the boy received cefazolin. A one-year follow-up did not result in relapse.

Surgery for pneumothorax consists of stapling ruptured blebs and resection of abnormal lung tissue. The approaches used include VATS (video-assisted thoracoscopic surgery), mini-thoracotomy and conventional thoracotomy. We usually use VATS, which provides adequate exposure for resection or stapling and an opportunity for pleurectomy, abrasion or chemical pleurodesis. The morbidity of VATS is less than with conventional or mini-thoracotomy, and recurrence rates are approximately 5%, although open thoracotomy and pleurectomy have the lowest recurrence rate. Given that in our previous cases, after repeated tube drainage, we did not have recurrences, we did not opt for VATS immediately after the first recurrence. Reading the literature, in the future we will be guided by the algorithm to initiate VATS after the first recurrence of pneumothorax.

420 INTERSTITIAL LUNG DISEASE DUE TO SFTPC MUTATION – A RARE CASE REPORT

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Interstitial Lung Disease due to SFTPC mutation -a rare case report Mutations of the surfactant protein (SP)-C gene SFTPC have been associated with neonatal respiratory distress syndrome and interstitial lung disease.

If accurate diagnosis and proper management are delayed, irreversible respiratory failure may occur. A girl born following dizygotic twin pregnancy born at 37+3 weeks, born in good condition, no active resuscitation. However noted to have a fast-respiratory rate. Subsequently has had several admissions to hospital with faltering growth, persistent tachypnoea and oxygen requirement. High resolution CT angiogram showed atelectatic changes in the dependant portions, genetics work up identified a variation in a gene called SFTPC.

Introduction Pulmonary surfactant is a complex mixture of phospholipids and proteins that reduces surface tension of the alveolus by forming a surface-active film at the air-liquid interface. Of the four surfactant associated proteins the hydrophobic SP-B and ASP-C are especially essential in the dynamics of phospholipids by promoting the transition between the storage form and the functional surface film.

SP-C is synthesized from a 197 –amino acid precursor (proSP-C)which is encoded by SFTPC on human chromosome 8p21.3. In this report we present a case of a child who has childhood interstitial lung disease due to SFTPC mutation (c.289G>T).

Child Details A 5-week-old initially presented with possible aspiration; she was noted to be persistently tachypneic (up to 90 breaths/min) needing minimal oxygen to maintain saturations. She was reviewed weekly, where she remained tachypneic associated with feeding issues and faltering growth, was extensively investigated (GI/cardiac/metabolic). Had a prolonged stay at hospital and was subsequently discharged home on 0.5L O2, high calorie NJ feeds and anti-reflux medications (post barium study).

Over the course she has had several admissions to the hospital, she has been treated with methylprednisolone 6 doses,