

RING CHROMOSOME 7 PRESENTING WITH INTRAUTERINE GROWTH RESTRICTION AND MULTIPLE ANOMALIES

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SUMMARY

Objective: Ring chromosome 7 is a very rare chromosomal anomaly that may have a grave prognosis. Nevertheless, the clinical features associated with ring chromosome 7 are highly variable. Here, we report a case with ring chromosome 7 and the perinatal findings.

Case Report: A 32-year-old, gravida 1, para 0, woman was referred to our hospital because of intrauterine growth restriction (IUGR) and oligohydramnios at 35 weeks of gestation. Prenatal ultrasound revealed a severe IUGR fetus presenting with multicystic kidney, hydronephrosis and oligohydramnios. At parturition, the birth weight of this male infant was 1,720 g, and a battery of anomalies were also noted, including imperforate anus, hypospadias, micropenis, right cryptorchidism, severe IUGR, multiple nevi on the forehead, shoulder and left thigh, brain atrophy, right multicystic kidney, and left mild hydronephrosis. Cytogenetic study from cord blood revealed a ring chromosome 7.

Conclusion: Ring chromosome 7 is extremely rare and our case might be the 15th and youngest case in the medical literature. Our case had multicystic kidney and imperforate anus, which have not been reported previously. Prenatal diagnosis of ring chromosome 7 is very difficult. When fetuses present with severe IUGR, oligohydramnios and multicystic kidney, chromosomal aberrations should be kept in mind, and perinatal cytogenetic workup is warranted. [*Taiwanese J Obstet Gynecol* 2005;44(3):297-299]

Key Words: ring chromosome 7, intrauterine growth restriction, multicystic dysplastic kidney

Introduction

Ring chromosome 7 is a rare chromosomal anomaly. From the first description in 1973 [1], only 14 cases have been reported in the medical literature [1-13]. Clinical findings associated with ring chromosome 7 are highly variable; most reported cases have growth and mental retardation, microcephaly, and dermatologic abnormalities including nevus flammeus, café-au-lait spots, and dark pigmented nevi. The variation in pheno-

typic effects in different patients suggests different causal mechanisms that could include size variations in chromosomal deletions or abnormal chromosomal behavior such as ring instability leading to mosaicism in different tissues [10]. We report a case with ring chromosome 7 that presented with severe intrauterine growth restriction (IUGR), hydronephrosis with multicystic dysplastic kidney (MCDK), hydronephrosis, and oligohydramnios when referred to our unit at 35 weeks of gestation. Perinatal cord blood cytogenetic study confirmed a ring chromosome 7. Our case might be the 15th and the youngest case of ring chromosome 7 in the medical literature.

Case Report

A 32-year-old, gravida 1, para 0, woman was referred to our hospital at 35 weeks of gestation because of IUGR

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Figure 1. Prenatal ultrasound reveals right multicystic dysplastic kidney at 35 weeks of gestation.

and oligohydramnios. According to the local practitioner’s description, the fetus presented with IUGR from 22 weeks of gestation; otherwise, prenatal findings were negative. Her history and family history were both unremarkable.

In our hospital, prenatal level II ultrasound showed right MCDK (Figure 1) with hydronephrosis. Severe IUGR was also noted; the biparietal diameter was 7.5 cm (30.1 weeks), abdominal circumference was 28.4 cm (34.1 weeks), femur length was 5.5 cm (30.5 weeks), and the estimated fetal weight was 1,750 g (31.6 weeks). The amniotic fluid index was 2.7 cm, indicating oligohydramnios.

One week later (36 weeks’ gestation), severe variable deceleration was noted during a routine non-stress test. Therefore, emergency cesarean section was undertaken

to deliver an asphyxic IUGR male baby with birth weight 1,720 g and Apgar scores at 1 and 5 minutes of 6 and 7, respectively, after neonatal resuscitation. A battery of anomalies were noted, including imperforate anus, hypospadias, micropenis, right cryptorchidism, severe IUGR, and multiple nevi on the forehead, shoulder, and left thigh.

