

## SHORT COMMUNICATION

### DELETION 18p ASSOCIATED WITH A SINGLE MAXILLARY INCISOR: A CASE STUDY

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#### ABSTRACT

We describe a patient with del(18p) syndrome and with a single central maxillary incisor (SCMI). There are few reports on this association. SCMI could be a less severe form of holoprosencephaly in del(18p) syndrome.

#### INTRODUCTION

Single central maxillary incisor (SCMI) can be associated with short stature, insufficient growth hormone or even with normal growth. There are few reports on the association of SCMI with del(18p) syndrome (Dolan *et al.*, 1981; Boudailliez *et al.*, 1983; Aughton *et al.*, 1991). Here we describe a case from Cuba.

#### CLINICAL REPORT

The proband (Figure 1), a female and second child of a 23 year old mother and 25 year old father, was born after 42.3 uncomplicated weeks of gestation. Her data at birth was weight 3040 g, length 49 cm and cephalic circumference (CC) 35 cm, Apgar score of four and eight at one and five minutes, respectively; psychomotor development was retarded. On examination at 39 months, the height was 87 cm (10th centile), weight 13500 g (50th centile) and CC 51 cm (50th centile). There was a dolichocephalic skull with bipatieral bossing, prominent forehead, upslanting palpebral fissures with bilateral epicanthal folds and a low nasal bridge. The nose was small

with short columella and the philtrum long. The patient had a narrow palate with a prominent median palate raphe, single central maxillary incisor and multiple dental caries. The ears were large, protruding, thickened and simply formed. The chest was broad with inverted nipples. Abdominal examination did not show any enlarged organs or masses. The hand had normal palmar creases, absence of distal crease of right fifth digit, clinodactily of both fifth fingers and partial bilateral syndactily of toes 2/3. Neurologic examination was normal. Computed tomography of the cranium was normal; T<sub>3</sub>, T<sub>4</sub>, cortisol, FSH, LH and estradiol performed were normal for the age; lymphocyte chromosomes, studied by G banding had the karyotype 46,XX, del(18p) (pter-p11) (Figure 2); parental karyotypes were normal.

#### DISCUSSION

Six out of 18 cases with SCMI have been found to have chromosome abnormalities, four of them presenting an 18p deletion (Dolan *et al.*, 1981; Boudailliez *et al.*, 1983; Aughton *et al.*, 1991, and this case) and the remaining two a 7q deletion (Masuno *et al.*, 1990). Patients with these chromosome aberrations can also show holoprosencephaly, which occurs with a frequency of about 10% in del(18p) syndrome (Schinzel, 1983). Holoprosencephaly is a malformation sequence with impaired midline cleavage of the embryonic forebrain. It

