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CASE REPORT

MULTIPLE CELL LINES IN A SHORT STATURE PATIENT

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ABSTRACT

Short stature is a term applied to a child whose height is two standard deviations (SD) or more below the mean for children of that sex and chronological age. Turner syndrome is an important consideration in girls with short stature and especially growth failure, because shortness may be the presenting feature of this syndrome, other physical abnormalities can be variably expressed. Here we present a case of short stature, with some congenital abnormalities. The cytogenetic analysis from peripheral venous blood revealed four cell lines, 46, XX (10%) /45, X (10%) /46, X, i (Xq) (60%) /46, XX (ter rea) (20%), which is a rare finding.

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INTRODUCTION

Short stature is common reason of referral, for pediatric subspecialty evaluation and is a common indication for genetic evaluation. The purpose of genetic evaluation of short stature is to provide accurate diagnosis and to provide information to the patient and family regarding natural history, prognosis, available treatment, genetic basis and recurrent risk (Laurie Seaver *et al.,* 2009). The recumbent length or height of 95 percent of normal children lies between the 3rd and 97th percentiles of height or within 2 standard deviations above or below the mean height for the age. If the length or height of the child is below the 3rd percentile or less than 2 S.D. from the mean, he or she is considered to be short in stature (Ghai, 2007). Short stature may either be proportionate or disproportionate. One of the causes for proportionate short stature is chromosomal disorder (Ghai, 2007). Turner syndrome is an important consideration in girls with short stature and especially growth failure, because shortness may be the presenting feature of this syndrome, other physical abnormalities can be variably expressed. Virtually all girls with Turner syndrome have short stature, with an average adult height about 20 cm shorter than predicted by mid parental height. Incidence of Turner syndrome is approximately 1 in 4000 female live births. More than 99% of 45, X conceptuses abort spontaneously, accounting for one fifth of all spontaneous abortions (Nussbaum *et al.,* 2004).

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CASE REPORT

A 10 year old female child was referred to Genetics division for karyotyping with chief complaints of failure to gain in height. The learning capacity of the child was average (studying in 4th standard). Family history revealed 2nd degree consanguinity in parents. Proband had 2 elder sisters and one younger brother, aged 15 years, 13 years and 6 years respectively with the normal height. Both sisters have attended menarche. Mother had history of abortion after first daughter. Clinical examination showed short stature, shield chest with widely placed nipples, short neck, cubitus valgus, large ears, wide gap between 1st and 2nd toe bilaterally, small 3rd and 4th toes, hyper convex and upturned nails. External genitals were normal with normal vaginal orifice. Cardiovascular and urinary system was normal. On anthropometric measurements**,** height was 110 cms.

The normal height for age, 3rd percentile, according to CDC 2000 standards is 126cm (OP Ghai,2007).Weight was 16.6 Kgs and span was 108 cms .On ultrasonography it revealed uterus: 20X8X4mm (Small in size) and bilateral streak gonads. Buccal mucosa was scraped and stained with thionine for sex chromatin analysis, showed only 5% cells positive for Barr body. Peripheral venous Sample of the patient was subjected for cytogenetic analysis using lymphocyte culture. GTG – banding analysis of metaphase chromosomes revealed four cell line mosaicism i.e 46, XX (10%) /45, X (10%) /46,X,i(Xq) (60%) /46, XX (ter rea) (20%).

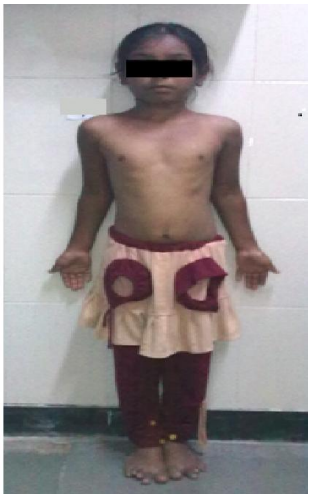

Fig. 1. Showing patient with short stature, widely placed nipples, cubitus valgus

