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*RESEARCH ARTICLE*

# A RARE CASE REPORT OF KARTAGENER'S SYNDROME IN MOTHER AND OFFSPRING.

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## *Abstract*

Kartagener's syndrome (KS), a subgroup of the immotile-dyskinetic cilia syndrome, is a rare congenital disorder in the axoneme of the cilia characterized by the classic triad of recurrent rhinosinusitis, bronchiectasis, and situs inversus. It is generally considered that males with KS are almost all infertile. Some women with this syndrome are infertile, whereas some have successfully conceived. Although pregnancy is not impossible in women with KS, they are at high risk for sterility. We report a case of Kartagener's syndrome with normal fertility and the same disease inherited by the offspring. Both baby and mother after getting treatment for lower respiratory infection, were discharged in good condition.



## Introduction:-

Kartagener's syndrome (KS) or"Afzelius syndrome", a subgroup of the immotile-dyskinetic cilia syndrome, is a rare congenital disorder in the axoneme of the cilia,with mutation in Dynein gene DNAI and DAH5 and characterized by the classic triad of recurrent rhinosinusitis, bronchiectasis, and situs inversus with autosomal recessive inheritance. The prevalence of this syndrome has been estimated at 1 in 30,000 to 1 in 40,000 in general population. No difference in incidence in males and females. It is generally considered that almost all males with KS are infertile. Some women with this syndrome are infertile, whereas some have successfully conceived. The dyskinetic ciliary activity (or immotility) in the reproductive tract explains the fertility (or lack of it) in women with KS. Although pregnancy is not impossible in women with KS, they are at high risk for sterility. Although there is no cure for this syndrome treatment can be given for the management of symptoms like frequent respiratory, sinus, ear infections ,infertility etc. We present here a case of fertile woman with successful pregnancy outcome with same disease inheritance to offspring.

**Case Presentation:-**

A 32 year old female gravida3,para 1,live 1 married since 12 years, last child birth 10 years with previous LSCS, came with complaints of cough with expectoration and lower abdominal pain. She was a known case of Kartagener's syndrome with situs inversus, dextrocardia and bronchiectasis since childhood. She had recurrent episodes of URI and LRI since then. She was married for a period of 12years and with 3rd degree consanguinity. She conceived spontaneously after two years of marriage and delivered by LSCS indication being primi breech. Later after 10 years she conceived again spontaneously. There is no history of contraceptive use and was living with husband. Her 27 weeks gestation scan showed fetal situs inversus totalis. She presented to casualty with complaints of lower abdominal pain and severe cough and expectoration at 37 weeks of gestation.

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On examination patient afebrile, mild anaemic with clubbing of nails, no pedel oedma,PR-90/mt, BP-120/80 mm Hg,RR-18/mt, CVS normal S1S2 heard, RS- NVBS, bilateral wheeze and crepitations. Per abdomen uterus term, suprapubic scar present,head unengaged and FHR good. Patient was in early labour. After giving nebulisation and antibiotics and bronchodilators she was taken up for emergency repeat LSCS in view of previous LSCS with CPD in labour. She delivered an alive term male baby was admitted in NICU for respiratory distress and evaluation.

Her basic investigations were normal, ECG showed right axis deviation, ECHO – dextrocardia ,situs inversus, EF 61%, sputum culture showed Klebsiella pneumonia growth , CT chest scan revealed e/o ectatic changes with air fluid level in suprabasal, medial, posterior segment of left lower lobe, lingular lobe- Kartagener's syndrome with situs inversus totalis with infected bronchiectasis. Cardiology and thoracic physician opinion was obtained suggested iv antibiotics, nebulisation and bronchodilators. Meanwhile, first male child was evaluated and was found to be normal and the new born was evaluated in NICU, baby was with abnormal nasal discharge, hyperinflated chest X ray and ECHO showing dextro cardia and situs inversus, USG showed liver left side stomach and spleen right side and the diagnosis of primary ciliary dyskinesia was made. Both mother and baby were treated for respiratory problems and discharged in good condition on 19$^{th}$ postoperative day. Currently she maintains regular follow up.

## Discussion:-

Cilliary movement disorders may be congenital or acquired. Congenital cilliary disorders are also known as primary cilliary dyskinesias (PCD). PCD is an inherited autosomal recessive condition characterized by bronchiectasis, sinusitis and otitis media. Approximately, one half of the patients with PCD have situs inversus . When situs inversus is associated with PCD, then it is referred to as Kartagener's syndrome. As PCD is a cilliary movement disorder and as normal cilliary beating is necessary for rotation of different viscera during embryonic development, half of the PCDs will have situs inversus because of random rotation and the other half will have normal situs. Numerous defects of cillia are encompassed under this category including structural abnormalities of the dynein arms, radial spokes and microtubules. Structural abnormalities of dynein arms are the most common. The cilllia become dyskinetic, their coordinated, propulsive action is diminished and bacterial clearance is impaired. The clinical effects include recurrent upper and lower respiratory tract infections such as sinusitis, otitis media and bronchiectasis. Males are generally infertile because of immotile sperms. Some males have completely normal spermatozoa. The dyskinetic ciliary activity (or immotility) in the reproductive tract explains the fertility (or lack of it) in women with KS. That is, fertility depends on the degree and efficiency of the ciliary beat in the fallopian tubes,which is variable in women with the syndrome. Although women with KS have a variable degree of fertility about 50-60%, they definitely have an increased risk for sterility.

