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Case Report

Management of primary infertility in a patient with Kartagener's syndrome: a case report

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ABSTRACT

Kartagener's syndrome (KS) is a subgroup of primary ciliary dyskinesia (PCD), an autosomal recessive hereditary condition characterised by chronic sinusitis, bronchiectasis, and situs inversus. The fundamental pathophysiologic issue in Kartagener's syndrome is abnormal ciliary structure or function, which results in decreased ciliary motility. The patient in the present case was a 31-year-old female with a twelve-year history of primary infertility. In vitro fertilization and embryo transfer (IVF-ET) was performed and the patient conceived successfully. PCD should be strongly considered in women whose main infertility is accompanied by persistent respiratory symptoms. In women with PCD, superovulation and intrauterine insemination would not improve the likelihood of conception. In infertile women with PCD, IVF-ET is considered to be the best therapeutic option.

Keywords: Kartagener's syndrome, Infertility, Primary ciliary dyskinesia, Chronic sinusitis, Bronchiectasis, Situs inversus

INTRODUCTION

Kartagener's syndrome (KS) is a subgroup of the primary ciliary dyskinesias (PCDs), a broader class of ciliary motility disorders.[1] KS is a genetic autosomal recessive condition characterized by deficits in ciliary movement activity.[2] Siewart initially reported this ailment in 1904, but details were provided by Manes Kartagener's in 1933, and it is now known as Kartagener's syndrome.[3] It is distinguished by the classical triad of dextrocardia, bronchiectasis, and sinusitis. According to estimates, its occurrence is roughly 1 in 30,000 live births.[2]

Normal ciliary activity is required for respiratory host defence, sperm motility, and appropriate visceral orientation throughout development. The dynein axonemal heavy chain 5 (DNAH5) and dynein axonemal intermediate chain 1 (DNAI1) gene mutations in KS cause decreased ciliary motility, which predisposes to recurrent sinopulmonary infections, infertility, and left-right body orientation problems.[4] Both males and females might experience issues with fertility due to ciliary dyskinesia. Male infertility is caused by sperm immotility, whereas cilia activity in the fallopian tubes may be absent in females.[5] Due to ciliary function issues in the fallopian tube, women with PCD are at increased risk of infertility and ectopic pregnancy.

A significant index of suspicion is required to make an early diagnosis so that effective treatment options for infertility can be given to these young individuals, wherever possible. In addition, although unproven, early diagnosis seems important to preserve pulmonary function, quality of life, and life expectancy in this disease.[1]

We report a female patient with infertility secondary to KS and successfully conceived via in vitro fertilization and embryo transfer (IVF-ET).

CASE REPORT

The patient in the present case was a 31-year-old female with a twelve-year history of primary infertility. She came to our fertility clinic. She had the typical triad of KS (dextrocardia, chronic sinusitis and bronchiectasis) with history of recurring respiratory issues from childhood, including difficulties in clearing sputum and dyspnoea.

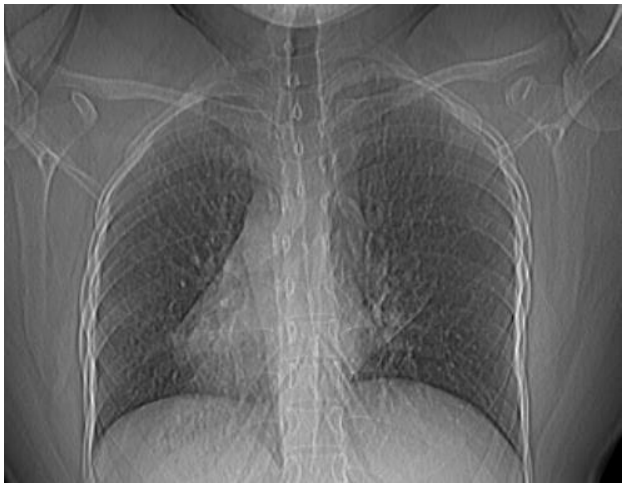

Figure 1: Chest X-ray showing dextrocardia.

