

Klinefelter's Syndrome in 10 Year-Old Boy: Case Report



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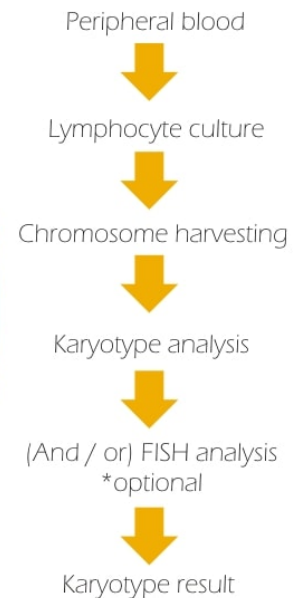
Introduction

A 10-year-old boy came to clinic at Cell Culture and Cytogenetics Laboratory, Faculty of Medicine Universitas Padjadjaran, Indonesia. He was referred by physician at Department of Pediatrics, Rumah Sakit Hasan Sadikin. Diagnosis showed that the patient has penoscrotal hypospadias and surgery was a feasible further action. However, chromosome analysis was prompt to determine accurate genetic condition of the patient. Patient came to clinic for more thorough examination. General look showed he has normal appearance of 10-year-old boy. With body height of 135 centimetres and weight 30 kilograms, he was in average of normal Indonesian elementary school aged boy. Examination revealed the patient has micropenis, hypospadias, both testicles were present, and 3 centimetres of penile phallus at relaxed condition. Many previous studies of patients conducted at Cell Culture and Cytogenetics Laboratory showed that condition would revealed a normal 46,XY karyotype. Still, chromosomal analysis was fundamentally needed before any further medical action.

Abstract

Klinefelter's syndrome is one of the most common sexual development abnormalities with incidence of approximately 1 in 500 live male births. It is the most familiar cause of delayed or incomplete puberty as well as leads to infertility. This abnormality is characterised by 47,XXY karyotype. This report presents a 10 year-old male with micropenis came to clinic. First clinical examination showed the patient has small penile phallus and hypospadias, but has no enlarged breast and shows other normal male child physical general appearances. Lymphocyte obtained from peripheral blood was cultured and chromosome analysis was performed using conventional karyotyping method. Karyotype analysis revealed 47,XXY confirming suspicion of Klinefelter's syndrome. Genetic counselling for patient and parents was conducted regarding this chromosomal condition. Counselling emphasised future medical treatment possible to overcome issues emerged from both physiological condition as well as psychological state of patient and family. Testosterone therapy will be started early to minimise the effect of hormonal deficiency on patient's physical condition.

Methods



Results



Figure 1. Genital appearance of patient

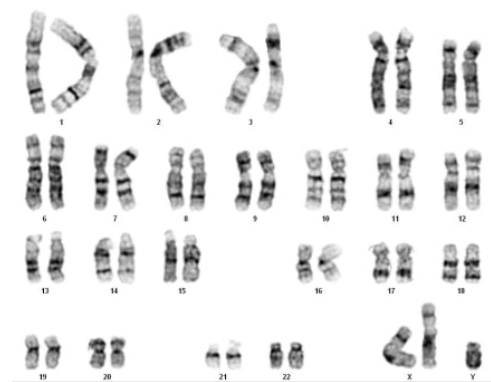


Figure 2. Karyotype result

Karyotype analysis showed that the patient has 47,XXY confirming Klinefelter syndrome. It is the most common chromosomal aneuploidy in male population of 1 in 500 newborn. The prevalence rate is 5-20 times higher in individual with mental retardation. Genetic counselling for patient and parents had been conducted regarding this condition to inform future medical treatment possible. In future, physician had planned to conduct testosterone therapy early to minimise hormonal deficiency effect on patient.

Conclusion

It was confirmed the patient has Klinefelter syndrome. Further study was needed to achieve comprehensive knowledge regarding physiological condition of particular patient as well as psychological condition.

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Keyword: Klinefelter syndrome, karyotype, disorder of sexual development, genetic counselling