KLINEFELTER SYNDROME WITH SHORT STATURE AND MICROCEPHALY: AN UNUSUAL COMBINATION.

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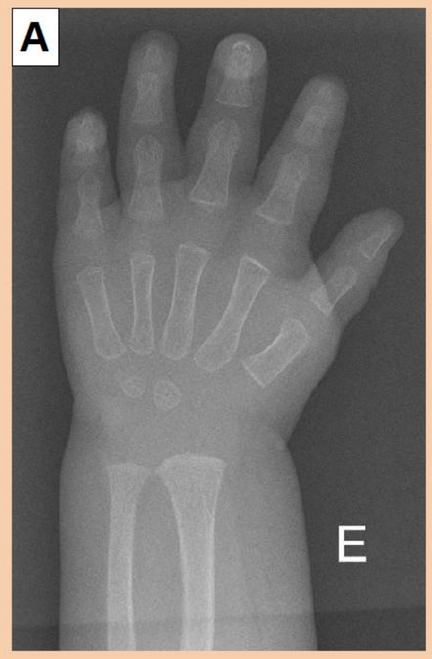
INTRODUCTION

Patients with Klinefelter syndrome (SK) have a 47, XXY karyotype and tall stature as a result of overexpression of the SHOX gene.

CLINICAL DESCRIPTION

A 2-year, 4 month-old boy was referred for study of growth retardation. The product of a first gestation of 39 weeks.
BW: 2.850 g (–1.3 SD), length: 47.5 cm (–1.7 SD), PC: 33 cm (–1.0 SD), of healthy non-consanguineous parents of normal stature.

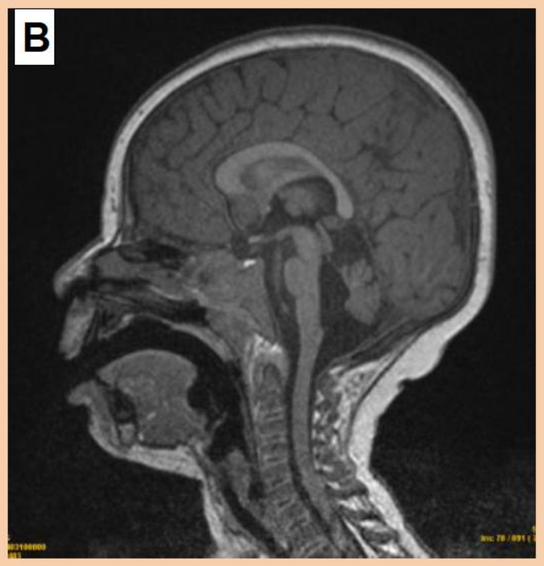
Bone age: 2 years
Fig. A

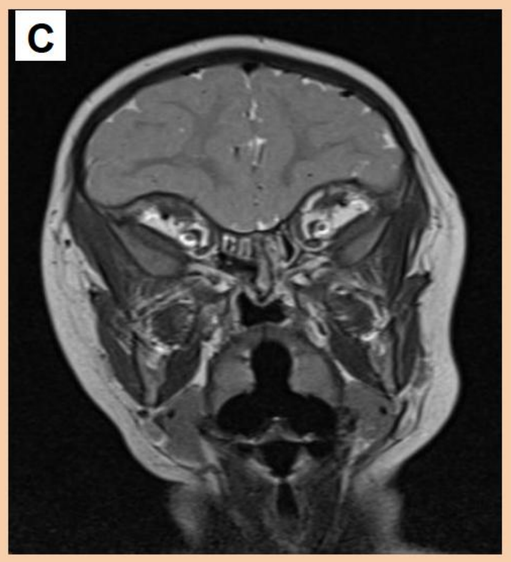


Physical exploration:
Weight: 8.6 kg (–3.0 SD), length: 78 cm (–3.6 SD), CP: 40 cm (–2.0 SD).
Phenotype: peculiar facies with narrow forehead and hypoplasia of nostrils. Microcephaly.
Hypoplastic external genitalia. Testes in 1-cc sacs.
Mild psychomotor retardation and axial hypotonia.

