KARTAGENER SISTERS AND CILIARY DYSKINESIA HERITAGE

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ABSTRACT: Primary Ciliary Dyskinesia is characterized by structural and motility abnormalities of cilia, two sisters who had history of chronic respiratory infections during childhood were studied. Both patients had situs inversus, chronic sinusitis and bronchiectasis. In both cases was performed radiological studies that confirmed the presence of Kartagener syndrome.

KEY WORDS: Primary Ciliary Dyskinesia, Kartagener Syndrome, Bronchiectasis, chronic sinusitis.

INTRODUCTION: Kartagener syndrome also known as Primary Ciliary Dyskinesia (PCD) is an autosomal recessive defect characterized by structural and motility abnormalities of cilia, where the ciliary movement is dyskinetic, ineffective or absence of cilia which leads to an alteration in the mechanism of clearance of mucus in both upper and lower airways, which manifests as a clinical disease of sinus-lung and gonadal tissue, among others 1. It consists of a clinical triad characterized by situs inversus, chronic sinusitis and bronchiectasis. The role of imaging methods is valuable to characterize this condition with adequate clinical suspicion. We present the case of two patients with primary ciliary dyskinesia (PCD) identified in the outpatient services of Isidro Ayora Hospital, and we make a review of the literature focusing on the imaging diagnostic as a valuable tool for the characterization of this pathology.

CASE REPORT

Two sisters patients of female sex between 18 to 21 years old, with a history of recurrent infections of respiratory tract in childhood with a relief period during adolescence and reactivation from approximately about 5 years, were diagnosed with asthma and referred to the Imaging Department for complementary studies.

MATERIALS

It was performed tomographies of the paranasal sinuses and thorax with a Siemens Somatome Emotion 16 slices where multiplanar reconstructions were performed and in high-resolution for thorax.
DISCUSSION

The PCD, as mentioned in the literature, was initially known by Siewert, who associated this chart to bronchiectasis, sinusitis and situs inversus was Kartagener, but only Afzelius described it as dysmotility of cilia. It is an autosomal recessive disorder deduced by the Laws of Mendel that our patients have the autosomal defect, suspecting that one parent has the homozygous recessive gene defect while the other is single recessive carrier of the defect; although it is unknown which parent carries the disease. The clinical presentation of this entity is characterized by productive cough, respiratory tract infections, sinusitis, otitis media and infertility, affects both sexes with a prevalence of 1 in 32,000 births. For confirmation of the PCD was used the study of the frequency and pattern-shaped whipped cilia by video digital high resolution and high speed and electron microscopy biopsy of nasal mucus or bronchial for the study of ciliary ultrastructure, however CT provides the diagnostic with a sensitivity and specificity greater than 90.

CT values extension and morphology of bronchiectasis, it can define if there is total or partial situs inversus and observe lung structural damage. In the case of our patients, they had a childhood with chronic respiratory symptoms and allergic treatments without effective response but no tests were done due to the lack of clinical suspicion. In paranasal sinuses tomography (PNS) we observe mucosal thickening of both maxillary antra with obstruction of drainage pipes and agenesis of the frontal sinuses (Fig. 1 and 2), this is consistent with what mentioned S. Ochoa-Linares in whose publication emphasizes this anatomical variant as the most common in these patients.

FIGURE 1. 21 years old woman with mucosal thickening of the maxillary antra, the arrow shows the clogging of drainage pipes

FIGURE 2. Sagittal reconstruction of a CT in the 21 years old patient where the arrow shows the absence of frontal sinuses.
In thorax tomography was identified cylindrical bronchiectasis in both cases being more prominent in the patient of 18 years old, in a paracardiac and bilateral distribution (Fig. 3 and 4).

**FIGURE 3.** Thorax HRCT of the 21 years old patient where it shows bronchiectasis for right hilar and the aorta with location inverted (arrow).

**FIGURE 4.** 18 years old patient with more cylindrical bronchiectasis than her older sister, seen on a coronal CT reconstruction.

There is situs inversus of the thorax and abdomen viscera, which creates a mirror image, allowing compliance the diagnostic criteria for this condition. (Fig. 5A and 5B)

**Figure 5A.** Coronal CT reconstruction of 18 years old patient where it is observed situs inversus totalis.

**Figure 5B.** CT axial section of the 21 years old patient where it is observed complete reversal of the location of the abdominal viscera.

The clinical diagnosis of DCP can be challenging for its unspecific symptoms, which is why it should conduct a thorough differential diagnosis with other pathologies with the same clinic so we can make an accurate diagnosis and the treatment could be immediately applied, thus avoiding complications and marked development of this entity. In the differential diagnosis to consider it is stated cystic fibrosis disease with similar symptoms, furthermore Young Syndrome also share similar clinical and radiological findings to
primary ciliary dyskinesia and cystic fibrosis, however the underlying pathogenesis is not yet completely elucidated, it is characterized by obstructive azoospermia in epididymis level and is thought to be the cause of infertility. The clinical triad is obstructive azoospermia, bronchiectasis and sinusitis. Another entity to perform a differential diagnosis is Allergic Bronchopulmonary Aspergillosis found only in patients with long-standing asthma, occasionally in patients with cystic fibrosis. Only rarely, it occurs in patients with any other identifiable lung disease. While the imaging diagnosis of DCP is relatively simple, it could lead to difficulties when the doctor examining a patient with a history of recurrent pulmonary infections, chronic bronchial infection and rhinitis, does not have in mind the possibility that there is a Kartagener syndrome. In this regard, is very clear the premise that mentions, "You cannot see what the head does not know".

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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