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Bloom syndrome. Clinical manifestations and chromosomal study in a Mexican child

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Abstract

Bloom syndrome is an extremely rare inherited disorder. We present a case of Bloom syndrome with a chromosomal study in a Mexican five-year-old patient who presented growth retardation, narrow facies with poikiloderma, café-au-lait, macules and photosensitivity. (Gac Med Mex. 2016;152:747-8)

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We present the case of a 5-year old Mexican female patient, born from non-consanguineous Mexican parents and grandparents at 42 weeks of gestation, with 2,140 g weight, 44 cm height and 29 cm head circumference. She was referred to pediatric endocrinology owing to short stature. Physical exam revealed younger-than-apparent age, short stature (95.5 cm; < 3.9 standard deviations [SD] from the mean), low weight (11.5 kg; < 2.9 SD from mean) and head circumference of 43 cm (< 5.2 SD from mean). She had narrow and elongated facies, short forehead, epicanthus, prominent nose and high-pitched voice (Fig. 1A). Dermatological examination showed multiple café-au-lait oval-shaped maculae, of 5-30 mm in diameter, on the posterior face of the trunk (Fig. 1C), and poikiloderma on the malar region and nasal bridge (Fig. 1B). Genetic assessment found bone age to correspond to chronologic age, and the karyotyping result was 46 XX with a chromosomal spontaneous rupture. Sister chromatid exchange by direct immunofluorescence showed an average higher than 25 exchanges per cell in the 25 analyzed metaphases, which confirmed the Bloom syndrome diagnosis (Fig. 2). The patient has remained under regular surveillance for the development of any malignancy, with preventive measures such as sunscreens and sunlight avoidance.

Bloom syndrome is a rare autosomal recessive disorder characterized by photosensitivity, facial telangiectasias, short stature, different degrees of immune disorders and a high predisposition to several malignancies, associated with underlying chromosomal instability. It was first described in 1954 by David Bloom[1]. It can also be associated with other conditions such as diabetes mellitus, hypogonadism, infertility

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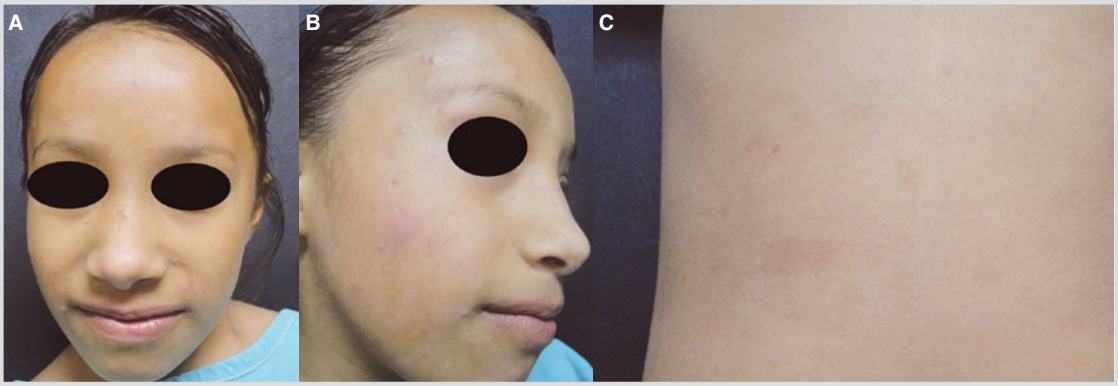

Figure 1. A: *Elongated and narrow facies, short forehead, deep eye implantation and prominent nose.* **B:** *poikiloderma on the cheeks.* **C:** *multiple café-au-lait oval-shaped maculae of 5-30 mm in diameter on the trunk.*

Figure 2. *Sister chromatid exchange test by direct immunofluorescence showing more than 25 exchanges per cell.*

and ocular disorders[2,3]. To date, few cases have been reported in different countries, with one third of cases being found in Ashkenazi Jew population[1].

Sister chromatid exchange test is currently the standard method to confirm the Bloom syndrome diagnosis[4]. Differential diagnoses include Rothmund-Thomson syndrome, erythropoietic protoporphyria and Cockayne syndrome[5].

Early diagnosis is fundamental, since Bloom syndrome has a poor prognosis, with high mortality secondary to malignant tumors. Patient management is usually symptomatic and with a multidisciplinary approach. Close follow-up is important for opportune detection of malignant tumors and infections.

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