A case report of prenatally diagnosed tetrasomy 18p

Phill-Seung Jung¹, Hye-Sung Won¹, In-Ji Cho¹, Min-Kyung Hyun², Jae-Yoon Shim¹, Pil-Ryang Lee¹, Ahm Kim¹
Department of Obstetrics and Gynecology, ¹University of Ulsan College of Medicine, Asan Medical Center, Seoul; ²Inha University Hospital, Inha University College of Medicine, Incheon, Korea

Tetrasomy 18p, one of the most commonly observed isochromosomes, consists of two copies of the p arms on chromosome 18[i(18p)]. It is known as a de novo occurrence of non-disjunction or centromeric mis-division during meiosis II in the vast majority of cases. It has a prevalence of 1/140,000–180,000 live births and affects both genders equally. A 28-year-old woman was referred at 33+2 weeks gestation to rule out fetal congenital heart disease. Her prenatal ultrasonography showed intrauterine growth retardation, cardiomegaly, and imperforate anus. Doppler ultrasonographic finding showed fetal anemia. Tetrasomy 18p was confirmed by conventional karyotyping and fluorescence in situ hybridization. Because of its very low prevalence rate, only several cases of tetrasomy 18p has been reported worldwide and it has not yet been reported in Korea before. Therefore, we report a case of prenatally diagnosed tetrasomy 18p.

Keywords: Isochromosome 18p; Prenatal diagnosis; Tetrasomy 18p

Introduction

Isochromosomes are supernumerary chromosomes that are made up of two copies of the same arm on a chromosome [1-3]. Tetrasomy 18p was first reported by Froland et al. in 1963 [4]. It is a very rare chromosomal anomaly with a prevalence of 1/140,000–180,000 but is also one of the most commonly observed isochromosomes, and affects both genders equally [1-4]. Tetrasomy 18p syndrome is characterized by nonspecific morphologic features; low birth weight, microcephaly, low-set ears, strabismus, abnormalities in muscle tone and deep tendon reflex [1,3,5,6]. Feeding difficulties and developmental retardation are also followed [2,6]. Cardiac and renal malformations are rare, therefore, mortality rate is low [1,5,7]. Because of its very low prevalence rate, tetrasomy 18p has not yet been reported in Korea. Herein, we report the first case of prenatally diagnosed tetrasomy 18p.

Case report

A 28-year-old primi gravid woman was referred to our fetal treatment center because of suspected fetal congenital heart disease at 32+4 weeks of gestation. The ultrasonography showed asymmetric intrauterine growth retardation (IUGR) with 5-week smaller abdominal circumference. The fetal echocardiography demonstrated dextrocardia with cardiomegaly (cardio-thoracic ratio, 0.64), mild pericardial effusion, and decreased left ventricular function (modified myocardial performance index, 0.68). There was no intracardiac abnormality. Doppler findings in the middle cerebral artery revealed increased peak systolic velocity (71 cm/sec, 1.3–1.5 MoM), which suggested fetal anemia. Imperforate anus was also suspected. The cordocentesis was performed at 33+2 weeks of gestation for identifying the karyotype, hemoglobin, and the presence of viral infection. The karyotyping confirmed tetrasomy 18p by G-banding and fluorescence in situ hybridization with 5-week smaller abdominal circumference. The fetal echocardiography demonstrated dextrocardia with cardiomegaly (cardio-thoracic ratio, 0.64), mild pericardial effusion, and decreased left ventricular function (modified myocardial performance index, 0.68). There was no intracardiac abnormality. Doppler findings in the middle cerebral artery revealed increased peak systolic velocity (71 cm/sec, 1.3–1.5 MoM), which suggested fetal anemia. Imperforate anus was also suspected. The cordocentesis was performed at 33+2 weeks of gestation for identifying the karyotype, hemoglobin, and the presence of viral infection. The karyotyping confirmed tetrasomy 18p by G-banding and fluorescence in situ hybridization.
Phill-Seung Jung, et al. A prenatally diagnosed tetrasomy 18p (Fig. 1). Fetal hemoglobin level was 9.7 g/dL and there was no evidence of viral infection such as toxoplasma, rubella, cytomegalovirus, and herpes simplex virus.

The male infant was delivered at 36+6 weeks of gestation; weighing 2,256 g, which was below 10 percentile. Apgar score was 6, 8 at 1, 5 minutes, respectively. Because of low oxygen saturation (76%), the baby was admitted to the neonatal intensive care unit. Initial hemoglobin level was 11.4 g/dL, and after transfusion of packed red blood cells, his oxygen saturation got over 97% without applying oxygen. Low-set ears with small auricles were shown (Fig. 2) and muscle tone was increased. The postnatal echocardiography and cardiac computerized tomography revealed dextrocardia and mild cardiomegaly without pericardial effusion, and the cardiac function was within normal range. As prenatally suspected,