Laboratorial study:
Normal biochemical profile with normal coeliac disease markers.
Hormone study: thyroid hormones: normal, LH <0.07 U/L, FSH 0.61 U/L, inhibin-B 113.0 pg /ml, ACTH 25.9 pg/ml, cortisol 18.6 mcg/dl, prolactin 6.1 ng/ml, IGF-1: 51.4 ng/ml, IGFBP-3: 3.6 ng/ml.
Glucagon test: basal GH 4.4 ng/dl and maximum peak: 7.6 ng/dl.

Brain MRI:
Pituitary hypoplasia, ectasia of the optic nerve sheath, ponto-cerebellar hypoplasia, mild demyelination of white matter.
Fig. B y C

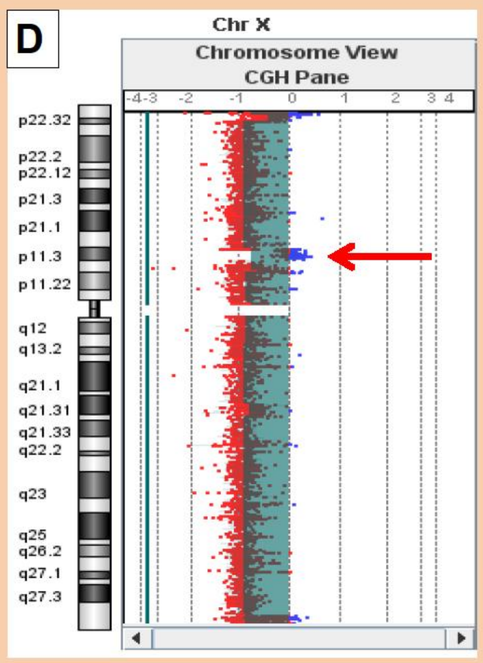


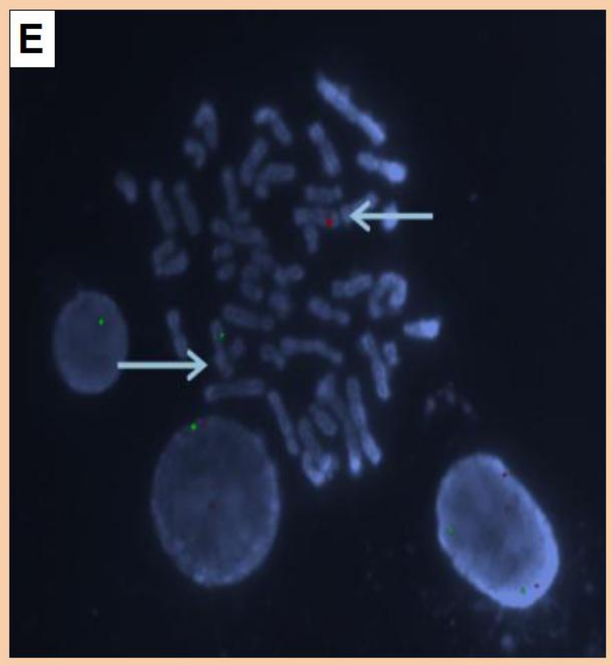


Karyotype: 47, XXY (Klinefelter syndrome).

Array-CGH (ISCA v2, 8x60K, Agilent): presence of two X chromosomes and a deletion in Xp11.3 involving the CASK, NDP, KDM6A, GPR34, GPR82, MAOA, MAOB, EFHC2, FUNDC1 and DUSP21 genes.

FISH study with BAC RP11-24p:
Confirmed the Xp11.3 deletion in the patient and was normal for the parents.
Fig. E





Fundus: retinitis pigmentosa.

Treatment: awaiting start of GH.

CONCLUSION

Array-CGH techniques are useful to orient the diagnosis of patients with severe growth retardation and dysmorphic phenotype. The combined effect of haploinsufficiency of the CASK gene and pituitary hypoplasia may be responsible for the short stature in our patient.

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