Proptosis, Micrognathia, Low Set Ear and Chest Deformity in a Patient with Extra Marker Chromosome 22

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Abstract- There is a number of syndromes, associated with proptosis, micrognathia, low-set ear and chest deformity. Herein, we report a 9-year-old female with such phenotype who was presented with a vaginal neuroma. The result of karyotype showed 47XX, with extra marker chromosome 22. Although such a manifestation had not been reported in the literature, it should be considered as a very rare manifestation of the disease.

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Keywords: Cat eye syndrome; Ocular coloboma; Congenital heart defects; Vaginal neuroma.

Introduction

Several syndromic disorders have already been described, associated with proptosis, micrognathia, low-set ear and chest deformity. One of them is Cat eye Syndrome (CES) which is a rare chromosomal disorder, clinically characterized by ocular coloboma, congenital heart defects, and anorectal and urogenital disorders, ears with preauricular tags and/or pits and varying degrees of mental deficiency.

CES (OMIM 115470) is a rare chromosomal disorder, caused by duplication of part of the long arm of chromosome 22, resulting in supernumerary dicentric marker chromosome with satellites at the ends 22 (pter->q11.2:q11.2->pter). The clinical features comprise of ocular coloboma, congenital heart defects, and anorectal and urogenital disorders, ears with preauricular tags and/or pits and varying degrees of mental deficiency (1,2).

Herein, we describe a female child with clinical features of CES presented with vaginal bleeding.

Case Report

The patient was a 9-year-old female, from non-consanguineous and healthy parents with an uneventful pregnancy, who was referred to the Children's Medical Center, the Pediatrics Center of Excellence because of vaginal bleeding since two months ago.

On examination, she had proptosis, micrognathia, low-set ear and chest deformity (Figure 1).

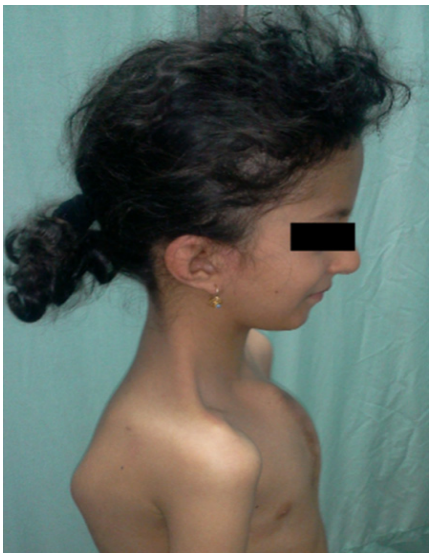

Figure 1. Clinical phenotype of the patient with CES

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The patient had 124 cm height and weighed 21 Kg (<5 percentile).

No sign of puberty was seen. In the past medical history, she underwent open heart surgery due to ventricular septal defect (VSD) and pulmonary stenosis (PS). Abdominal computed tomography (CT), and brain magnetic resonance imaging (MRI) were normal.

Her bone age was 7-year-old according to by the Greulich-Pyle method. The histological examination of vaginal and cervix biopsy revealed disorganized neural bundles with fibrosis consistent with a neuroma.

Considering a syndromic phenotype, karyotype was performed, which showed a female karyotype 47XX, with extra marker chromosome 22, NOR positive at both sides (bisatellites) (Figure 2).

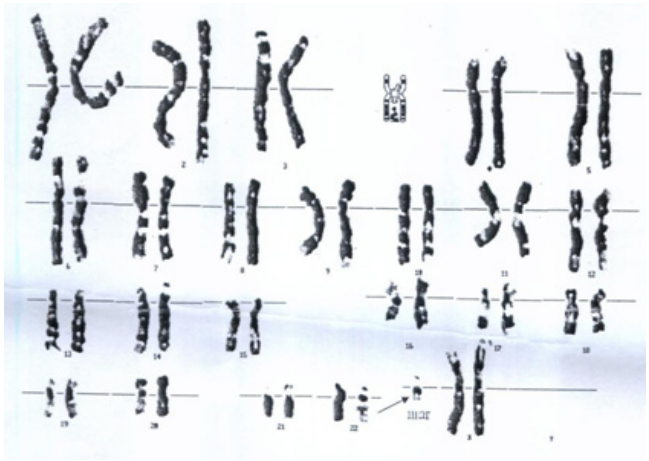

Figure 2. An extra marker chromosome in karyotype

Twenty metaphase spreads were studied on the basis of GTG and NOR binding technique at 500 band resolution

Discussion

CES has wide variable features mainly involve the eye, ear, anorectal, urogenital system, and heart. Minor manifestations include abdominal malformations (*e.g.* biliary atresia), neurologic disorders (*e.g.* dysregulation of mascular tonus), orthopedic malformations (e.g. scoliosis), ocular disturbances apart from coloboma (*e.g.* ocular motility defect) and intellectual disabilities. A Classical feature of CES with biosatellite marker chromosome 22 is manifested in 41% of affected patients (1). The current patient had one feature of CES including congenital heart defects (VSD, PS), but lacking iris coloboma and anal atresia, which make concerns on the definite diagnosis of the case. It should also be noted that to prove that the marker is related to chromosome 22, a FISH probe for that chromosome is needed, which a limitation of this report was.

Some rare and unusual complications have been reported, including anatomical asplenia (3), isolated idiopathic hypogonadotropic hypogonadism (4), hemifacial microsomia (5), schizophrenia (6), and extragonadal mature teratoma (7). Vaginal neuroma has rarely been documented. It may ascribe to trauma and surgery on the vagina (8). Further studies are needed to show if it is a new syndrome or an atypical CES.

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