

## Case Report

### A New Diagnosis Klinefelter Syndrome Refer To Hospital with Dyspeptic Complaints

Hatice Beyazal Polat<sup>1</sup>, Aydın Demirbaş<sup>2</sup>, Teslime Ayaz<sup>3</sup>, Sibel Demirbaş<sup>4</sup>

<sup>1,2,3,4</sup> Recep Tayyip Erdogan University, Faculty of Medicine, Department of Internal Medicine, Rize, Turkey

#### Abstract:

Klinefelter Syndrome is the most common sex chromosomal disorder in males. The clinical features of Klinefelter syndrome are characterized by significant testicular dysfunction, hypogonadal symptoms, atrophic testes and gynecomastia. These patients can apply to the physician with symptoms specific to the syndrome or general symptoms. In addition to specific physical examination findings, the hormone profile is consistent with primary hypogonadism. The definitive diagnosis is made by karyotype analysis of peripheral blood lymphocytes. Early diagnosis and treatment are important. With hormone replacement therapy, muscle mass, bone mineral density and libido are improved. In general, patients with Klinefelter Syndrome are infertile, but fertilization can be achieved with assisted reproductive techniques. We present a case with dyspeptic symptoms and suspected physical examination findings and diagnosed as Klinefelter Syndrome.

**Keywords:** Dyspepsia, Infertility, Klinefelters syndrome.

#### Introduction

Klinefelter syndrome is the most common sex chromosomal disorder in males, characterized by additional one or more X chromosomes. The prevalence is approximately 1 in 600 males at birth. Although the diagnosis is easy, only 25% of the patients can be diagnosed. Klinefelter Syndrome is a systemic problem, but most of the cases diagnosed from hospital admission with infertility. Typical clinical features of Klinefelter Syndrome are eunucoide masculinization owing to the lack of androgens and long stature, gynecomastia, small testes, azoospermia and infertility. Patients are diagnosed during and after puberty. Laboratory and clinical findings in adult patients are consistent with hypergonadotropic hypogonadism. The definitive diagnosis is made by chromosome analysis from lymphocytes in the peripheral blood. Genetically, 80% of patients in the form of classical structure 47XXY, while the remaining 20% of mosaic genetic structure is observed (1).

In addition to infertility, patients with Klinefelter Syndrome have also been shown to have significant health problems leading to high morbidity and mortality (2,3). Therefore, early diagnosis of these patients is important.

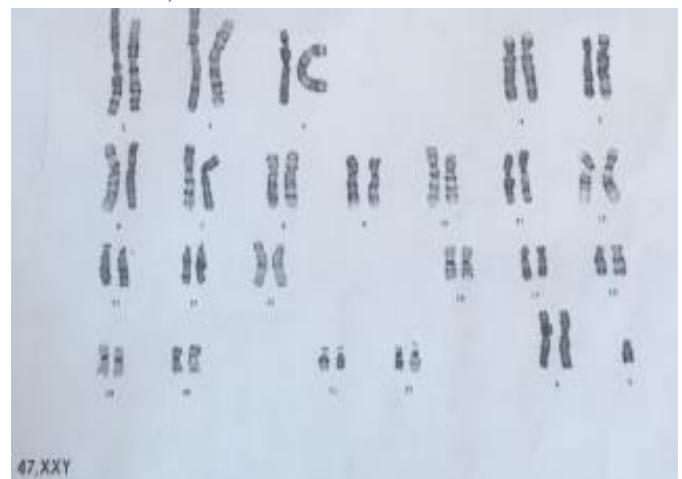
We present a case of dyspeptic symptoms that we diagnosed with Klinefelter Syndrome. Our aim is to emphasize that the physical examination of patients is still very important with this case.

#### Case

A 46-year-old married male patient presented to our clinic with complaints of bilateral lateral pain, epigastric pain and abdominal distention. He had no special medical history accept of previous operation with peptic ulcer ,20 years ago. Family history was unremarkable. On physical examination,

his weight was 88 kg and height was 177 cm. Phenotypically, hypogonadism was observed. There was a reduction in the

beard and mustache. He had gynecomastia. There was a reduction in axillary and pupil hair. At the genital system examination, the penis was normal in size, the meatus structure was normal, the testes were atrophic. In testicular USG, both of testes were atrophic, highly heterogeneous and microlithiasis were seen. When his story was deepened, it was learned that he had been married for 15 years and had no children. The patient, who had never been investigated due to infertility, was examined further. Blood biochemistry and hemogram were normal. The patient's alkaline phosphatase (ALP) and parathormone (PTH) level were within normal limits. Vitamin D deficiency was also detected. FSH: 36.46 mIU / ml (0.95-11.95), LH: 19.34 mIU / ml (0.57-12.07), Testosterone: 102.55 ng / dl (142.4-923,1) Prolactin: 10.85 ng / ml (3.46-19.4). DEXA: Left femur: T score was -3.1. Azoospermia was detected in the patient's semen analysis. Karyotype analysis was performed. Chromosome analysis was found to be 47, XXY.



**Figure 1: Chromosome analysis by GPL banding after peripheral blood cell culture of the patient.**

**Discussion**

Klinefelter Syndrome is the first identified chromosome anomaly. It is characterized by hypogonadism and eunucoide body structure. Klinefelter Syndrome is the first chromosomal anomaly. It is characterized by hypogonadism and anterior body structure. No significant phenotypic differences occur until puberty. Puberty time is normal, but the testes are small and secondary sex characters are underdeveloped . Although serum testosterone levels are not always very low, it is an important laboratory finding that serum follicle-stimulating hormone levels are above normal. The studies show that the prevalence of Klinefelter syndrome is increasing due to advanced father age (4).

Klinefelter's syndrome is a syndrome that is usually diagnosed during adulthood due to late puberty and infertility. It may be difficult to diagnose because of the inability to detect a typical phenotype in each case (5). Klinefelter Syndrome is detected in 11% of azoospermic male and in 4% of applicants with infertility. A typical finding in semen analysis in the majority of patients with Klinefelter Syndrome is azoospermia. Although the patients with Klinefelter Syndrome are considered as infertile, it can be fertilized with some assisted reproductive techniques. In our case there was infertility. Hormone profile was consistent with primary hypogonadism.

Osteoporosis is frequently seen in patients with Klinefelter Syndrome. The reason for this is thought to be the antiresorptive effect of testosterone hormone, leading to an increase in bone density in the lumbar spine. In a study comparing 823 Klinefelter cases in Denmark with 4022 same age group control cases, significant health problems such as osteoporosis and diabetes mellitus were increased in individuals with Klinefelter syndrome (6).

We've detected low levels of testosterone in our case. We determined that bone mineral density was low in the DEXA measurements of the patient.

Klinefelter Syndrome can lead to serious health problems that cause not only infertility but also significant morbidity and mortality.

Therefore, the evaluation of men with Klinefelter Syndrome by multidisciplinary approach is important for determining the morbidity of patients.

As a result, Klinefelter Syndrome is most commonly diagnosed while investigating the cause of infertility. However, it should be kept in mind that these patients may also consult the physician with the disease-specific other than infertility and disease-unrelated symptoms. And Klinefelter Syndrome can be diagnosed with systemic evaluation of patients .

The aim of this case report is to emphasize that physical examination findings may be a clue to the syndromes apart from the complaints that bring the patient to us. It should be kept in mind that the presence of infertility together with physical examination findings may be associated with Klinefelter Syndrome.

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