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Kartagener syndrome at the stage of cor pulmonale: a case report

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Abstract

Kartagener syndrome is highlighted by a triad of symptoms that encompassed bronchiectasis, chronic sinusitis, and situs inversus. It is a rare disease which is characterized by primary ciliary dyskinesia. Also, Kartagener syndrome is defined as genetic disorder along with autosomal recessive inheritance. We report a case of Kartagener syndrome among 30-year-old man. The diagnostic was make on clinical and paraclinical evidences. A cough associated with chronic bronchorrhea for about 10 years, a exertional desaturation, and signs of right heart failure marked the clinical picture of the patient. The sinus X ray -(Blondeau incidence)- and the chest-abdominal CT scan confirmed: the situs inversus, the sinusitis, as well as the bronchiectasis. Pulmonary arterial hypertension and right cavities dilatation have been observed on Doppler ultrasound. The patient has benefited of daily respiratory physiotherapy, empiric antibiotherapy based on amoxicillin clavulanic acid for 10 days, oxygen therapy, and bronchodilators. The patient's condition remained stationary with persistence of the exertional desaturation. Kartagener syndrome requires early diagnosis in order to limit the disease progression to chronic respiratory failure.

Keywords: *kartagener syndrome, bronchiectasis, chronic sinusitis, situs inversus*

Introduction

Kartagener syndrome (KS) is characterized by the clinical triad of bronchiectasis, situs inversus and chronic sinusitis. The estimate incidence of this rare disease is approximately 1/32000 births[1,2].

Kartagener syndrome is associated along with primary ciliary dyskinesia (PCD) or "immotile cilia syndrome" in 50% of cases [1, 2].

Primary ciliary dyskinesia is a rare genetic disorder with autosomal recessive inheritance. Its prevalence is estimate 1/15000 [3].

The main underlying mechanisms of this desease are functional and ultrastructural organization abnormality of cilia, leads to organs damage among which respiratory system, middle ear and nose.

In Madagascar the prevalence of this disease remains unknown.

In the present paper we report a rare case of kartagener syndrome at the stage of cor pulmonal.

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Observation

A 30-year-old man, married for 7 years and had no children, has been hospitalized in the Pneumology department of Antananarivo University Hospital for a cough evolving over ten years, marked by a recent worsening and occurrence of dyspnea on exertion.

His medical history revealed otitis episode during childhood. His parents are in apparent healthy and It is no repported any concept of consanguinity.

On clinical examination, his saturation was of 81% while breathing ambient air, fever at 38.3°C. Respiratory system exam revel a bilateral basal crackles. Furthermore it is noted a digital clubbing.

Cardiovascular examination revealed lower legs edema, jugular veins turgescence, apical beat, and heartbeat perceived in right side, with no extra cardiac noises.

Other system examination area was normal.

Furthermore, the laboratory investigations has evinced an inflammatory syndrome described through C reactive protein levels increase at 145 mg / l, leukocytosis at 12 G / L into a blood count with 70% of neutrophil.

Blood sugar levels, renal and liver function status were normal. The sputum investigation for acid-fast bacilli and HIV serology were negative.

Chest X-ray has showed a dextrocardia and bilateral hilo-basal interstitial opacities (figure 1).

Sinus radiography into Blondeau incidence squeezed out thickening sinus maxillary (figure 2).

The chest-abdominal CT (figure 3) found a situs inversus as well as bronchiectasis. Ultrasound Doppler heart has allowed discovering right atrium and right ventricular dilatation, in contrast a normal left ventricular ejection fraction and pulmonary arterial pressures of 40 mmHg.

However, our patient has refused the seminogram, and cilia examination has not been carried out fault of technical support center.

Diagnosis of Kartagener syndrome of chronic pulmonary heart stage was evoked considering the clinical triad (situs inversus, bronchi dilatation, as well as sinusitis) and pulmonary arterial hypertension along with right heart cavities dilatation.

Patient has therefore received a respiratory physiotherapy daily, probabilistic antibiotic therapy such as amoxicillin clavulanic acid for ten days and an oxygen therapy.

Moreover, the following-up was emphasized through the desaturation persistence on exertion.

Discussion

In 1904, Siewert [4] described a case of situs inversus coupled with the bronchiectasis. However, the link between these two signs could not establish.

In 1935, Kartagener the Swiss doctor [5], was the first who identify a clinical syndrome combining a sinusitis chronic and bronchiectasis as well as situs inversus total along with family cases.

Thus, these signs were grouped under the name of "triad of Kartagener."

Afterwards, other signs such nasal polyposis, otitis and agenesis frontal sinuses extended this triad, and the term of"Kartagener syndrome" was given.

However, in half of the cases, Kartagener syndrome is associated with primary ciliary dyskinesia [1,2] which is a heterogeneous genetic disease with autosomal recessive inheritance, involving respiratory diseases related with the constitutional ciliary anomaly [3].

