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KARTAGENER'S SYNDROME- A DELAYED DIAGNOSIS DURING PREGNANCY

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Abstract: Kartagener's Syndrome (KS) is a group of ciliary motility disorders called primary ciliary dyskinesias (PCDs). It is a genetic condition with an autosomal recessive inheritance, with a triad of Sinusitis, Bronchiectasis and Situs Inversus. The case presented here proves that females with KS also can successfully conceive and carry pregnancy to term. However, they are likely to have respiratory complications during the course of their pregnancy. These complications eventually will subside post pregnancy and hence the patient has to be managed to reduce respiratory complications.

INTRODUCTION

Kartagener's Syndrome (KS) is a group of ciliary motility disorders called primary ciliary dyskinesias (PCDs). It is a genetic condition with an autosomal recessive inheritance, with a triad of Sinusitis, Bronchiectasis and Situs Inversus.[1] It was first described by Siewart in 1904, It was Kartagener who recognized therelation between the elements of the triad and reported 4 cases in 1933[2] The estimated prevalence of PCD is about 1 in 30,000,[3] PCD ranges from 1 in 12,500 to 1 in 50,000.[1] In KS, the genetic defect leads to impaired ciliary motility which causes recurrent chest infections, sinus infectionsand infertility.

Early diagnosis treatment options may be given to manage infertility in these youngsters. Although not proven, early diagnosis can guide in preserving and maintaining of pulmonary function there by improving the quality of life, and life expectancy in these patients.[4,5] However, further large prospective studies are needed to confirm this.

CASE STUDY

This was a 23-year-old pregnant female (G2A1), born to consanguineous parents. She presented to the Outpatient Department referred from another institute for evaluation and management of chronic cough, wheeze & SOB which got exacerbated after she conceived.

On examination she had tachycardia (HR – 110/min) SpO2 -96% (RA) BP-110/70m6mHg. She had Ichthyotic skin and clubbing (Grade III). Retrospective history was taken in which she had a

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spontaneous miscarriage 8 weeks a year ago. She also complained of frequent chest infections and URIs since childhood which used to subside with a local doctor's prescription.

Radiological imaging was avoided as the patient was pregnant but was under regular follow up and was symptomatically better with inhalers, bronchodilators and anti-allergic medication. However, she still had wheeze on follow up at OPD visits. She was suspected of having CTD with ILD.

On 9th May 2023, this patient was presented to the primary hospital with premature rupture of membranes and was referred to our institute for delivery as she had bilateral wheeze, crackles and SOB. She was given a trial for vaginal delivery but however taken up for LSCS as there was nonprogress of labor. She delivered an alive male baby with good birth APGAR.

On post op day 1, she underwent a HRCT Chest which showed left sided bronchiectasis, situs inversus totalis. CT PNS was also done which revealed sinusitis. The patient and her family were informed regarding the triad and Kartagener's Syndrome.

Her baby's screening was done which had no significant radiological abnormality. Genetic screening was sent and is negative. The patient was discharged in a stable condition and was reviewed after 4 weeks with symptomatic relief of respiratory complaints. She has been advised for regular follow-ups.