She had a regular menstrual cycle and no additional symptoms or surgery. Hysterosalpingography was normal. Her partner is 35 years old with normal secondary sexual characteristics, laboratory workups including semen analysis. She underwent five cycles of intrauterine insemination (IUI) at an outside clinic. Unfortunately, the patient was unable to conceive. On female evaluation ultrasound revealed normal sized uterus, right ovary normal with 5 antral follicles and left ovary with 4 antral follicles. Then she underwent IVF treatment with antagonist protocol. 8 eggs collected. Intracytoplasmic sperm injection (ICSI) was done with husband sperm. Six day 3 embryos were frozen. She underwent artificial cycle frozen embryo transfer. Three day 3 embryos were transferred and beta-human chorionic gonadotropin (hCG) test was positive. During her pregnancy, she developed intermittent respiratory tract infections. At 28-week period of gestation, she had oligohydramnios with preeclampsia. At 34-week period of gestation, the amniotic fluid index was 2.9 cm. In view of severe oligohydramnios, she underwent an emergency lower segment caesarean section (LSCS). she gave birth to a healthy female baby weighing 2.5 kg. Post-operative period was uneventful.

DISCUSSION

Kartagener's syndrome (KS) or type 1 primary ciliary dyskinesia is a hereditary condition where there is absent or inefficient ciliary action. It has an autosomal recessive inheritance pattern, and affected individuals might be heterozygous or homozygous for the disorder. PCD is a phenotypically and genetically heterogeneous condition in which the predominant abnormality is in cilia ultrastructure or function.[1] Approximately one-half of PCD patients have situs inversus. When situs inversus is associated with PCD, the condition is known as Kartagener's syndrome.[6]

It is typically diagnosed in childhood, only a few cases diagnosed in adults.[7] It has the characteristic triad of dextrocardia, bronchiectasis, and sinusitis. Kartagener's syndrome can be situs inversus totalis, when all the viscera are located on the opposite side, or situs solitus, where just dextrocardia is present. The patient in this report had situs solitus.[3]

The ultrastructure of the cilia in KS patients has been shown to consist of Dynein arm abnormalities. The absence or dysfunction of dynein arms, radial spokes, and ciliary microtubules are recognised structural and functional defects of ciliary ultrastructure. The majority of disease-causing mutations are thought to involve two genes that code for the DNAH5 and DNA I1. These faulty genes cause the cilia to be the wrong size or shape or move the wrong way, causing the cilia to have defective motility.[4]

Clinical effects include recurrent upper and lower respiratory tract infections such as sinusitis, otitis media, bronchiectasis, and infertility in both males and females.[3]

The recommended diagnostic criteria for this syndrome include a history of chronic bronchial infection and rhinitis since early childhood, along with one or more of the following characteristics: situs inversus or dextrocardia in the patient or a sibling, alive but immotile spermatozoa, absent or impaired tracheobronchial clearance, and cilia exhibiting a distinctive ultrastructural defect on electron microscopy. Exhaled nasal nitric oxide level measurement and the saccharin test for measuring nasal epithelial mucociliary function are two laboratory screening procedures. Confirmatory laboratory procedures include high-speed video microscopy to examine ciliary beat frequency and pattern, transmission electron microscopy to detect ultrastructural ciliary defects, and genetic testing for DNAI1 and DNAH5 mutations.[4]

Here patient presented with recurrent episodes of sinopulmonary infections. Imaging findings revealed bronchiectasis, dextrocardia, and situs inversus, which met the diagnostic criteria for KS.

As a hereditary condition, KS has no definite treatment. Patient management is symptomatic and includes intermittent or continuous oral or intravenous administration of antibiotics to treat respiratory infections. Bronchiectasis and pneumonia should be treated with inhaled bronchodilators, mucolytics, oral corticosteroids,

and chest physiotherapy. Vaccinations against influenza and pneumococcus are also required to avoid recurrent infections.[8]

Women with KS have a lower fertility rate; spontaneous conception is not possible in them because fertility is dependent on the degree and effectiveness of the ciliary beat in the fallopian tubes, which varies in women with the condition.[1,9] Here, the patient was considered to be a case of primary infertility due to tubal ciliary dysfunction secondary to KS.

As the present patient had 5 cycles of IUI failure, the next treatment of choice for this patient was IVF-ET. IVF-ET was done to the patient, and she conceived successfully with the help of the procedure.

CONCLUSION

In conclusion, KS may affect female fertility to various degrees because the ultrastructural defects in the ciliary axoneme are heterogeneous and lead to variable functional defects. Although these women are at risk for infertility, they should be encouraged by the possibility of spontaneous pregnancy. If they are found to be infertile, IVF-ET is a reasonable approach to try than IUI.

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