Some authors [6] have nevertheless reported X-related forms.

Primary ciliary dyskinesia and Kartagener syndrome lead repeated upper and lower respiratory tract infections because of mucociliary transport alteration and symptoms onset are often from childhood [7].

This abnormality can also defect sperm's flagella and lead to primary infertility. This is due to similarity between the ciliary structural and flagella.

Afzelius [8] was the first one who describe the relationship between Kartagener syndrome and male infertility by reporting this syndrome cases and lack of dynein arms in the spermatozoa and cilia in electron microscopy. That is the case of our patient who presented a primary infertility.

Clinically, pulmonary involvement is early.
This is overall a daily cough, bronchial congestion with exacerbation stages phases. Pulmonary symptoms are often the signs of discovery of this disease. Indeed it they reported by several authors [9-12].
Our case presents a chronic cough with chronic bronchorrhea. Thoes symptoms are the signs of discovery of Kartagener syndrome.
Bacteriological, respiratory secretions analysis among patients with primary ciliary dyskinesia and Kartagener syndrome proved that the bacteria in respiratory tracts are abundant and varied [13]. Pathogens that grow in culture are frequently Haemophilus influenzae, Staphylococcus aureus and Streptococcus pneumonia, Pseudomonas aeruginosa and non-tuberculous mycobacteria.
Radiologically, bronchiectasis appears early and is observed in 60-70% of cases at the time of diagnosis [14]. Imaging (abdomen-chest and sinus CT scan) confirms the classic triad of Kartagener syndrome substantiating the presence of bronchiectasis, sinusitis and laterality defect.
All of these abnormalities were found out in our patient. However, At this time there is no available ciliary dyskinesia standard diagnostic reference test [15]. According to the recommendation of the European respiratory society (ERS) of 2017 [16], the current diagnosis requires a combination of technically demanding investigations, sucha nasal nitric oxide testing, analysis of the frequency and rhythm ciliary beats using video-microscopic analysis at high-speed, transmission electron microscopy, genotyping and immunofluorescence. About our case, we do not have a capacity to achieve these all specifics investigations owing to inadequate of technical platform.
Concerning therapeutic regimen of the PCD /KS there was no etiological treatment yet, it remains symptomatic.
In 2009, the ERS [17] has developed a recommendation on management of the disease. However it highlighting that the recommendation is of low level of evidence.
Treatment consists in one hand on performing daily respiratory physiotherapy in order to drain bronchial secretions and to restrict the infection risk.
In the other hand an antibiotic therapy is recomended in case of infectious episode (ear, nose, and throat [ENT] or respiratory).
Antibiotic therapy should target thee germs commonly encountered [13] and shose regardind the antibiogram results.
Due to bronchial obstruction encountered during the bronchiectasis, the inhaled bronchodilator could be therefore used to improve respiratory symptoms.
A lobectomy is indicated in some cases of severe and localized bronchiectasis.
Furthermore, bilateral pulmonary transplantation is a necessary option for end-stage respiratory failure. Successfully surgery cases [18,19] have been reported. In the ENT area, nasal hygiene measures are very important.They must be done carrying out a saline nasal wash with effective blowing.
In fact, surgical sinus treatments may be considered in some rare cases for reduce secretions.
Vaccination against influenza and pneumococcal is however, in all cases is recommended.
The evolution of the disease is variable and often slow.
The acute evolution is marked by outbreaks of exacerbations with recurrent infections.
Moreover, in case of advanced and extended bronchiectasis disease may progress to the chronic respiratory failure, occurrence of pulmonary arterial hypertension and impact on the right heart.
It is unfortunately at this stage that our patient has been diagnosed.
Shas A et al [20] reported that the majority of adults with primary ciliary dyskinesia were diagnosed late along with impaired forced expiratory volume in one second (FEV(1)) and incremented colonization with Pseudomonas aeruginosa.

However, if diagnosis is make precociously and patient well treated, -as the case of 12-years-old boy reported by Sanjay Gupta et al. [21]-, evolution will favorable.
Hence the needing of early diagnosis, suitable treatment of symptoms, as well as monitoring of respiratory function so as not to reach the stage of chronic respiratory failure.

Conclusion

Kartagener syndrome and primary ciliary dyskinesia are rare heterogeneous genetic diseases. Diagnosis is based on clinical evidences and the implementation of specific investigations.
The clinical picture is dominated through respiratory and ENT symptoms.
Current treatment is always symptomatic.
Early diagnosis and appropriate treatment consists a guarantor for normal life to patient with the disease.

