Lethal multiple pterygium syndrome

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Abstract

The multiple pterygium syndrome is consist of wide range of fetal malformations which have a genetic linkage. A defect in embryonic acetylcholine receptor which can be inherited as autosomal recessive, autosomal dominant, or X-linked fashion is the cause of this syndrome. We present a sporadic case of lethal multiple pterygium syndrome.

Keywords: Cleft palate, club foot, cystic hygroma, Escobar syndrome, multiple pterygium syndrome, pterygium

Introduction

Multiple pterygium syndrome (MPS) is a rare disorder with spectrum of congenital anomalies such as multiple webbing of skin, cystic hygroma, cleft lip/palate, and joint contractures. Its clinical manifestation varies with in utero death in severest form (OMIM 253290). We present a sporadic case of lethal multiple pterygium syndrome.

Case Report

A 26-year-old G4P3L3A0 was referred to our hospital for USG detected multiple fetal malformations at 23 weeks of gestation. The patient had nonconsanguineous marriage. Her blood group was AB−ve, and she had received anti D in all previous deliveries. Per abdominal examination showed an irritable uterus of 28-week size with breech presentation. On p/v, os was closed with no show.

The two-dimensional (2D) ultrasound showed a 24 weeks fetus in breech presentation, with ascites and subcutaneous edema (suggestive of fetal hydrops) [Figure 1a], large multi separte cystic hygroma [Figure 1b], short humerus, bilateral pleural effusion with pulmonary hypoplasia [Figure 1c], and bilateral club foot [Figure 1d].

There were shortening and fusion of lumbar vertebra. The heart was four-chambered with normal outflow track. The amniotic fluid was at increased suggested by the amniotic fluid index of 15 cm, but uterine artery, umbilical artery, and middle cerebral artery Doppler study was normal. The indirect Coombs test was negative, which ruled out Rh isoimmunization.

Within a week, the patient went into spontaneous labor with delivery of 1.4 kg female still born with a large cystic hygroma, depressed nasal bridge, low-set ears with malformation of pinna, cleft lip with partial cleft palate, and bilateral rocker bottom deformity of feet. There were multiple pterygia involving fingers bilateral antecubital, popliteal, and intercrural areas [Figure 2a-d]. The fetus had marked ascitis and edema. The umbilical cord had three vessels, and there were no other anomalies visible. Necropsy and chromosomal studies were refused by the patient due to their cultural belief.

Discussion

The nonlethal form of MPS is called Escobar syndrome (OMIM 265000), which has an autosomal recessive inheritance. The features are pterygia, cardiac defects, pulmonary hypoplasia,
The lethal form of MPS (OMIM 253290) has a wide clinical spectrum of malformations such as webbing of skin (pterygia) of elbow, knee, neck, cystic hygroma, left lip/palate, rocker bottom feet deformity, pulmonary hypoplasia, cryptorchidism, joint contractures, fetal akinesia, cardiac defects, kyphoscoliosis, fetal growth restriction, and intestinal malrotation. The mode of inheritance can be either autosomal recessive, autosomal dominant, or X-linked dominant. The exact etiology is unknown, but the condition has an association with embryonic acetylcholine receptor mutations. The genes of embryonic acetylcholine receptors are CHRNA1 (OMIM 100690), CHRND (OMIM 100720), CHRNA2 (OMIM 100730), RAPSN (OMIM 601592), DOK7 (OMIM 610285), CNTN1 (OMIM 600016), and SYNE1 (OMIM 608441) located on long arm of chromosome 2. The mutations in abovementioned genes are also associated with fetal akinesia deformation sequence, which has an overlapping clinical spectrum with lethal MPS. The early diagnosis of MPS is difficult because increased nuchal translucency and fetal hydrops are the only sonographic features present in the first trimester. In later, half of pregnancy joint contracture develops which reduce the fetal movements. The presence of persistent cystic hygroma, joint contractures, akinesia cardiac defects, kyphoscoliosis, and malrotation of the gut in 2D sonography along with a normal karyotype may suggest toward MPS. The differential diagnosis includes Bartoscas-Papas, Neu-Laxova, and arthrogryposis multiplex congenital. As the diagnosis of MPS is confirmed usually in the later half of pregnancy, the multimodal approach should be taken consisting of obstetrician, radiologist, genetic counselor, and psychological counseling. Genetic counseling is essential for future pregnancies as both lethal and Escobar variant of MPS has autosomal/X-linked inheritance.

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Conflicts of interest
There are no conflicts of interest.

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