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PRIMARY CILIARY DYSKINESIA IN ADULTS – A TALE OF TWO CASES OF KARTAGENER SYNDROME

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INTRODUCTION: Kartagener syndrome (KS), the most serious form of primary ciliary dyskinesia (PCD), is a rare genetic disease occurring in both males and females, with an estimated incidence of 1 in every 32,000 live births. KS accounts for approximately half of PCD cases and is defined by a triad of bronchiectasis, sinusitis and complete situs inversus. We herein report the unique clinical features of KS in two young adults.
CASE PRESENTATION: Case 1 – A 32-year-old Caucasian female with KS, diagnosed at four years of age, was recently followed-up in the ambulatory respiratory care clinic. She is a current smoker (six cigarettes a day, three pack years). At three years old, the patient began having respiratory symptoms consisting of recurrent sinusitis and purulent bronchitis. She was only four when she underwent a right lower lobectomy for the associated bronchiectasis. Frequent respiratory infections requiring antibiotic treatment have plagued her since childhood. She has severe allergic asthma (i.e., sensitivity to house dust mites) for which she consistently uses inhaled corticosteroids and bronchodilators. The pulmonary function tests show stable, though moderately severe, restrictive and obstructive features. A nasal polypectomy is currently being planned. Case 2 – A 19-year-old Caucasian male with KS, diagnosed at birth, was seen a few months ago for follow-up. He is a never-smoker. This young man has been hospitalized several times for chest drainage of recurrent pneumothorax of the left lung. His chronic sinusitis with polyps was surgically managed. He has non-allergic mild asthma. The pulmonary function tests have now normalized, however in recent years, they have been much worse and variable. For the past two

years, he has been consistently taking antibiotics and bronchodilators, and his cough has diminished. His respiratory cachexia has improved significantly over the previous year with nutritional drink supplements.
DISCUSSION: Both patients described above have been closely followed by pulmonologists since being diagnosed with KS. The importance of early recognition of KS cannot be overemphasized. Treatment of this rare congenital disorder is often personalized, but generally includes antibiotics (intravenous or oral, intermittent or continuous) to treat respiratory infections or for prophylaxis, inhaled bronchodilators for obstructive lung disease, mucolytics, and chest physiotherapy.
CONCLUSIONS: This report serves to: 1) raise awareness about KS in adults, 2) remind clinicians that this condition must not be missed in adults without a previous diagnosis of KS in childhood, and 3) illuminate the coexistence of different pulmonary diseases with KS.