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Case Report

Prenatal diagnosis and molecular cytogenetic characterization of mosaicism for a small supernumerary marker chromosome derived from chromosome 11

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ABSTRACT

Objective: We present prenatal diagnosis and molecular cytogenetic characterization of a small supernumerary marker chromosome (sSMC) derived from chromosome 11.

Case report: A 37-year-old, gravida 3, para 2, woman underwent amniocentesis at 17 weeks of gestation because of advanced maternal age. Amniocentesis revealed a karyotype of 47,XX,+mar[18]/46,XX[4]. The parental karyotypes were normal. Level II ultrasound findings were unremarkable. Array comparative genomic hybridization (aCGH) on the DNA extracted from cultured amniocytes revealed no genomic imbalance. The sSMC was characterized by spectral karyotyping (SKY) using 24-color SKY probes and fluorescence *in situ* hybridization (FISH) using a whole chromosome paint (wcp) probe and a CEP11 (D11Z1) probe. The result was 47,XX,+mar.ish(11)(SKY+, wcp11+, D11Z1+)[16]/46,XX[4], indicating that the sSMC was derived from chromosome 11. A healthy female baby was delivered at 37 weeks of gestation with no phenotypic abnormalities. The cord blood had a karyotype of 47,XX,+mar[32]/46,XX[8]. Polymorphic DNA marker analysis of the blood excluded uniparental disomy 11. The female infant was normal in growth and psychomotor development during follow-ups at two months of age.

Conclusion: aCGH, SKY and FISH are useful in prenatal diagnosis of an sSMC derived from the centromeric region of a non-acrocentric chromosome.

Introduction

A small supernumerary marker chromosome (sSMC) has a size equal or smaller than that of chromosome 20 and cannot be identified or characterized its structural abnormalities by conventional cytogenetics [1]. Prenatally ascertained sSMCs occur in 0.075% of the cases at prenatal diagnosis [1–3] and have an overall 13% risk for phenotypic abnormalities [4]. An sSMC derived from a non-acrocentric chromosome has a 28% risk for phenotypic abnormalities comparing with a lower 7% risk in an sSMC derived from an acrocentric chromosome [5]. Liehr and Weise [6] additionally found that a prenatally ascertained sSMC derived from a

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non-acrocentric chromosome carries a 30% risk for phenotypic abnormalities.

We previously reported prenatal diagnosis of sSMC derived from non-acrocentric chromosomes with phenotypic abnormalities [7–9]. Here, we additionally report prenatal diagnosis of an sSMC derived from the centromeric region of chromosome 11 without phenotypic abnormalities.

Case report

A 37-year-old, gravida 3, para 2, woman underwent amniocentesis at 17 weeks of gestation because of advanced maternal age. Cytogenetic analysis of cultured amniocytes revealed mosaicism for an sSMC and a karyotype of 47,XX,+mar[18]/46,XX[4]. Among 22 colonies of cultured amniocytes, 18 colonies had a

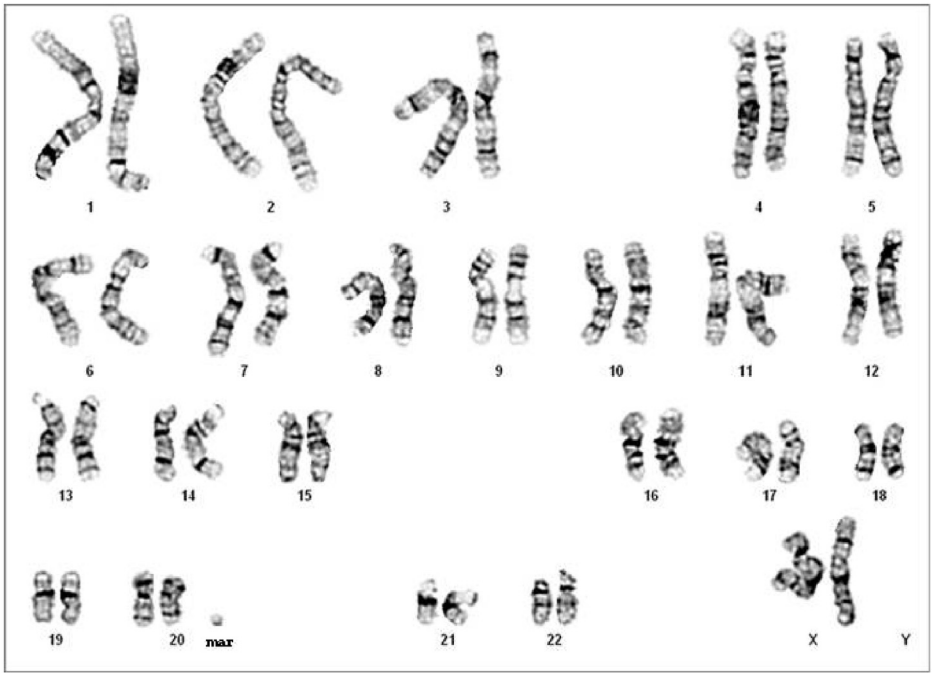

Fig. 1. A karyotype of 47,XX,+mar. mar = marker chromosome.

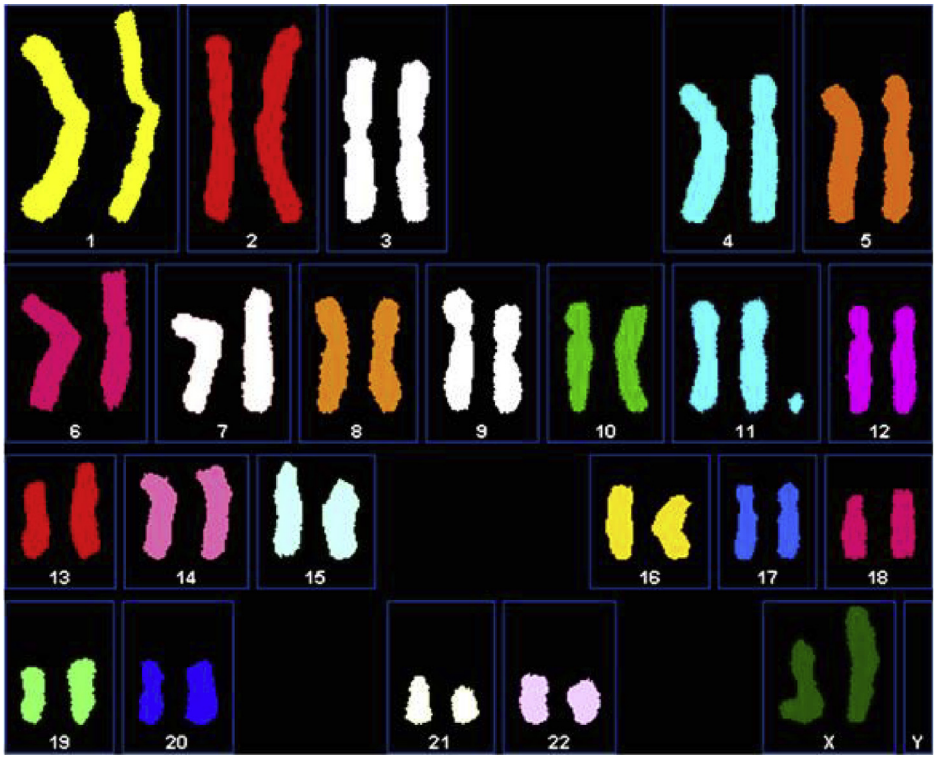

Fig. 2. Spectral karyotyping (SKY) using 24-color SKY probes demonstrates a small supernumerary marker chromosome derived from chromosome 11.