Klinefelter Syndrome Associated With Short Stature Due To Iatrogenic Cushing

Renata M. Pinto, MD, MSc[1,2,3,4]; Lucas S. Steinmetz[4] ; Julio M. G. Barbosa[4] ; Arthur F. C. S. Mendes[4] ; Damiana Mirian Da Cruz Cunha, MSc[1,2,5]; Aparecido D. da Cruz, MSc, PhD[1,2,5,6]

[1] Department of Biology, Replicon Research Center, Catholic University of Goiás, Goiânia, GO, Brazil; [2] Postgraduate Program in Genetics, Catholic University of Goiás, Goiânia, GO, Brazil; [3] Health Sciences PhD Program, Federal University of Goiás, Goiânia, GO, Brazil; [4] Department of Pediatrics, Federal University of Goiás, Goiânia, GO, Brazil; [5] Laboratory of Human Cytogenetics and Molecular Genetics, Secretary of State for Health of Goiás (LACEN/SESGO), Goiânia, GO, Brazil; [6] Biotechnology and Biodiversity Graduate Program, University of Brasília, Brasília, DF, Brazil

INTRODUCTION

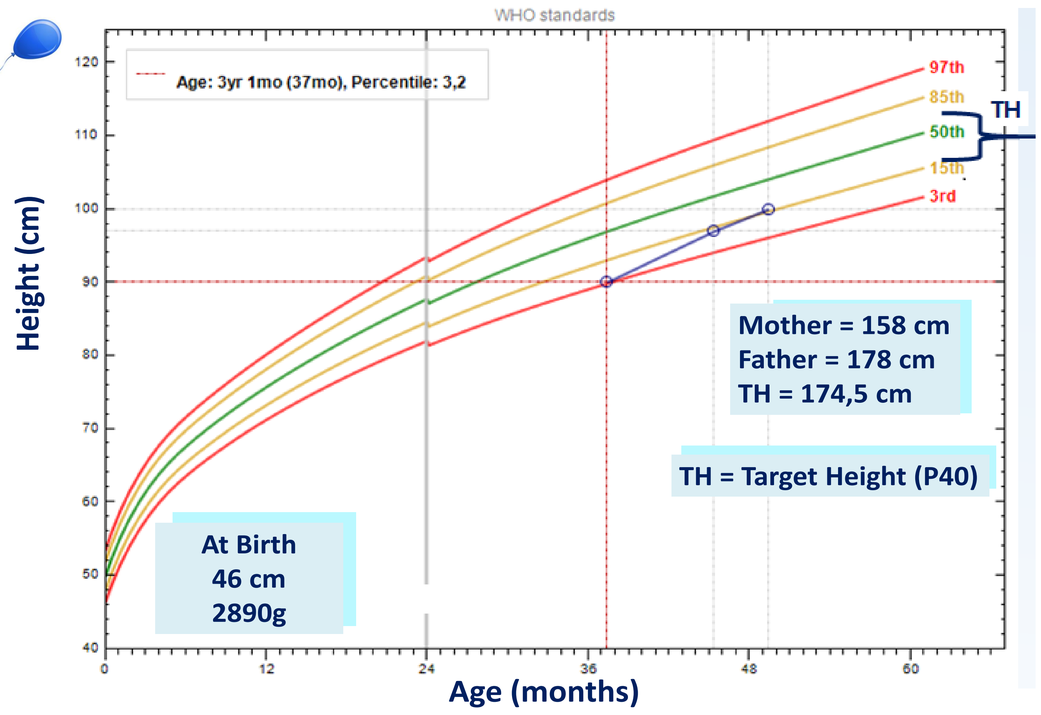
Klinefelter syndrome (KS) is a form of aneuploidy resulting from 2 or more X chromosomes in a male. The most common karyotype is 47,XXY. KS affects physical and intellectual development to varying degrees, commonly causing hypo-development of secondary sexual characters and high stature.

CASE REPORT

JLV, male, 3 year 1 month age, the first child of a young non-consanguineous couple, was referred to the pediatric endocrinology department with Short Stature (SS) main complaint. The mother's height is 158cm, and the father's height 178cm (Target height 174.5cm).

He had clinical history of atopic dermatitis and allergic rhinitis, and because of theses diagnosis the patient used during the last year: nasal beclomethasone (twice a day), nasal fluticasone (three times a day), topical hydrocortisone on the scalp (once a day) and oral methylprednisolone during periods of wheezing. All drugs were prescribed by doctors.

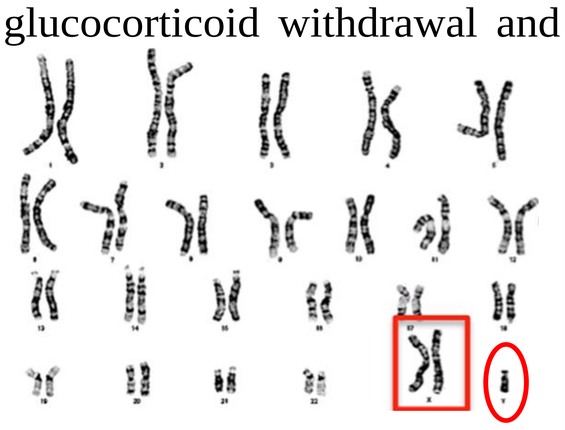
Physical examination revealed the increase of lanugo throughout the body and syndromic facies: retroverted small ears with low implantation, ocular hypertelorism, wide nasal base, oval palate. Normal segmental examination, G1P1. 90.5cm (- 1.99 SD) and 10kg (- 3.17 SD).



General biochemical tests, thyroid, renal and hepatic functions were normal. Basal ACTH was 6.2 pg/ml (Normal range: 7.2 - 63.3pg/ml), basal cortisol was 5.7 mcg/dl and one hour after synthetic ACTH was 18.7 mcg/dl.

The child was referred to an allergist who introduced other classes of medications, enabling glucocorticoid withdrawal and posterior recovery of growth rate.

Karyotype = 47 XXY



CONCLUSION

The systemic manifestations of prolonged exposure to glucocorticoids are widely reported; however, iatrogenic Cushing Syndrome (CS) is very common. In this case CS was able to mask one of the striking features of the KS carriers, which is high stature.

We observe the delay in the diagnosis of KS and the induction of CS due to poor medical practice. The prescription of glucocorticoids should be judicious: precise indication and for a determined time in prescription, adequate dosage and clear orientation to the patient about the risks of the occurrence of CS with the indiscriminate use.

This case exemplifies how essential it is for every physician to value the clinical method, based on thorough anamnesis and physical examination, aiming for the patient's well being.

REFERENCES

LOS, Evan; FORD, George A.. Klinefelter Syndrome. 2018. Available at: https://www.ncbi.nlm.nih.gov/books/NBK482314/>

GENETICS HOME REFERENCE. U.s National Library Of Medicine. Klinefelter syndrome. 2018. Available at: https://ghr.nlm.nih.gov/condition/

ESTRADA-CHÁVEZ, Guadalupe et al. Cushing Syndrome due to Inappropriate Corticosteroid Topical Treatment of Undiagnosed Scabies. Tropical medicine and infectious disease, v. 3, n. 3, p. 82, 2018.

LODISH, Maya. Cushing's syndrome in childhood: update on genetics, treatment, and outcomes. Current opinion in endocrinology, diabetes, and obesity, v. 22, 2015.