Refractory leg ulcers in a patient with 48,XXYY, a rare variant of Klinefelter's syndrome

S. Läuchli, J. Hafner and G. Burg

SUMMARY

A case of a 17-year old boy with extensive ulcers on both lower legs is presented. Clinical examination showed an unusually tall stature, obesity and eunuchoid habitus. Chromosomal analysis revealed a karyotype of 48,XXYY, a rare variant of Klinefelter's syndrome (47,XXY). After treatment with moist gauze and many recurrences, rapid ulcer healing could be achieved with topical application of Factor XIII. Patients with 48,XXYY seem to be particularly prone to developing extensive leg ulcers. Possible etiologies include chronic venous insufficiency and microvascular disturbances as a consequence of fibrinolytic abnormalities. Differences between Klinefelter's syndrome (47,XXY) and 48,XXYY are discussed.

KEY WORDS

male, Factor XIII, fibrinolysis, karyotyping, Klinefelter syndrome, leg ulcers, plasminogen activator inhibitor 1, sex chromosome aberrations, venous insufficiency

Introduction

Klinefelter's syndrome is one of the most common chromosomal variations in humans. Patients with this syndrome have at least one extra X chromosome which results in a karyotype of 47,XXY or, in less common variations, 48,XXXY, 49,XXXXY and XY/XXY mosaic (1). One rare variant is the 48,XXYY syndrome (2). Boys with the latter syndrome are, like Klinefelter patients, usually of a tall stature with a eunuchoid habitus and small testes. There are, however also some important differences: more often than in Klinefelter syndrome, patients with a 48,XXYY karyotype show cognitive or behavioral problems and extensive ulcers of the lower limbs (3). Patients with a female phenotype and extra X chromosomes can have a karyotype of 47,XXX and usually show very little, if any, clinical abnormalities, which might include tall stature and neuromotor developmental delay, but normal sexual development.

Case report

A 17 year-old boy was seen at our clinic for the first time in 1995 with extensive ulcers on both legs, which had persisted in spite of intensive therapy. His family history was unremarkable for genetic diseases. He had just started an apprenticeship as a cook, a job which involved long periods of standing. Initial examination