Conflict of interest:

The author(s) declare(s) no conflict of interest

Figure 1: Chest X-ray showed dextrocardia and hilo-basal infiltration opacities

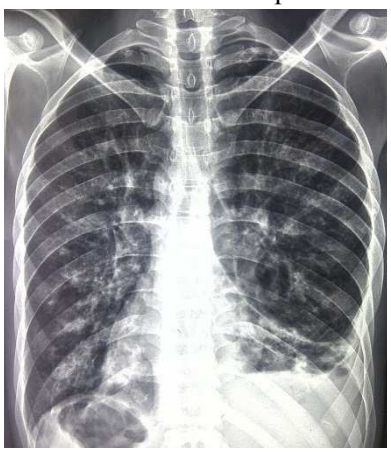


Figure 2: X-ray of the sinus -incidence of Blondeau- showed thickening of the maxillary sinuses

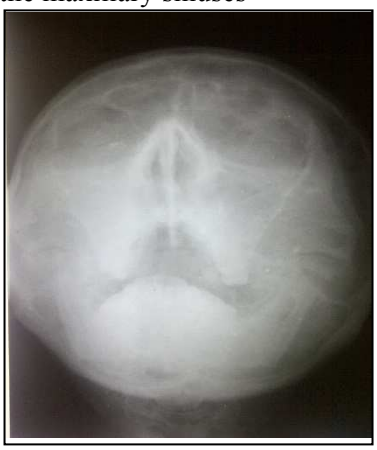
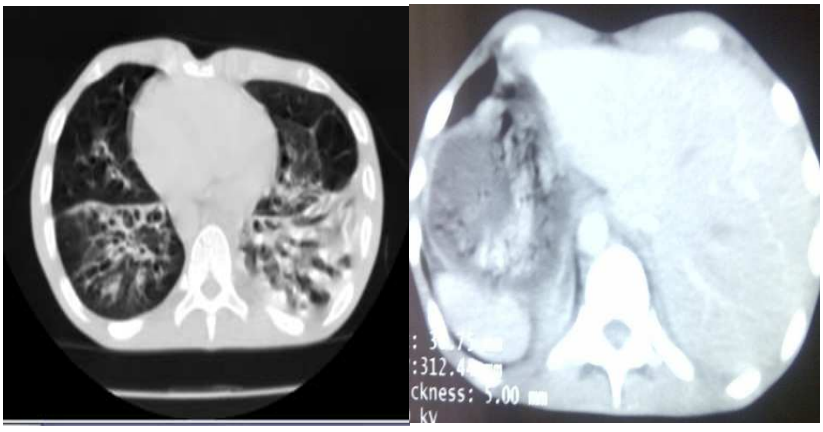


Figure 3: : Abdominal chest CT scan showed bronchectasis and situs inversus



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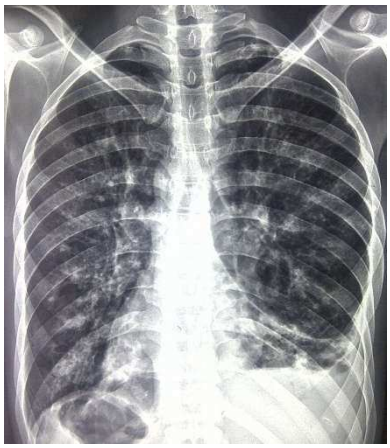
FIGURES



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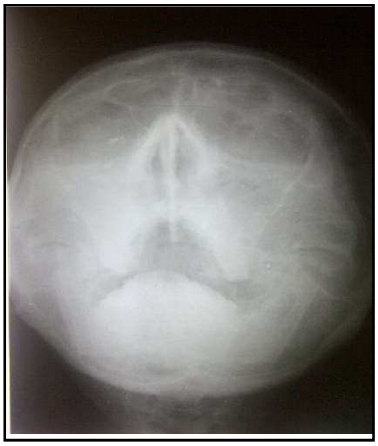

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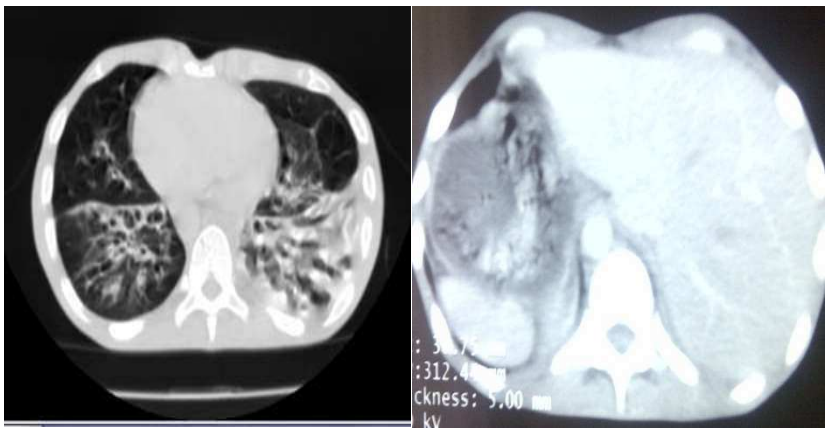

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