International Journal of Biomedical and Advance Research
ISSN: 2229-3809 (Online); 2455-0558 (Print)
Journal DOI: https://doi.org/10.7439/ijbar
CODEN: IJBABN

Case Report

Shrouded co morbidity of severe osteoporosis in a young female with Kartagener-Afzelius syndrome - An unrivalled case report

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***Article History:**
Received: 04/07/2018
Revised: 13/07/2018
Accepted: 13/07/2018
DOI: https://doi.org/10.7439/ijbar.v9i7.4834

Abstract

A 23 year old young female presented with recurrent chest infection since childhood with joint pain, diagnosed as a rare congenital autosomal recessive case of Kartagener syndrome with severe osteoporosis.

Keywords: Primary ciliary Dyskinesia, Kartagener syndrome, Dextrocardia, Situs inversus.

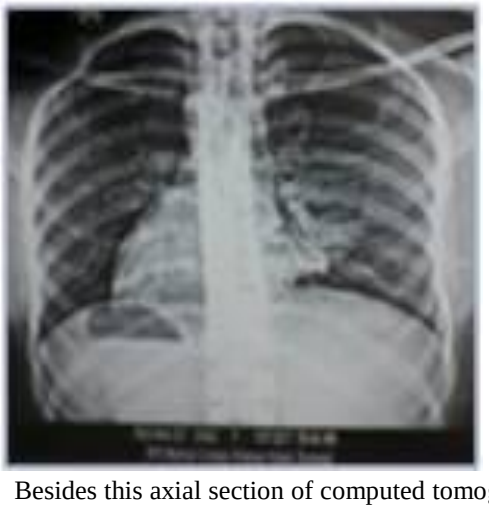
1. Introduction

Kartagner syndrome is an autosomal recessive disorder as well as rare subgroup of primary ciliary dyskinesia (PCD) occurring with frequency of 1:30,000 to 1:40,000. Although Siewert first described this condition in 1904, Kartagener just recognized the etiological correlation between the elements of the triad and reported four cases in 1933. [1] In the 1970s, Bjorn Afzelius, reported cilia immobility in infertile males. In which half of the cases had Kartagener's triad since then it is also called as Kartagener-Afzelius syndrome. [2] There is no specific predominance of disease among male or females. To till date approximately 30 genes are known to be associated with (primary ciliary dyskinesia) PCD, accounting for > 60% of PCD cases. [3] During the embryonic stage, organ position is determined by uniform ciliary beating but in KS, due to ciliary dysmotility heart along with the other organs fail to move on to the left side, resulting in dextrocardia and situs inversus. Kartagener's syndrome may be either situs solitus attributed only dextrocardia or situs inversus totalis where all the viscera are on the opposite side. [1] The young female in this report had rare situs inversus totalis with extremely unusual combination of sever osteoporosis, not reported hitherto.

2. Case Study

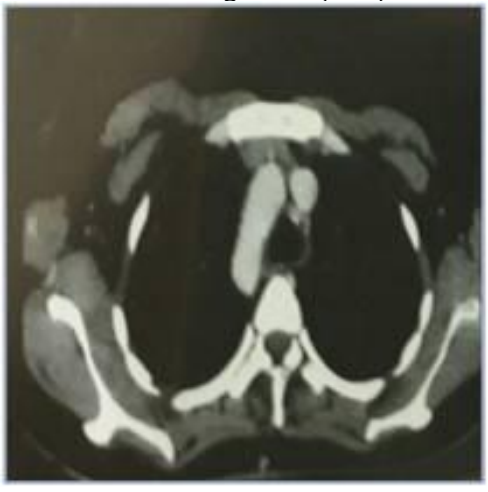
We are reporting a case of 23 years old female with anosmia, rhinorrhoea, cough with expectoration and joint pain since childhood. Symptoms used to get aggravated during seasonal change and she has to take treatment on and off. Upon investigation of chest skiagram shows dextroposition of cardiac shill haut (Figure 1)

Figure 1: CXR in PA view. Situs inversus totalis



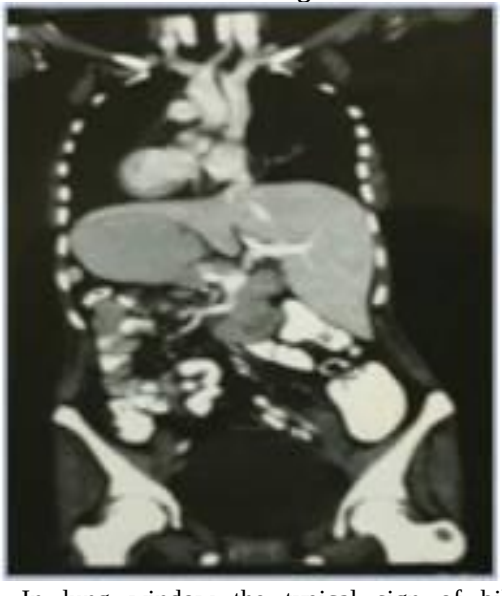
Besides this axial section of computed tomography of thorax reveal right sided aortic arch (Figure 2).

