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Short Communication

Molecular cytogenetic characterization of inv dup del(8p) in a fetus associated with ventriculomegaly, hypoplastic left heart, polyhydramnios and intestinal obstruction

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ABSTRACT

Objective: To present molecular cytogenetic characterization of inv dup del(8p) in a fetus with congenital malformations.

Materials and Methods: A 19-year-old, primigravid woman underwent cord blood sampling at 31 weeks of gestation because of prenatal ultrasound findings of polyhydramnios, intestinal obstruction, right ventriculomegaly, and hypoplastic left heart. Preterm precipitous labor and delivery occurred at 32 weeks of gestation. Array comparative genomic hybridization (aCGH), conventional cytogenetic analysis and metaphase fluorescence *in situ* hybridization (FISH) were applied on cord blood lymphocytes. aCGH was also applied on the umbilical cord. Conventional cytogenetic analysis was applied on parental bloods.

Results: aCGH detected an 11.35 Mb deletion in 8p23.3-p23.1 encompassing *SOX7* and *GATA4*, and a 31.99 Mb duplication in 8p23.1-p11.1 in the fetus. Metaphase FISH confirmed inv dup del(8p). The fetus had a karyotype of 46,XX,der(8)del(8)(p23.1) inv dup(8) (p11.1p23.1). Parental karyotypes were normal. A malformed fetus was delivered with facial dysmorphism.

Conclusion: Fetuses with inv dup del(8p) may present central nervous system (CNS) abnormality and congenital heart defect on prenatal ultrasound. Prenatal diagnosis of concomitant CNS and cardiac abnormalities should include a differential diagnosis of chromosome 8p inverted duplication deletion syndrome.

Introduction

The chromosome 8p inverted duplication deletion syndrome caused by inv dup del(8p) is very uncommon and has been found in one in 10,000–30,000 live births [1]. The clinical manifestations of this disorder include mental retardation, facial dysmorphism, central nervous system (CNS) abnormality, hypotonia, orthopedic abnormalities, scoliosis/kyphosis, and congenital heart defects [1–4]. Soler et al [5] first reported prenatal diagnosis of inv dup(8p) with deletion of the distal 8p23 region and duplication of the remaining 8p in a fetus with clubfeet, clenched left hand, subcutaneous edema, and bilateral hydrocephalus. We additionally

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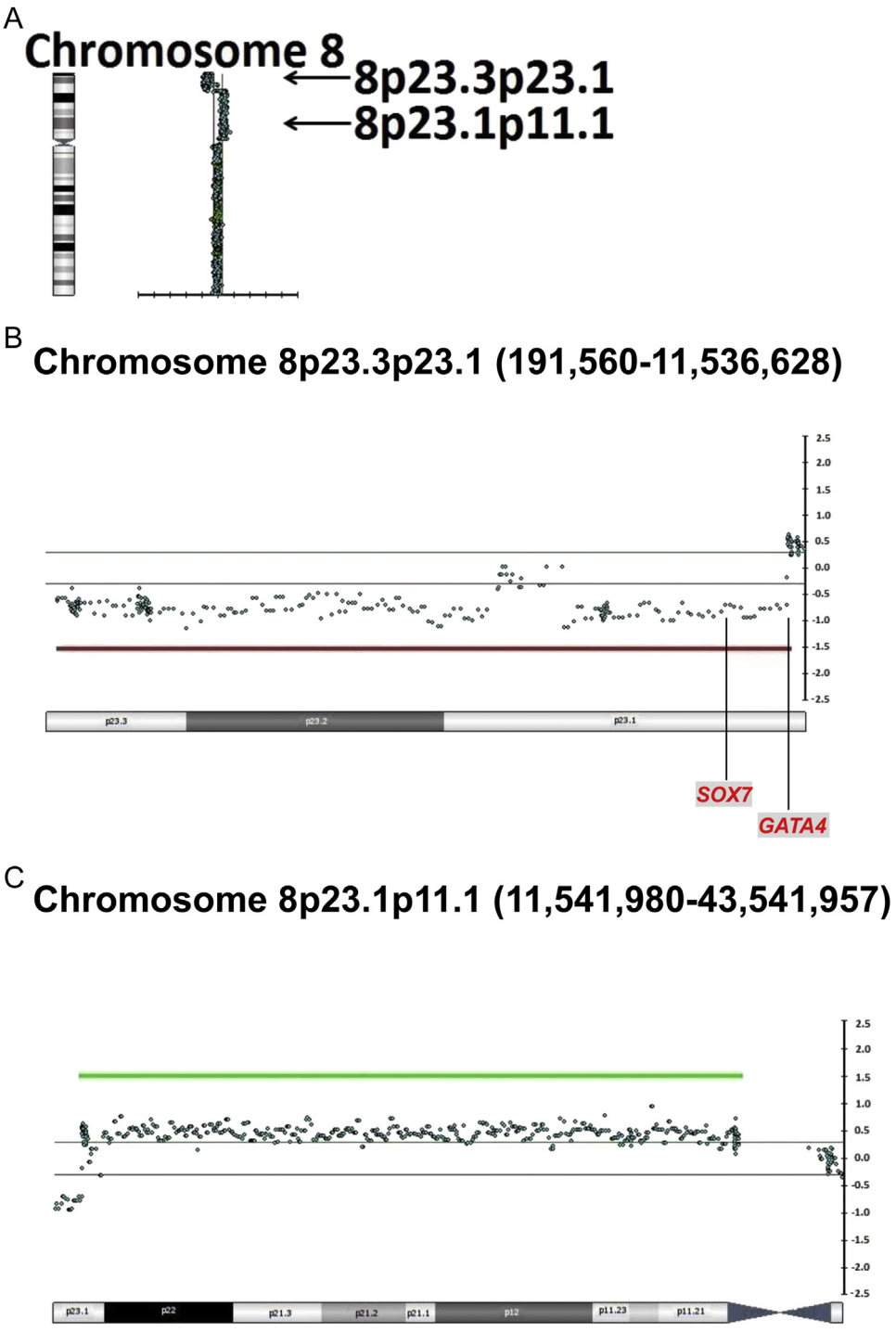

Figure 1. Array comparative genomic hybridization of DNA extracted from the umbilical cord shows (A) an 8p23.3-p23.1 deletion and an 8p23.1p11.1 duplication with (B) an 11.35 Mb deletion in 8p23.3-p23.1 encompassing *SOX7* and *GATA4*; and (C) a 31.99 Mb duplication in 8p23.1-p11.1.

report molecular cytogenetic characterization of inv dup del(8p) in a fetus with ventriculomegaly, hypoplastic left heart, polyhydramnios, and intestinal obstruction.

Materials and methods

Clinical description

This was the first pregnancy of a 19-year-old woman. Her husband was aged 30 years, and there was no family history of congenital malformations. The pregnancy was uneventful until 31 weeks of gestation when right ventriculomegaly, hypoplastic left heart, polyhydramnios, and intestinal obstruction were first noted. She underwent cord blood sampling at 31 weeks of gestation. Preterm precipitous labor and delivery occurred at 32 weeks of gestation. Array comparative genomic hybridization (aCGH), conventional cytogenetic analysis, and metaphase fluorescence *in situ* hybridization (FISH) were applied on cord blood lymphocytes. aCGH was also applied on the umbilical cord. Conventional cytogenetic analysis was applied on parental bloods.