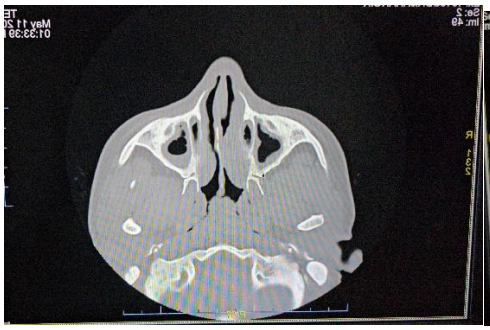

Fig 1- Sinusitis

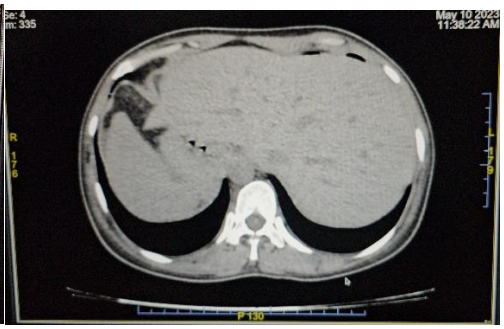

Fig 2 – Situs inversus of abdomen

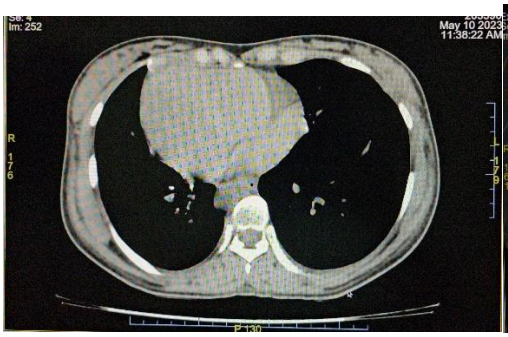

Fig 3 – Dextrocard

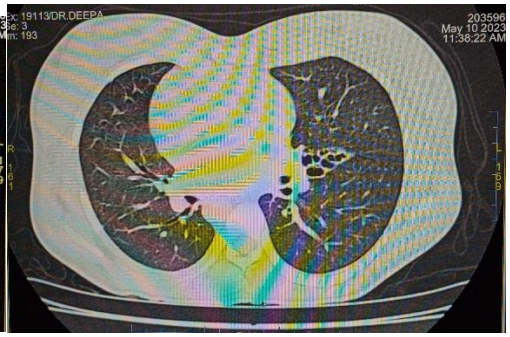

Fig 4 – Bronchiectasis and situs inversus

DISCUSSION

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Disorders of ciliary motility may be congenital/acquired. Congenital disorders are also referred to as PCDs. Almost 50% of PCD patients have situs inversus. Cases of PCD with situs inversus are known as Kartagener's Syndrome.[6]

PCD is a phenotypically and genetically heterogeneous condition in which the primary defect is in the functional integrity of cilia.[7,8] Almost 90% [9] of these PCD patients have these defects. It predominantly involves the outer dynein arms, inner dynein arms, or both. [9] 38%of the PCD patients carry mutations of the dynein genes *DNAI*[10] and *DNAH5*.[6]

This underlying defect leads to accumulation of secretions and thereby causing recurrent sinusitis, bronchiectasis, infertility, and situs inversus. The symptoms and the age at which the condition is diagnosed varies from individual to individual, even though the symptoms may be present from infancy.[11,12] Kartagener's syndrome sometimes associated with reversible airflow obstruction.[13] Clinical progression of the course of the disease may be variable sometimes requiring lung transplantationin severe cases.

The following are the Diagnostic criteria :-[14] clinico-radiological picture suggestive of recurrent chest infections, bronchiectasis, and chronic sinusitis, 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.

In addition to the above clinical criteria, two laboratory screening tests include exhaled nasal nitric oxide measurement, which is low in PCD, and saccharin test to assess mucociliary function of nasal epithelium. Diagnosis can be further confirmed by ciliary beat pattern and frequency analysis using video recording, and electron microscopic confirmation of the ultrastructural ciliary defect. The samples for these tests for examining motility and ultrastructure of cilia may be obtained by biopsy of nasal mucosa and laparoscopic biopsies of tubal mucosa in females, which was done by Halbert *et al*. In our case, the diagnosis was purely clinic-radiological, laboratory tests were not performed due to logistic reasons.

The fertility potential of women with Kartagener's Syndrome varies. Fallopian tube ciliary motility, in contrast to bronchial ciliary motility is not indispensable and hence several studies/case reports of pregnancy in KS have been reported. Yet, motile cilia are considered by many workers to be essential for normal ovum transport. More recently, bizarre ciliary motion has been described in the respiratory cilia of Kartagener's women.[16]

There is a case of successful IVF – ET done in Japan by Goro Kuramoto in KS lady who had a pretermCaesarean section delivery due to aggravation of lung disease during pregnancy but post-delivery, her lung function returned to pre labor period within 6 weeks.[17]

Even in our patient, from 4 weeks post-delivery, the patient was symptomatically better and had lesser respiratory symptoms when compared to during pregnancy.

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CONCLUSION

In conclusion, we presume that females with KS can successfully conceive and carry pregnancy to term. However, they are likely to have respiratory complications during their pregnancy in view of their underlying lung disease. Hence, watchful expectancy, timely diagnosis and prompt intervention will minimize the morbidity and ensure that the pregnancy continues to term.

These complications eventually will subside post pregnancy and hence the patient has to be managed to reduce respiratory complications and should be counseled about the expected complications and need for close monitoring thorough the course of her pregnancy.

We also conclude that diagnosis of KS in rural population in our country is still a challenge and are not diagnosed till they reach their middle age. We hope that this will change in the years to come.

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