ARCHIVOS DE LA SOCIEDAD ESPAÑOLA DE OFTALMOLOGÍA

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Short communication

Cataract in a patient with 47,XYY sex chromosome aneuploidy☆

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ARTICLE INFO

Article history:
Received 21 June 2017
Accepted 9 November 2017
Available online 2 April 2018

Keywords:
Congenital cataract
47,XYY syndrome
High myopia

ABSTRACT

Case report: The case concerns a 16-year-old boy with a history of high myopia and unilateral congenital cataract, tall stature for age, facial dysmorphism, hypermobile metacarpal-phalangeal joints, as well as behavioral problems. The mother had a history of recurrent pregnancy loss. Chromosomal analysis of the peripheral blood lymphocytes reported 47,XYY.

Discussion: Patients with sex chromosome aneuploidy 47,XYY have higher risk of congenital malformations, although ophthalmological anomalies are unusual. Evaluation of patients with tall stature and behavioral problems should include a chromosomal analysis in order to determine the etiology.

Catarata congénita en un paciente con aneuploidía cromosómica 47,XYY

Palabras clave:
Catarata congénita
Síndrome 47,XYY
Miopía alta

RESUMEN

Caso clínico: Paciente masculino de 16 años de edad con antecedente de miopía alta y catarata congénita unilateral, talla alta, dismorfias faciales, hiperlaxitud de falanges y alteraciones de la conducta. La madre tenía antecedente de 3 pérdidas gestacionales. Se realizó cariotipo en sangre periférica reportando 47,XYY.

Discusión: Los pacientes con aneuploidía 47,XYY tienen mayor riesgo de malformaciones congénitas, las alteraciones oftalmológicas no son frecuentes. La evaluación de pacientes con talla alta y alteraciones de la conducta debe incluir cariotipo como parte del abordaje diagnóstico.

☆ Please cite this article as: Medina-Andrade A, Villanueva-Mendoza C, Arenas S, Cortés-González V. Catarata congénita en un paciente con aneuploidía cromosómica 47,XYY. Arch Soc Esp Oftalmol. 2018;93:303–306.

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Introduction

The 47,XYY syndrome is an aneuploidy of the sex chromosomes with an incidence of approximately one case per 1000 live-born males.[1] It is considered an underdiagnosed condition in up to 85% of cases.[2] The clinical characteristics express during the first decade of life, notably developmental delay, behavioral problems and high stature. Motor development may be normal as well as IQ, however they may have learning difficulties, attention deficit hyperactivity disorder (ADHD), autism and aggressive behavior, which has been related to increased brain volume, gray and white matter reduction in the insular region and abnormal development of white matter tracts in the frontal region.[3,4] Published ophthalmological features include the "morning glory" anomaly,[5] iris coloboma and myopia.[6]

Clinical case

Male patient, 16, evaluated in an ophthalmological hospital for presenting antecedents of unilateral congenital cataract in right eye (RE) and high myopia in both eyes.

Antecedents include healthy parents, not consanguineous, with 3 gestational losses during the last quarter of gestation (Fig. 1). During gestation, abortion threats were described without teratogen exposure. The patient was birthed by Caesarean section at 7 months of gestation, presenting fetal distress with meconium aspiration. Development was borderline, with language delay. At age 4, strabismus surgery was performed in RE and at 7 cataracts phacoaspiration surgery was performed with intraocular lens placed in RE, followed by visual rehabilitation for amblyopia.

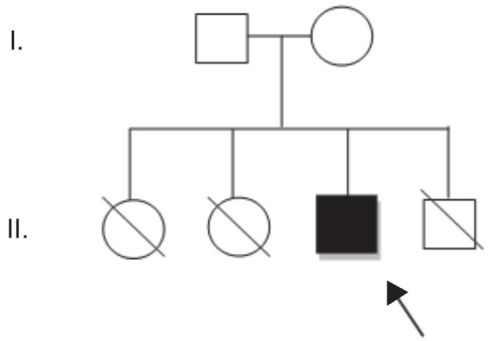

Fig. 1 – Genealogical tree showing a unique case with antecedents of 3 gestational losses in the third quarter.

The patient presents a visual acuity of 20/600 in RE and 20/50 in left eye (LE), with refraction of $-2.25\ -3.00 \times 15^{\circ}$ in RE and $-10.00\ -0.25 \times 45^{\circ}$ in LE and residual esotropia of 20° Hirschberg. In biomicroscopy pseudophakic RE was found, the posterior pole had choroidal fundus in both eyes and the rest of the exploration showed no further alterations. In the physical examination he presented a size above the family average, a relation of arm length/size 1.02; supernumerary nipples, minor facial dysmorphism, phalanges' hypermobility and long fingers (Fig. 2). Parents reported that he exhibited learning problems and challenging social behavior.

Due to the parents' antecedent of gestational losses and the characteristics found, a peripheral blood karyotype was performed, resulting in 47,XYY in 25 metaphases studied with 400–500 bands of resolution. In all the metaphases an extra chromosome of the sexual pair was observed. No structural alterations were detected (Fig. 3).

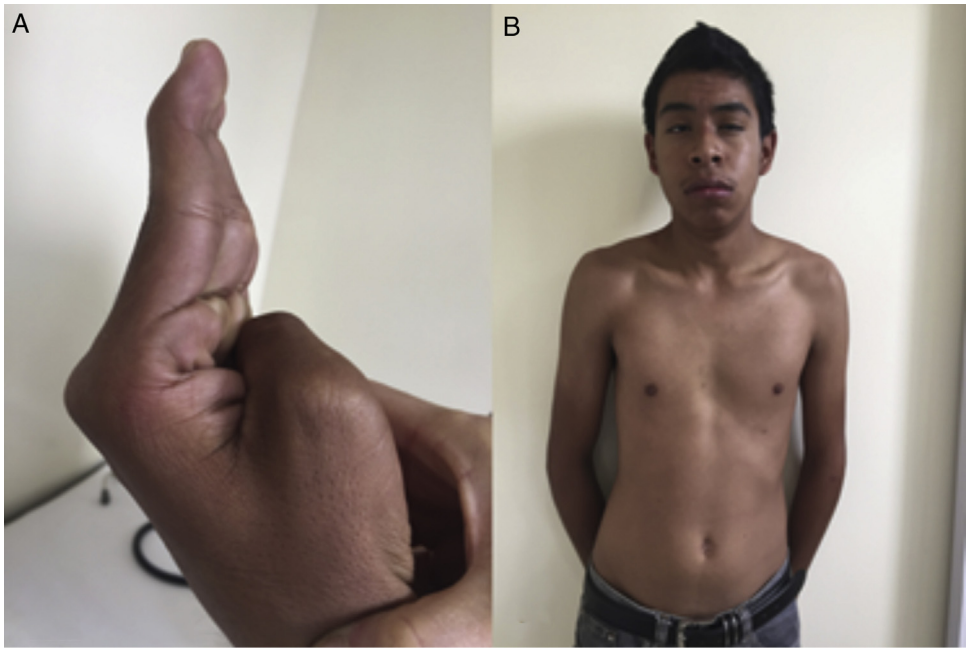

Fig. 2 – Phenotype of the index case. (A) Left hand showing interphalangeal hyperlaxity. (B) Elongated face, broad nose base, thick lips, long-lined thorax.