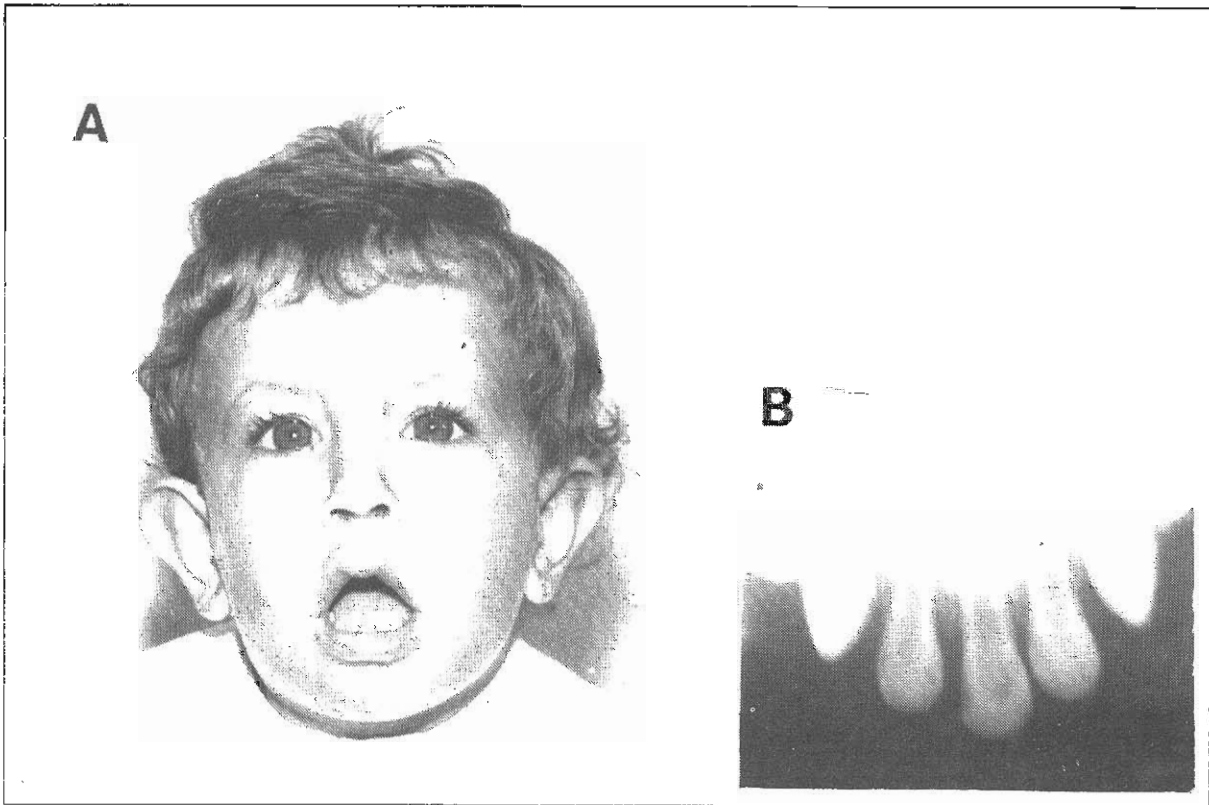


Figure 1 - (a) Patient at 39 months of age. (b) Periapical X-ray showing a single central incisor.

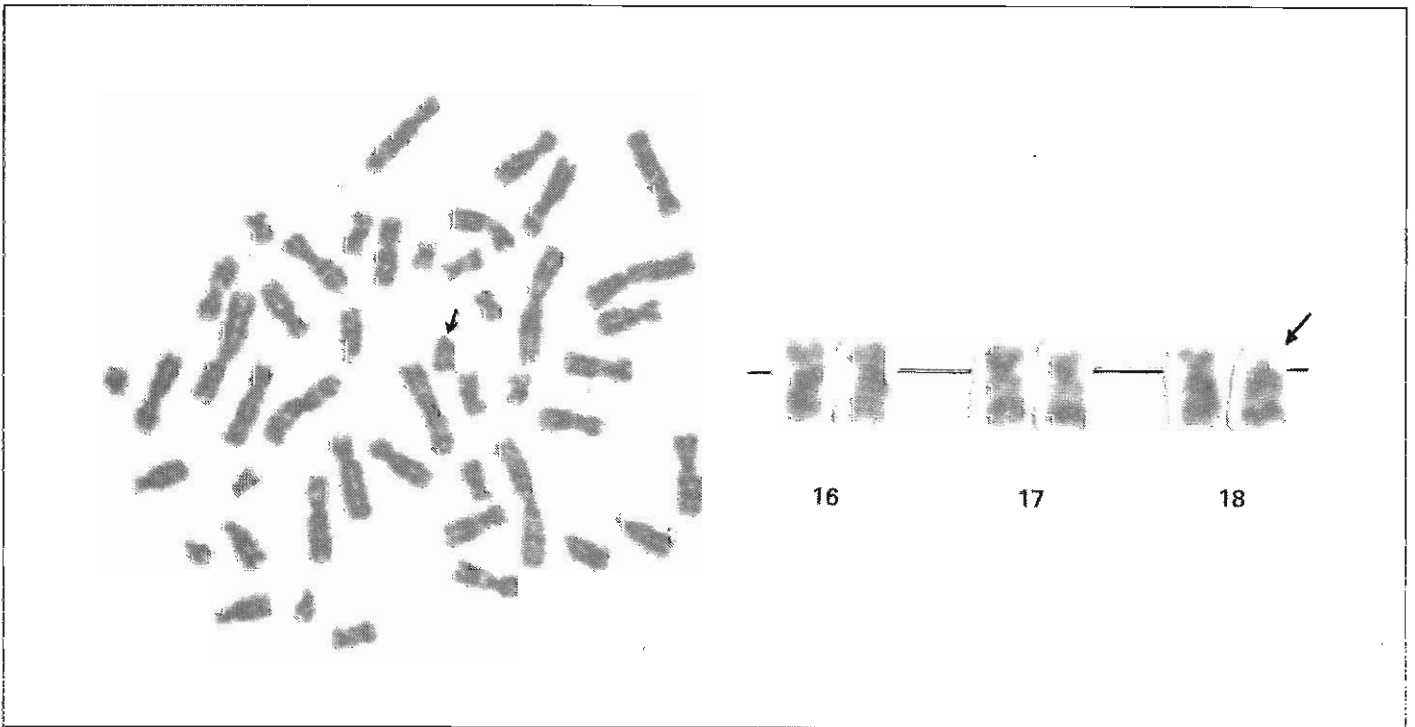


Figure 2 - Partial karyotype showing del(18p), after GTG banding.

is associated with various gradations of mild facial malformation that can vary from more severe forms such as cyclopia, to less severe ones like premaxillary agenesis or even hipotelorism. Because SCMI has been observed in

relatives of individuals with autosomal dominant holoprosencephaly, it has been considered as a less severe form of the spectrum of mild facial malformation that can be seen in holoprosencephaly (Berry *et al.*, 1984).

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## RESUMO

Apresentamos uma paciente com síndrome del(18p), com um único incisivo central superior (UICS). UICS poderia ser uma forma menos grave de holoprosencefalia na síndrome del(18p).

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