FT73

A Case Of Primary Ciliary Dyskinesia And The İmportance Of Anatomical Side Markers İn Direct Radiography

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ABSTRACT

OBJECTIVE:

If situs inversus is seen on chest x-ray and the patient's clinic is compatible with primary ciliary dyskinesia (PSD), it should be examined in more detail and screened for siblings in case of PSD. In addition, we aimed to state that even if the directional markers are not used correctly on the chest radiography, even an obvious condition such as situs inversus may be omitted or radiographs may be misinterpreted by the physician.

CASE:

Twelve years old girl with primary ciliary dyskinesia was evaluated. The presence of recurrent sinopulmonary infection and laterality defect on chest x-ray led to the investigation of PSD and the diagnosis of PSD was confirmed by homozygous variant in CCDC39 gene.

CONCLUSION:

The literature review shows that some of the radiographs have incorrect or no anatomical direction markers. In our case, if the radiography contains an incomplete directional marker, it may lead to delayed diagnosis and wrong treatment for the patient. Screening of siblings for a genetic disease such as PSD will provide many benefits.

ÖZET

AMAÇ:

Akciğer grafisinde situs inversus görülüyorsa ve hastanın kliniği PSD ile uyumluysa, PSD ayrıntılı olarak incelenmeli ve bu hastalık tespit edilirse kardeşler de taranmalıdır. Ayrıca, direk grafilerdeki yön belirteçleri doğru kullanılmazsa situs inversus gibi bariz bir durumun bile gözden kaçabileceğini veya radyografilerin hekim tarafından yanlış yorumlanabileceğini belirtmeyi amaçladık.

OLGU:

On iki yaşında PSD'li bir kız hasta değerlendirildi. Akciğer grafisinde tekrarlayan sinopulmoner enfeksiyon ve lateralite defekti varlığı PSD'nin araştırılmasına neden oldu ve PSD tanısı CCDC39 genindeki homozigot varyant ile doğrulandı.

SONUÇ:

Literatür taraması, bazı radyografilerde yanlış anatomik yön belirteçleri olduğunu veya hiç olmadığını göstermektedir. Bizim vakamızda, eğer radyografi eksik bir yön belirteci içerirse hasta için gecikmiş tanı ve yanlış tedaviye yol açabileceği açıklanmıştır. Kardeşlerin de PSD gibi genetik bir hastalık için taranması birçok fayda sağlayacaktır.

***Keywords**: **Ciliary Motility Disorders**, Genetic Diseases, Congenital Abnormalities,*

Anatomical side markers

INTRODUCTION:

Primary ciliary dyskinesia (PSD) is also known as kartagener syndrome and immotile cilia syndrome. This syndrome is a hereditary disease which characterized by impaired cilia function and leads to various clinical manifestations such as chronic sinopulmonary disease, middle ear effusions, infertility, and laterality defects. Situs inversus is a rare congenital anomaly characterized by transposition of abdominal organs, internal organs and vessels. Situs inversus occurs in approximately 50% of patients with PSD. In the first approach to the patient with situs inversus is likely to be detected by chest x-ray (1,2). Physicians should pay attention to side marker and x-ray type (anterior or posterior) and interpret them appropriately (3).

We present a patient with primary ciliary dyskinesia who diagnosed at the age of twelve. We emphasized that the patient had recurrent sinopulmonary disease and the laterality defect on the chest radiography was the first to be considered in this disease, and that the radiography could be misinterpreted by the physician if the directional markers were not used correctly. We would like to point out that even a very cautionary condition such as situs inversus may be omitted if we do not use these markers.

CASE:

Twelve years old female patient was admitted to our hospital with fever and cough for 2 days. The patient had a history of recurrent lung infection and was first diagnosed to have pneumonia at the age of one month, but after the treatment her complaints continued to increase. The patient was brought to our hospital because of fever, vomiting and cough at the age of 2 months. For further investigation she was hospitalized in the pediatric chest diseases department and then chest x-ray and echocardiography was performed. After her first hospitalization, he had 12 more hospitalizations for lung infection. She also frequently used antibiotics for recurrent sinusitis and otitis. There was no pathology related to birth of the patient and no kinship between her parents. Her siblings had no known disease. In physicial examination, patient's height and weight were between 3-10 percentile. Respiratory system examination revealed diffuse rhonchi in both lungs and rales in some areas. Chest x-ray showed infiltration areas and dextrocardia was observed (figure 1).

Primary ciliary dyskinesia was considered with her clinic and history, also occurence of situs inversus totalis. A homozygous variant was found in the CCDC39 gene and the diagnosis of primary ciliary dyskinesia was confirmed. Genetic analysis was performed in the siblings of our case and no primary ciliary dyskinesia was detected.

DISCUSSION

The mutation in any protein in the structure or function of the ciliary mechanism may theoretically cause disease. PSD is a genetically heterogeneous disease involving many genes. PSD has an autosomal recessive inheritance and some cases is shown with X-linked inheritance in the literature. (1). There is no gold standard diagnostic method for this syndrome, the recommended diagnostic criteria is the history of chronic bronchial infection and rhinitis in early childhood with one or more of the following characteristics: (a) patient or sibling with situs inversus or dextrocardia, (b) detection of viable but inactive spermatozoa (c) impaired or no tracheobronchial clearance (4).

Reading the chest radiography can be very challenging even for specialists. It is recognized worldwide that all radiographic images should have an accurate anatomic side marker. It is

important to define the image orientation and position in the view in order to evaluate a direct radiography with the clinic. Anatomical side markers are defined in the image as descriptions of "right" or "left" side (5). Malpractice potential is high in cases where the the wrong side is marked. Anatomical side markers on chest radiographs should be correctly labeled to avoid potentially harmful consequences for the patient especially in cases of dextrocardia. The importance of accurate radiographic anatomical side markers should not be underestimated.
When we look at the literature on anatomic side markers, Barry et al. Found that 5.8% of the 400 images had an incorrect or incomplete anatomic side marker (3). In the study of Platt et al., 1% of the images had no anatomical markers (6). There are lots of studies which studied anatomical side markers error and its consequences.
In our case, if the radiography contains an incomplete directional marker, it may lead to delayed diagnosis and wrong treatment for the patient. It is very beneficial for individuals and public health that physicians are familiar with the clinic of PSD and siblings should be screened as soon as possible for a genetic disease such as PSD.

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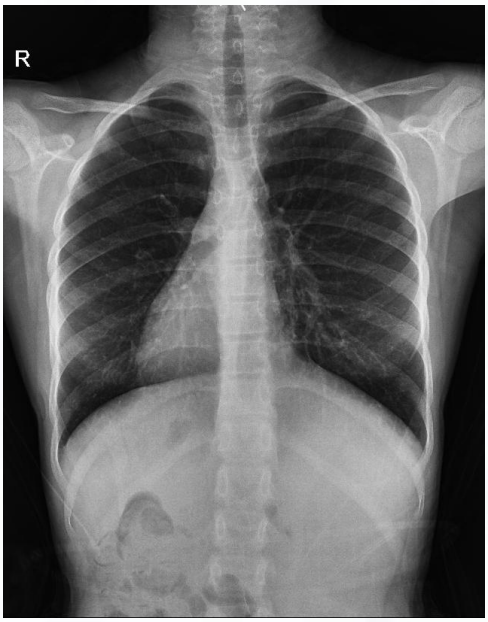

Figure 1: Chest radiography shows dextrocardia.