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Case Report

Prenatal diagnosis of partial monosomy 5p (5p15.1→pter) and partial trisomy 7p (7p15.2→pter) associated with cystic hygroma, abnormal skull development, and ventriculomegaly

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ABSTRACT

Objective: Prenatal diagnosis of concomitant chromosome 5p deletion syndrome and chromosome 7p duplication syndrome in a fetus with abnormal prenatal ultrasound is presented.

Case Report: A 34-year-old woman was referred for amniocentesis at 22 weeks of gestation because of an irregular-shaped skull, bilateral ventriculomegaly, and nuchal cystic hygroma. Amniocentesis revealed a derivative chromosome 5 with a distal 5p deletion and an addendum of an extra unknown chromosomal segment at the breakpoint of 5p. Cytogenetic analysis of parental bloods revealed a karyotype of 46, XX, t(5;7)(p15.1;p15.2) in the mother and a karyotype of 46,XY in the father. The karyotype of the fetus was 46, XX, der(5) t(5;7)(p15.1;p15.2)mat consistent with partial monosomy 5p (5p15.1→pter) and partial trisomy 7p (7p15.2→pter). A malformed fetus was subsequently delivered with an irregular-shaped skull, a large anterior fontanelle, brachycephaly, hypertelorism, a high and prominent forehead, a large nuchal cystic hygroma, large low-set ears, a short and flattened nose, and micrognathia. Array comparative genomic hybridization analysis of the placenta revealed the result of arr 5p15.33p15.1 (22,179-18,133,327)×1.0, 7p22.3p15.2 (54,215-25,551,540)×3.0, indicating an 18.11-Mb deletion of 5p (5p15.33-p15.1) and a 22.5-Mb duplication of 7p (7p22.3-p15.2). Cord blood sampling revealed a karyotype of 46, XX, der(5)t(5;7) (p15.1;p15.2)mat.

Conclusion: Fetuses with 5p deletion syndrome and 7p duplication syndrome may present ventriculomegaly, abnormal skull development, and cystic hygroma on prenatal ultrasound.

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Introduction

The chromosome 7p duplication syndrome is characterized by dolichocephaly or brachycephaly, large fontanelles, large low-set malformed ears, hypertelorism, down-slanting palpebral fissures, a high or prominent forehead, a broad or prominent nasal bridge, and micrognathia [1–10]. The critical region for the 7p duplication syndrome has been suggested to be in the segment of 7p21-7p22 [11,12].

The chromosome 5p deletion syndrome, or the cri-du-chat syndrome (OMIM 123450), is characterized by the common features of a high-pitched, monotonous cat cry, microcephaly, broad nasal bridge, epicanthic folds, micrognathia, abnormal dermatographics, growth delay, and psychomotor and mental retardation [13–18]. In the chromosome 5p deletion syndrome, the critical

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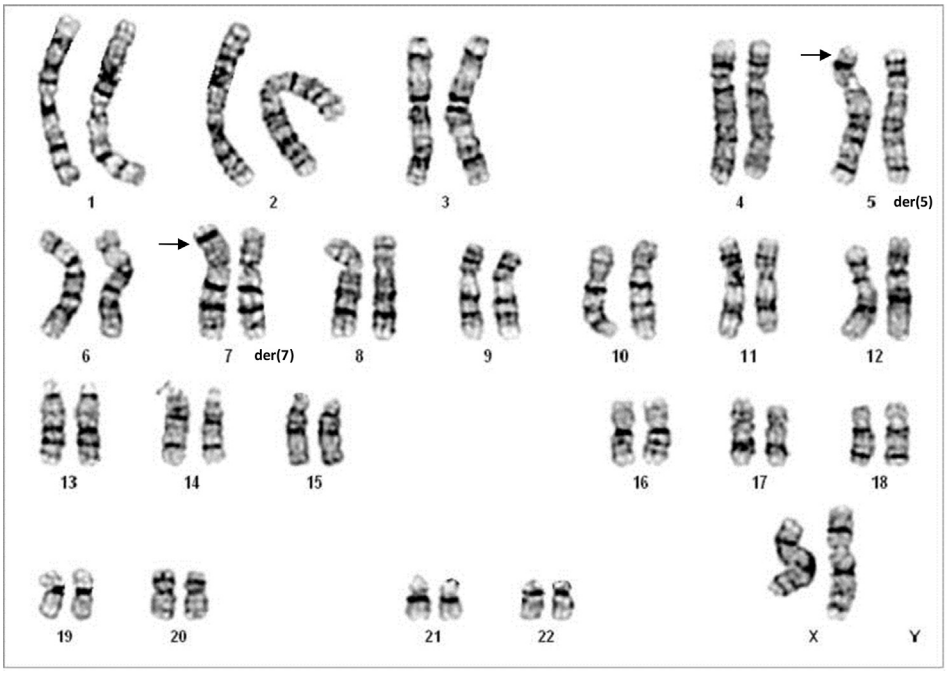

