Case Report

Kartagener Syndrome: A Rare Cause of Infertility

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ABSTRACT

Kartagener’s syndrome is defined as motility dysfunction of the epithelial cilia lining the respiratory tract, fallopian tubes and the flagella of the sperm and genetically classified as a rare autosomal recessive disease consisting almost half of all primary ciliary dyskinesia cases. In this brief case review we aim to share our experience concerning Kartagener’s syndrome from a gynecologic point of view regarding an unexplained infertility patient.

Keywords: Kartagener, Infertility


Introduction

Kartagener’s syndrome is a rare autosomal recessive genetic disorder characterized by a defect in the action of the cilia lining the respiratory tract, the fallopian tube, and also of the flagella of the sperm in males presenting with bronchiectasis, sinusitis, dextrocardia or situs inversus and infertility and it is responsible for almost half of the Primary Ciliary Dyskinesia cases which has a fairly low prevalence of 1 in 20000 live births.

First described by Maner Kartagener in 1933, he presented four cases including the triad to support his findings and therefore the disease was named after him. Later in 1998 Afzelius first mentioned ultrastructural deficiency causing ciliary dyskinesia with the help of electron microscope. Electron microscopic evaluation of these patients reveal that there are ultrastructural abnormalities in the inner and/or outer dynein arms of epithelia cilia, microtubules or radial spokes and the most common ultrastructural defect is the absence of dynein arms.1,2,3,4,5

In this article we aim to share our experience of a patient diagnosed with undefined infertility which later was revealed to be Kartagener’s syndrome, in regard of the current literature.

Case Report

A 33 years old nulliparous patient with regular menstruation, adequate ovarian reserve and a normal hysterosalpingo-graph, diagnosed with undefined infertility, was admitted to our clinic. Upon first examination and ultrasonography no pathological findings could be documented.

Patient history revealed that she used to have frequent respiratory tract infections from early years in life up to now and this statement led us to suspect Kartagener’s disease for the cause of infertility, in addition there was no record of Kartagener’s disease reported in the patient’s family history therefore all available diagnostic tools were aimed at this direction.

First a whole body computerized tomography was performed which revealed dextrocardia, complete obliteration of maxillary sinuses due to chronic sinusitis and signs of chronic lower respiratory tract infections namely tubular bronchiectasis.

This triad along with undefined infertility is adequate to diagnose the patient as Kartagener’s syndrome but Genetic testing for mutations in the genes DNAI1 and DNAH5 was performed by our genetics department laboratories showing biallelic mutations hence supporting the diagnosis.

Patient was given genetic counsel upon the characteristics and behavior of the disease and its autosomal recessive genetics.

Patient was later prepared for in vitro fertilization and managed to conceive a singleton pregnancy in the first cycle of her treatment which was healthily delivered vaginally at 38th week of gestation.

Discussion

With a prevalence of 1/20000 Kartagener’s syndrome or Primary Ciliary Dyskinesia is not the first thing to bear in mind when faced with undefined infertility moreover it is a diagnosis often missed even in recurrent respiratory tract infections let alone infertility.
A defect in ciliary clearance is known to result in recurrent infections especially in the respiratory tract.6,7 Moryan et al.8 had defined 3 sub-groups in ciliary dysfunction and Rosman et al.9 distributed them according to their frequency.

1: Dynein defect (95.5 %)
2: Radial Spoke defect (72.5 %)
3: Transposition of microtubules (31.5 %).

Our patient was in the first group with documented dynein defect.

As mentioned in the introduction the triad needed for the diagnosis of Kartagener’s syndrome was also present in our patient.

Concerning female infertility along with recurrent respiratory tract infections one should bear the possibility of Kartagener’s syndrome in mind and refer the patient for in vitro fertilization once the diagnosis is made in order to prevent the loss of time and protect the remaining ovaries that might otherwise spent on other assisted reproductive techniques.

Kartagener Sendromu: Nadir Bir İnfertilite Nedeni

ÖZET

Kartagener sendromu; respiratuvar trakt, fallop tüpü ve ayrıca sperm flagellasındaki epitelyumun hareket bozukluğuyla karakterize oldukça nadir, otozomal resesif geçiş gösteren genetik bir hastalıktır ve primer silyer diskinezili vakalarının neredeyse yanını oluşturur. Bu olgu sunumunda nedeni açıklandan- mayan infertilite tanısı olan fakat daha sonra Kartagener sendromu olduğu anlaşılan bir hastayı ve sendromu jinekolojik bir bakış açısından değerlendirerek istedik.

Anahtar Kelimeler: Kartagener, İnfertilite

References