Case report

A case of complete tetraploidy in amniotic fluid culture with normal karyotype in the repeat

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Abstract

We report a case of complete tetraploidy in amniotic fluid culture obtained at 14 weeks of pregnancy. Amniocentesis was performed in this pregnancy because of high maternal age and history of offspring with meningomyelocele. Ultrasonography at that time, revealed a single fetus with normal fetal activity and heart beat. Amniotic fluid volume was normal. The amniotic fluid obtained was yellow and clear. It was cultured in two flasks. Growth was very slow in one culture with no growth in the other. Harvest was possible after 3 weeks which revealed tetraploidy in all studied plates. Alpha feto protein of amniotic fluid was 24.1 KIU/m (normal range 11.1-48, for 15 weeks). A repeat culture was performed at 18 weeks of gestation and a FISH analysis was performed using X and Y centromeric probes. Repeat culture revealed 46, XY pattern in 89 out of 90 studied plates. Only one plate revealed tetraploidy. Two hundred interphase cells were studied for the FISH analysis and 98% had one single X and one single Y signal. Ultrasonography at 18 weeks of pregnancy revealed no abnormality. A healthy male infant was born at term and is doing well. We conclude that abnormal karyotypes in poor growth cultures could be misleading and have to be confirmed with repeat cultures.

Key words: Amniocentesis; Tetraploidy; Karyotype

Introduction

Tetraploidy is a relatively common finding in spontaneously aborted fetuses (1). It is however rare in live born infants. The incidence of tetraploidy is 2.0 to 3.2 percent in spontaneous abortions and 5 to 6 percent among abortions with chromosomal abnormalities (2). Thirteen live born infants with tetraploidy have been described in the literature to date (3-6). Anatomical abnormalities most often reported in these infants include positional limb defects, craniofacial abnormalities, and urinary tract abnormalities.

In amniocytes, two types of mosaicism can be

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