Primary ciliary dyskinesia (Kartagener syndrome) in a 38-year-old Egyptian male: A rare case

Motaz Badr Abdellatif Ibrahim

ABSTRACT

Introduction: Kartagener syndrome is a rare, ciliopathic, autosomal recessive genetic disorder with an estimated incidence of about 1 case per 32,000 live births. It always causes a triad which is situs inversus, chronic sinusitis and bronchiectasis.

Case Report: We present a case of a 38-year-old male patient complaining of productive cough and hemoptysis since 15 days, headaches since one month and easily fatigability two years ago. He suffered from similar complains during childhood with episodic fever, worsening of symptoms and he was wrongly diagnosed with tuberculosis. After 28 years, he complicated by severe empyema leading to surgical intervention. Laboratory workup was unremarkable except for leukocytosis ($>12,000/\text{mm}^3$) and hypoxia, semen analysis showed defective motility (asthenozoospermia) with grade c while other parameters were found normal including sperm count (35 million per milliliter), morphology and volume, chest X-ray and computed tomography (CT) scan were done, and he was diagnosed with Kartagener syndrome. He took medical treatment in the form of antibiotics (augmantin, 40 mg/kg/day PO), antipyretics (ibuprofen, 1 tablet PO q8h), mucolytics (guaifenesin, 100–400 mg PO q4h), inhaled bronchodilators (albuterol/ipratropium, nebulizer solution: 3 mL inhaled q6h) and a regular follow-up is done with chest X-ray.

Conclusion: In this case report, we intend to remind physicians by Kartagener syndrome despite being rare because early diagnosis is the key for those patients to avoid complications and to give those patients a better lifestyle, another conclusion is that our case is a fertile male which is uncommon in such syndrome.

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CASE REPORT

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Keywords: Kartagener syndrome, Bronchiectasis, Primary ciliary dyskinesia

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INTRODUCTION

Siewert described primary ciliary dyskinesia as a combination of situs inversus, chronic sinusitis, and bronchiectasis in 1904 [1]. However, the clinical triad of Kartagener syndrome was recognized by Manes Kartagener [1] in 1933 and that is why it bears his name until now. Kartagener syndrome is an autosomal recessive pattern in which its symptoms and signs are due to defective cilia motility. The frequency of Kartagener syndrome is 1 case per 32,000 live births.

CASE REPORT

A 38-year-old male presented to our hospital with a few days' history of chest pain, productive cough with yellowish sputum and significant shortness of breath. He retired two years ago from his job because of his easy fatigability and persistent productive cough forced him to do so. He denied any history of smoking. No family history of such a case in his first degree relatives.

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The condition started since he was 10-year-old with recurrent pneumonia and bronchiectasis in which he was wrongly diagnosed with tuberculosis so the doctor gave him antituberculous drugs (a 6-month course of isoniazid (INH) and rifampin, supplemented during the first two months with pyrazinamide) and antibiotics (augmentin) after which he was sent home after relieving.

After 28 years, he suffered from worsening of the symptoms and hemoptysis in which he was admitted to the hospital and diagnosed with severe empyema in which he underwent surgical decortication on the right side and application of chest tube for drainage to treat his severe empyema.

On general examination, it was found that his vital signs were: respiratory rate 28/min, blood pressure 110/70 mmHg, and temperature 37.4°C. Chest examination revealed on inspection limited chest movement more on the right side, apical pulsations on the right fifth intercostals space in the midclavicular line no any other pulsations noticed. By palpation a tracheal shift to the right side was found with a tactile vocal fremitus more on the right side. By auscultation diffuse ronchi, diminished air movement in both lungs and heart sounds were more pronounced at the right sternal border. Other physical examination findings were unremarkable.

Laboratory workup was unremarkable except for leukocytosis ($>12,000$ per mm^3) and hypoxia, semen analysis showed defective motility (asthenozoospermia) with grade c (non-progressive motility because they do not move forward despite the fact that they move their tails. Sometimes also denoted motility II), other parameters were found normal including sperm count (35 million per milliliter), morphology and volume.

The patient is fertile with three children by the help of assisted reproductive techniques.

Chest X-ray and computed tomography (CT) scan revealed a shifted mediastinum to the right side; the right lung field is totally replaced by cystic bronchiectatic changes showing thickened bronchial wall, hyperinflated left lung, dextrocardia and the stomach was normally on the left side due to the presence of a stomach bubble on X-ray which is confirmed by CT scan of abdomen (Figures 1 and 2).

