A Case of Kartagener Syndrome

Nasrin Ghasemi and Naeimeh Tayebi

The Kartagener syndrome is an autosomal recessive disorder characterized by bronchiectasis, sinusitis and situs inversus. This research reports a 28 years old woman with dextrocardia ask for genetic counseling to have a healthy child. She suffers from recurrent chronic sinusitis and bronchitis with situs inversus including right-sided heart and pancreas and left-sided liver. Kartagener syndrome was diagnosed according to her signs, symptoms, radiological and ultrasonographic evaluations.

Key words: Dextrocardia, sinusitis, bronchitis, Kartagener syndrome

¹Department of Medical Genetics, Infertility Research and Clinical Centre, Yazd Shahid Sadoghi Medical Sciences University, Yazd, Iran
Infertility Research and Clinical Centre, Yazd Shahid Sadoghi Medical Sciences University, Yazd, Iran
INTRODUCTION

Kartagener Syndrome (KS) is an autosomal recessive disorder with incomplete penetrance (Gorham and Merselis, 1959), which characterized by the triad of situs inversus, bronchiectasis and sinusitis (Casanova et al., 2006). The primary defect is abnormal function of cilia, which causes immotile cilia syndrome. The immotile cilia cause recurrent sinopulmonary infection (Chudhari et al., 2004). KS was seen in consanguinity especially first cousin (Moreno et al., 1965). It is likely that the Kartagener syndrome is a mendelian subgroup of situs inversus viscerum (Torgersen, 1956). There is no association with left-handedness. Dextrocardia may be absent and the patient may suffer from bronchiectasis, sinusitis and infertility (Guerrant et al., 1978). Patients with the Kartagener syndrome may have anosmia (Goldstein, 1979). The sperm of males with Kartagener syndrome are immotile, which is the same as every ciliary cell in the body (Azzenius et al., 1976). The spermatozoa have fertility capacity and with using assisted reproductive technique, they will fertilize the egg. Women with Kartagener syndrome usually are fertile, although eggs are moved down in fallopian tube by cilia. So using in vitro fertilization in the management of such patients is a possibility (Aitken et al., 1983).

CASE REPORT

This case report described a 28 years old woman sought genetic counseling to have a healthy child. She suffered from recurrent chronic sinusitis and bronchitis and with using antibiotic; the patient got rid of that signs and symptoms for a few weeks. She has situs inversus including right-sided heart and pancreas and left-sided liver in physical examination. For complete evaluation, ultrasonography of internal organs, chest X-ray and echocardiography were asked. Right-sided pancreas and left-sided liver were visualized in sonography. Dextrocardia with bronchiectasis were seen in chest X-ray. The apex of heart was right in echocardiography, but the valves and walls were normal. The sign of chronic sinusitis was seen in the radiography of sinus. Laboratory tests include fasting blood sugar, triglyceride, cholesterol, SGOT, SGPT, creatinine, sodium and potassium were normal (Table 1).

Table 1: The results of the laboratory test for this patient

<table>
<thead>
<tr>
<th>Lab test</th>
<th>FBS</th>
<th>TG</th>
<th>CHOL</th>
<th>AST</th>
<th>ALT</th>
<th>CREA</th>
<th>SOD</th>
<th>P.O.T</th>
</tr>
</thead>
<tbody>
<tr>
<td>Results</td>
<td>94*</td>
<td>176*</td>
<td>189*</td>
<td>34**</td>
<td>32**</td>
<td>0.9*</td>
<td>135*</td>
<td>4.5*</td>
</tr>
<tr>
<td>*ng dl^-1; **U L^-1</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

She had an 18 years old brother with the same problem, dextrocardia and situs inversus, but he did not accept to have more evaluation. In addition parents of them were second cousin. The pedigree of patient was drawn as follow:

![Pedigree Diagram]

DISCUSSION

Kartagener syndrome is originally described as the combination of situs inversus, bronchiectasis and sinusitis (Casanova et al., 2006). This syndrome is a mendelian disorder, which inherited as autosomal recessive. Namayan et al. (1994) suggested either X-linked or autosomal dominant inheritance for this syndrome.

Evaluation of this syndrome by genetic test is difficult because of genetic heterogeneity. In various linkage studies, this syndrome has been known as a disorder with a great genetic heterogeneity. It has not been determined any gene in the pathogenesis of this syndrome until now but some studies reported the association of Kartagener syndrome with linkage to chromosome 7 (Witt et al., 1999), heterozygous mutations in the DNAI1 (located on 9p21-p13) and DNAH5 genes (resides on 5p15-p14) (Guichard et al., 2001; Olbrich et al., 2002). In present study, genetic evaluation was not done, because of indefinite affected genes.

There are new reports of this syndrome which confirm diagnosis according to clinical and paraclinical findings (Ishiga et al., 2005; Naves et al., 2005).

In present study, a 28-years-old woman was described with recurrent chronic sinusitis and bronchitis. Right-sided pancreas and left-sided liver were visualized in sonography. Chest X-Ray confirmed dextrocardia with bronchiectasis. Also, the signs of chronic sinusitis were seen in the sinuses grapy. Therefore, Kartagener syndrome was diagnosed for this patient.

CONCLUSIONS

According to heterogeneity of this disease, genetic evaluation of it is very expensive and usually useless. Genetic counselor recommended this couples to have a
REFERENCES