Figure 2: CECT Thorax, Axial Section showing Arch of Aorta on right side (MW)



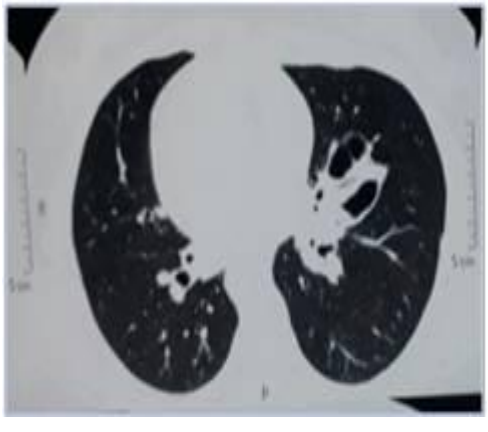
In coronal section of thorax and abdomen it is clear that entire viscera along with liver reciprocate over opposite side suggestive of situs inversus totalis (Figure 3).

Figure 3: Coronal section showing thoracic and abdominal organs



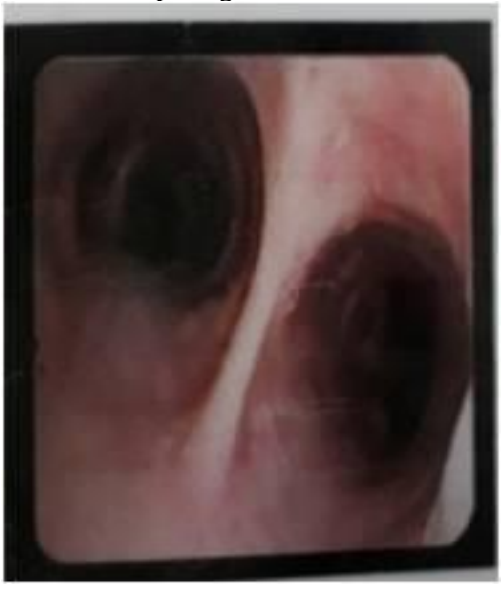
In lung window the typical sign of bilateral bronchiectatic changes appear with left sided predominance (Figure 4).

Figure 4: Axial section of CECT thorax showing central bronchiectatic changes (Lung window)



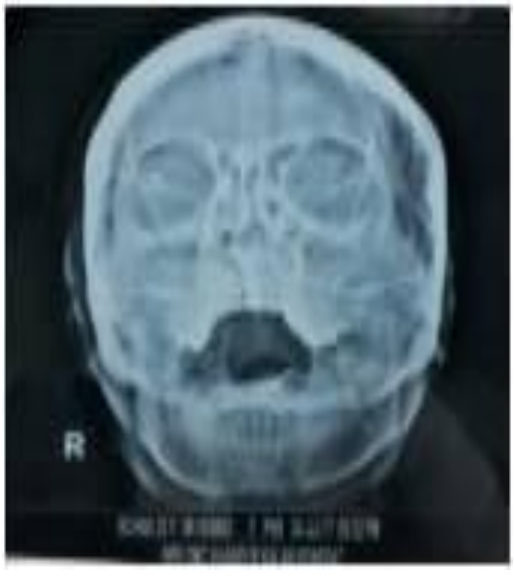
On further workup with fiberoptic video bronchoscopy it was reveal that anatomical opening of tracheobronchial tree also transposed (Figure 5).

Figure 5: Bronchoscopic view showing upper lobe opening in left side



PNS view also reveals maxillary haziness suggestive of chronic sinusitis (Figure 6).

Figure 6: XRAY PNS – maxillary sinus Haziness



Bone mineral density as well as Serum uric acid and ASO titer, RA factor was also estimated due to severe body ache, and severe osteoporotic changes revealed in the report at early age. Her bone mineral density report revealed mean T-score -3.9% (-43%) and Z-score had density of -3.9% (-44%) suggestive of severe osteoporosis. Rests of the investigations were found in the range. In addition to above said symptoms our patient was also experiencing erythmatous lesions on skin and hearing impairment. Her menstrual cycles were normal.