The clinical diagnosis of Kartagener syndrome was made and the condition was explained to the patient and he was treated with antibiotics (augmentin, 40 mg/kg/day PO), antipyretics (ibuprofen, 1 tablet PO q8h), mucolytics (guaifenesin, 100–400 mg PO q4h), inhaled bronchodilators (albuterol/ipratropium, nebulizer solution: 3 mL inhaled q6h) and a regular follow-up is done with chest X-rays.

The follow-up showed a good response to the medical treatment and the patient went back to his job and he is having a better lifestyle.

Figure 1: Chest X-ray revealed a shifted mediastinum to the right side, old bronchiectatic areas at the bases of the right lung, hyperinflated left lung and dextrocardia.

Figure 2: Computed tomography scan of chest revealed a shifted mediastinum to the right side, the right lung field is totally replaced by cystic bronchiectatic changes showing thickened bronchial wall; hyperinflated left lung, scattered areas of consolidative changes in the left lung, dilated pulmonary artery (3–4 cm) and dextrocardia.

DISCUSSION

Kartagener syndrome is a primary ciliary motility disorder in which the majority of the patients present with situs inversus, chronic sinusitis and bronchiectasis caused by pseudomonal infection [2, 3].

When taking a history, the patient mainly complains of chronic cough with unexplained respiratory distress and he/she always describes this cough as wet and productive, found in nearly all of infants [4]. In addition to the defective drainage of the sinonasal system in those patients, this will lead to congestion, rhinorrhea, and chronic middle ear effusions with suppurative otorrhea. Imaging for those patients will reveal situs inversus abnormalities which are relatively specific for Kartagener syndrome [5].

The initial diagnostic workup is started after proper history taking and physical examination.

In Kartagener syndrome, postpubescent males will have abnormal sperm motility (asthenozoospermia) which will be an obstacle in the future for those patients if they want children. However, this problem is solved by the new techniques of assisted reproductive techniques nowadays as in our case.

Despite the newly multiple diagnostic tests that emerged there is no full standard test to diagnose this syndrome and as an example of those tests:

1. Electron microscopy, which visualizes the ciliary ultrastructure. The samples are obtained by nasal scrape or brush biopsy.
2. High-speed videomicroscopy is used in some research centers for a better observation of the ciliary beats [5].
3. Nasal nitric oxide measurement is measurable in nasal air of normal subjects and found to be low in cystic fibrosis and very low in primary ciliary dyskinesia. Recently, it was suggested to play an important role in regulating ciliary motility.
4. The immunofluorescence diagnosis of primary ciliary dyskinesia, various components (proteins) of cilia structure is stained by specific antibodies. The cross section of a normal respiratory tract cilium shows the typical anatomy of the axoneme, which consists of nine peripheral microtubule doublets that surround two centrally located single microtubules (9+2 structure). The peripheral microtubule doublets are interconnected with each other by inner and outer dynein arms and to the central microtubules by radially oriented spokes. The outer and inner dynein arms are protein complexes of various dynein chains and associated proteins. A mutation in one of the genes that encodes a component of the dynein arms often results in a total loss of dynein arm structure.

In addition to the previously mentioned diagnostic tests, there is a large expansion into the field of genetic testing and isolation of Kartagener syndrome mutations.

Multiple new genes were discovered related to this syndrome based on the theory which states the existence of additional ciliary mutations which do not manifest in those patients. This is why future genetic testing will be one of the main diagnostic tools in diagnosing those patients [6].

The treatment regimen for those patients includes: antibiotics with good pseudomonal coverage, daily chest physiotherapy. Dnase, hypertonic saline and acetylcysteine may be tried in patients with recurrent infections and respiratory symptoms [7]. Surgical intervention for those patients can be beneficial in complicated bronchiectasis and when the disease is localized as in our case [8].

CONCLUSION

Kartagener syndrome must be in the differential diagnostic list of conditions leading to bronchiectasis in pediatric age group including (foreign body, tumor, lymphadenopathy, chronic obstructive pulmonary disease (COPD), immunodeficiency states with recurrent infections, IgG and IgA deficiencies, alpha-1antitrypsin deficiency, recurrent aspiration pneumonia, inhalation of poisonous dust and fumes) despite being rare because early diagnosis is the key for those patients to avoid complications and to give those patients a better lifestyle, another conclusion from our case is that our patient is a fertile male which is uncommon in such syndrome.

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Author Contributions

Motaz Badr Abdellatif Ibrahim – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

Guarantor

The corresponding author is the guarantor of submission.

Conflict of Interest

Authors declare no conflict of interest.

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