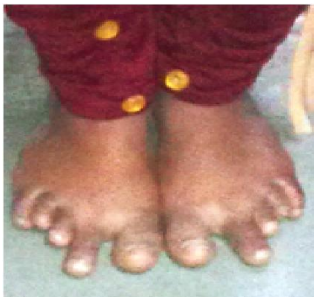

Fig. 2. Showing wide gap between first and second toe, small 3rd and 4th toes

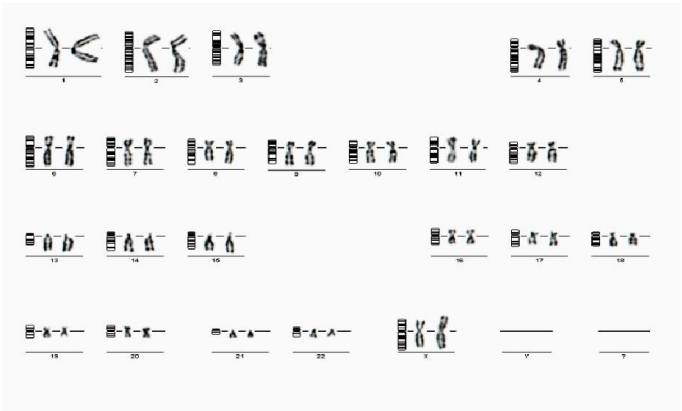

Fig. 3. Karyotype showing 46,X,i(Xq)

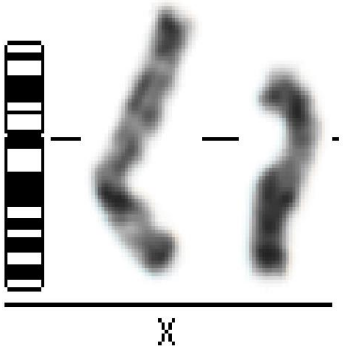

Fig. 4. ter-rea

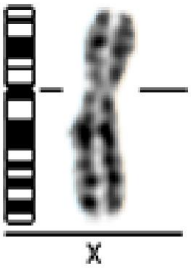

Fig. 5. Monosomy X

DISCUSSION

The karyotype 45, X is responsible for 40-60% of patients with Turner syndrome (A. de la Chapelle, 1983). However, about 50% of cases have other karyotypes. About 15% of patients are mosaics for 45, X and a normal cell line (45X/46XX). Others mosaics are with isochromosomes, 45X/46, X, i(Xq) and with rings, 45, X/46, X, r(X); or with fragments, 45X/46 fra (Robert Rapaport, 2004). In this patient, the normal cell line showing 46, XX karyotype is only 10%, Monosomy of X, is seen in 60% cell line. 45, X karyotype is responsible for short stature and other somatic abnormalities found are due to both cell lines that is 45 X, and 46,X.i(Xq). 46, XX (ter rea) is a rare but well known, it also leads to clinical features of Turner syndrome or to simple gonadal dysgenesis with stunted growth.

Management and Genetic counseling

One justification for treating short children is to improve their psychosocial adaptation (David *et al.,* 1994). Growth hormone therapy is now a standard treatment in Turner syndrome and can result in gain of 6 to 10 cm to the final height (Nussbaum, 2004). Replacement therapy with estrogens is indicated, but there is little consensus about the optimal age at which to initiate treatment. The psychological preparedness of the patient to accept therapy must be taken into account. Considering all these facts Genetic counseling of patient as well as parents was carried out for proper treatment and rehabilitation of patient.

REFERENCES

David, E., Sandberg, Amy E., Brook, Susna P., Campos, Pediatrics, December 1994, Volume 94/Issue 6.

De La Chapelle, A. 1983. Sex chromosome abnormalities, Principles and Practice of Medical Genetics, Vol 1, *Churchill Livingstone*, 193-200.

Ghai, O. P., Piyush Gupta, V. K. Paul, Disorders of Growth and Development, Ghai Essential Pediatrics, 6th edition, 2007: 50.

Ghai, O. P., Piyush Gupta, V.K., Paul, Growth and Development, Ghai Essential Pediatrics, 6th edition, 2007: 27.

Nussbaum, McINNES, Willard, Clinical Cytogenetics: Disorders of the Autosomes and the Sex chromosomes, Thompson and Thompson Genetics in Medicine, 6th Edition, 2004: 165-179.

Robert Rapaport, Disorders of the Gonads, Nelson Text book of Pediatrics, 17th Edition, 2004:1931.

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