3. Discussion

Upper respiratory tract infections may be caused by variety of disease i.e. bronchiectasis, rhinitis, rhinobronchitis, hypoproteinemia, COPD, foreign bodies, etc. but to come to diagnosis of KS need intensive exploration of investigation at treating facility, among all Kartagener-Afzelius syndrome is one of the rarest congenital disorder associated with recurrent infection due to ciliary dyskinesia.[4] Diagnostic criteria for this condition include clinical picture suggestive of recurrent chest infections, bronchiectasis, and rhinitis since childhood, along with one or more of the following: (1) situs inversus in the patient/sibling; (2) alive but immotile spermatozoa; (3) reduced or absent transbronchial mucociliary clearance; and (4) cilia showing characteristic ultrastructural defect on electron microscopy. Apart from fulfilling the criteria mentioned above, two types of tests are done for diagnosis of PCD – screening tests (exhaled nasal nitric oxide measurement which is usually low in PCD, and saccharin test to assess mucociliary function of nasal epithelium) and diagnostic tests (ciliary beat pattern and frequency analysis using video recording, and electron microscopic confirmation of the ultrastructural ciliary defect).[5] Initially, the term immotile cilia syndrome was used to describe this disorder; however, later studies showed that most cilia were motile, but exhibited a stiff, uncoordinated, and/or ineffective beat hence name was changed to “PCD”.[6]

Ciliopathy term given to collection of disorders related to impaired ciliary fuctions. There are two types of cilia viz primary (sensory) and motor cilia.[7] There is another class of cilia known as Nodal cilia which appears during the embryonic development stage of gastrula. Motor cilia are complex structure consisting of α and β subunits. These are separated from cytoplasm by gate or diffusion barrier constituting ciliary gate function. Primary cilia differ from motile cilia as it sense changes in external environment, essential for normal development, tissue differentiation. Due to this unique ubiquitous nature any defect in primary ciliary function responsible for not just a disease but variety of disease under the umbrella term as primary ciliary dyskinesia, attributed to impairment of ciliary function , exhibiting numerous syndromes such as Bardet-Bidel syndrome, retinitis pigmentosa, skeletal dysplasias.[8,9] Its been seen that nearly half of the primary ciliary dyskinesia patients have heterotaxy i.e. situs inversus totalis which leads to congenital heart ailments(40%), polyspleenia.[10] Any kind of impairment in ciliary function leads to excessive mucous production leads to colonization of bacteria later serving as nidus for secondary infections.[11] Alias movement of cilia is vital for organs such as respiratory tract, female reproductive tract, sperm flagella, while impairment in ciliary function leads to infertility in later stages of life, repeated respiratory tract infections and chronic sinusitis. Cilia rotation induces a leftward flow to the extra embryonic fluid. This flow may concentrate on the left side, or deplete on the right side, the critical factors that start the molecular cascade needed for normal lateralization. If the flow is not present, the factors are equally distributed and the lateralization is randomized May also be associated with situs inverses.[2] Apart from this cilia line the upper and lower respiratory tracts and are considered to be the first line of defense against harmful bacterial infections.[12] In present case report patient had recurrent upper respiratory tract infection since childhood. On workup by using advance diagnostic modalities like dexa scan, her bone mineral density surprisingly revealed early severe osteoporotic changes which are quite unusual to observe at young age of hers as compare to same gender. In recent study conducted by Mirra *et al*[13] on 22 PCD patients reported that Hypovitaminosis D is common in PCD patients besides this Vitamin D deficiency-to-insufficiency and sufficiency groups did not show any differences in age at PCD diagnosis which is responsible for recurrent infection though they have not commented on its association with severe osteoporosis[14]. However it is not possible yet to establish whether Kartagener Syndrome might play a pivotal role in the pathogenesis of osteoporosis, though it need more detailed comprehensive research in future to draw conclusion for early screening of osteoporosis as an integral part of management in Kartagener syndrome for better outcome.

4. Conclusion

Respiratory tract infection is a major complication of Kartagener syndrome though possibility of osteoporosis in early age can’t be overlooked to avoid associated complications. Hypovitaminosis D associated with Kartagener syndrome not merely increase the possibility of recurrent infection rather it may also culprit to reduce the density of bones may lead to pathological fracture in affected subject.

Recommendation

Bone density assessment in young female patient with Kartagener syndrome must be considered to avoid future complication of vertebral fracture.

Acknowledgement: Nil

Contributors Detail

Corresponding author responsible for the concept, design, manuscript preparation and statistical analysis, second author contributed in collecting literature, reviewing article and editing, rest of the author’s contribution in collecting data, compiling, searching review of literature.

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