Figure 1. A karyotype of 46,XX,t(5;7)(p15.1;p15.2) in the mother. The arrows indicate the breakpoints. der = derivative.

region for microcephaly has been assigned to be located more proximately to 5p15.31, for dysmorphism and psychomotor retardation in 5p15.2, for cat cry in 5p15.3, and for speech delay in a separate region in 5p15.3 [13,19].

Prenatal diagnosis of concomitant occurrence of chromosome 5p deletion syndrome and chromosome 7p duplication syndrome simply by abnormal fetal ultrasound is very rare. Here, we present such a case with array comparative genomic hybridization (aCGH) characterization.

Case report

A 34-year-old, gravida 2, para 0, woman was referred for amniocentesis at 22 weeks of gestation because of abnormal prenatal ultrasound. The woman had experienced one spontaneous abortion. Her husband was 34 years of age, and there was no family history of congenital malformations. Bilateral ventriculomegaly, an irregular-shaped skull, a nuchal cystic hygroma, and oligohydramnios were noted on fetal ultrasound at 21 weeks of gestation.

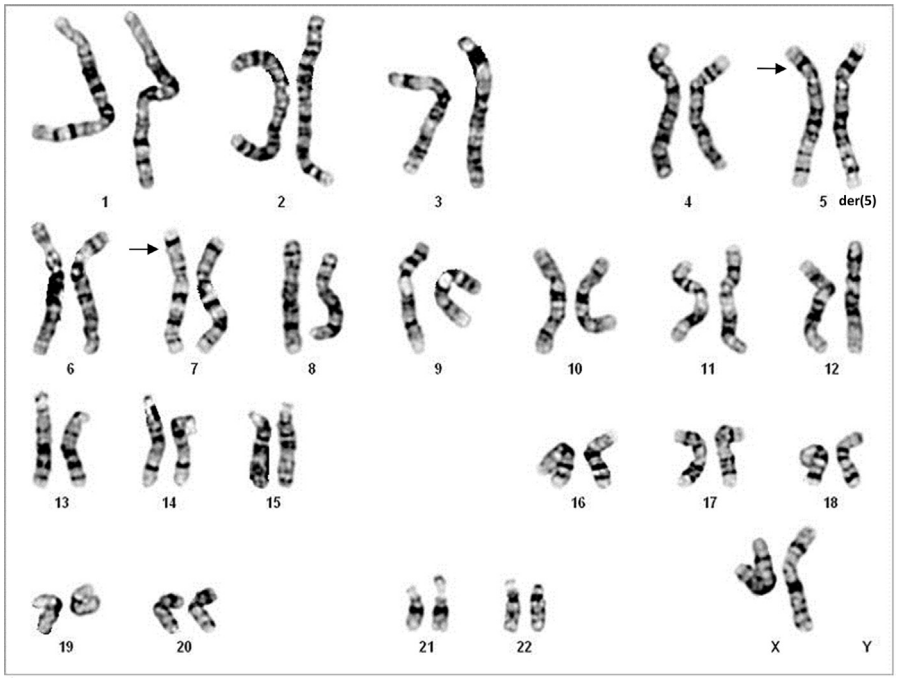

Figure 2. A karyotype of 46,XX,der(5)t(5;7)(p15.1;p15.2) in the fetus. The arrows indicate the breakpoints. der = derivative.

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