Diagnosis of KS is by detailed history,imaging studies like Xray, HRCT,nasal scrape or brush biopsy for electron microscopy,nasal nitric oxide measurement,saccharin test.

Since Kartagener's syndrome has no cure treatment involves managing symptoms and lowering risk of complications. And pregnancy as such is an immunocompromised state, active management of exacerbations need to considered. Here we can see that there was aggravation of symptoms. And baby inherited the same disease because of autosomal recessive pattern which increase with consanguinity.

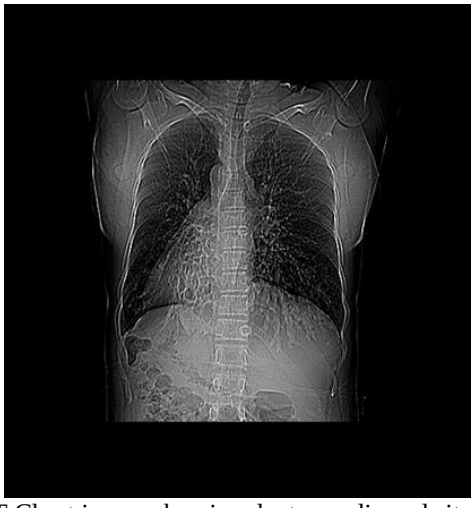

**Fig 1:-** CT Chest image showing dextrocardia and situs inversus

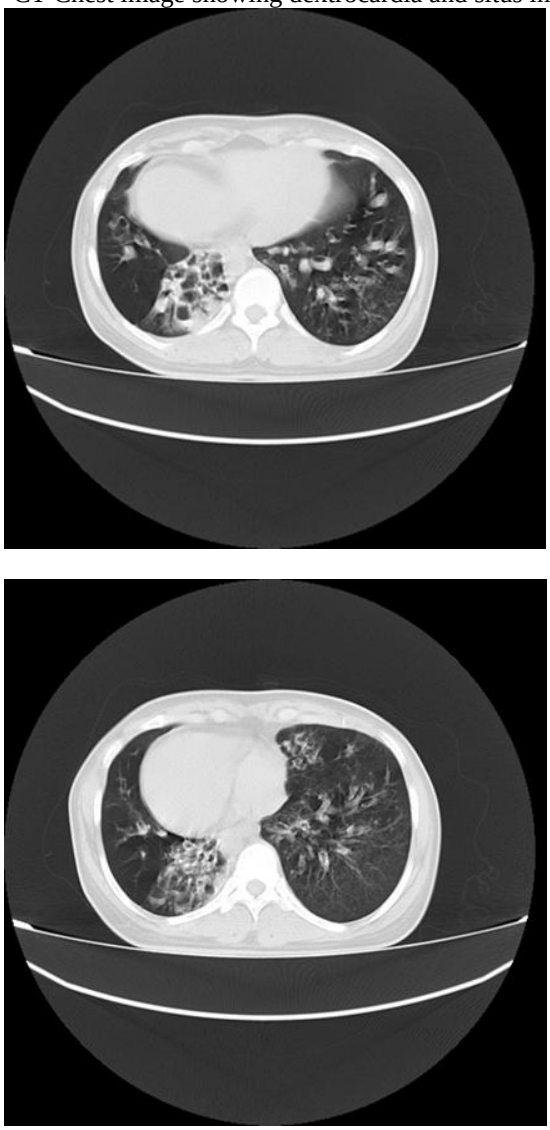

**Fig 2, 3:-** axial CT image showing wih e/o ectatic changes with air fluid level in suprabasal, medial, posterior segment of left lower lobe,lingular lobe.

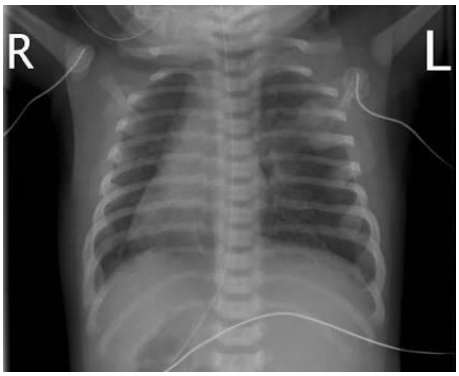


**Fig 4:-** Xray chest of newborn with dextrocardia,situs inversus

## Conclusion:-

KS can affect female fertility to various degrees because the ultrastructural defects in the ciliary axoneme are heterogeneous and lead to variable functional defects. These women should be counselled accordingly, they should be given genetic counselling and impact of consanguineous marriage, but should be encouraged by the possibility of conceiving and can have ordinary course of gestation. If they are found to be infertile, IVF-ET is a reasonable approach to try. Once a woman has successfully conceived, the KS is unlikely to affect adversely the course and outcome of the pregnancy.

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