Under the suspicion of chromosomal anomaly, a cytogenetic study from cord blood was undertaken, which revealed a ring arising from chromosome 7 (Figure 2). The ring chromosome 7 was formed by deletion of 7p22 and 7q36 with subsequent fusion of the 7p and 7q ends. Brain atrophy, heart ventricular septum defect, right MCDK, and left hydronephrosis were also detected in the neonatal intensive care unit.

Discussion

Ring chromosome 7 is very rare; chromosome analysis of consecutive newborns found it in one case out of 46,000 [11]. To the best of our knowledge, only 14 cases with a ring chromosome 7 have been published in the medical literature since 1973 [1–13], and no case has been reported in Taiwan.

Clinical findings of patients with a ring chromosome 7 are variable but most share some findings, such as mental retardation, growth deficiency, microcephaly, facial asymmetry, hypertelorism, abnormal palpebral fissures, small ears, limb and skeletal anomalies, and skin lesions including nevus flammeus, dark pigmented nevi, and café-au-lait spots [12]. A summary of the main clinical features of these cases is given in the Table. Our case has anomalies in common with those previously described, such as growth restriction, sacrum hypoplasia, brain atrophy, and multiple skin lesions. However, there were some novel anomalies that have not been reported before, including MCDK and imperforate anus.

The definite mechanism of the pathogenesis of ring chromosome 7 is still unknown. The most common theory is as follows: when a ring is formed, it requires two breaks and a union of the ends of the chromosome. The clinical consequences of such a ring depends on several factors, including the loss of small fragments of chromosomes distal from breaks. Large parts of a chromosome probably contain more genes and, consequently, their loss leads to more severe clinical findings than loss of smaller parts.

However, the genetic information in the chromosome material lost is also important [11]. In chromosomal abnormalities with unusual clinical manifestations, molecular cytogenetic studies could help explain the presence of such clinical features [12]. For example, a

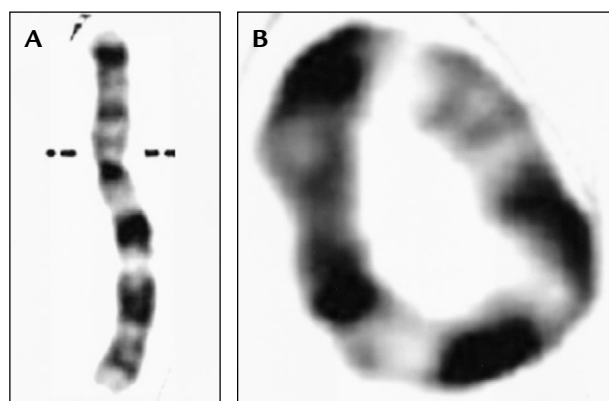


Figure 2. (A) Normal chromosome 7. (B) Ring chromosome 7.

Table. Summary of clinical features from reported cases with ring chromosome 7

Case	1 [1]	2 [1]	3 [2]	4 [3]	5 [4]	6 [5]	7 [6]	8 [7]	9 [8]	10 [9]	11 [10]	12 [11]	13 [12]	14 [13]	This case
Gender	M	M	M	F	M	F	M	M	M	M	M	M	F	M	M
Birth weight (g)	2,800	2,200	1,180	2,650	2,350	2,750	3,270	2,000	2,460	2,400	2,140	3,200	2,400	2,400	1,720
Age at detection	23 mo	16 mo	19 yr	8.8 yr	14 mo	9 mo	13 yr	39 yr	3 d	2 mo	5 mo	8 yr	1.7 yr	20 mo	Birth
Maternal/paternal age (yr)	35/40	32/34	29/32	28/34	29/38	30/33	29/26	30/32	26/28	35/35	/	/	32/33	27/27	32/33
Growth retardation	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
Brain anomaly	/	/	/	/	/	/	/	/	/	+	+	/	/	/	+
Ocular anomaly	+	-	+	-	-	-	-	+	-	+	+	-	-	-	-
Bone anomaly	/	+	+	+	+	/	+	+	/	/	/	/	+	+	+
Heart anomaly	/	/	/	/	/	/	/	/	+	-	-	/	/	/	+
Genital anomaly	+	-	-	/	/	-	-	+	+	+	+	-	/	-	+
Skin lesion	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
MCDK	/	/	/	/	/	/	/	/	/	/	/	/	/	/	+
Imperforate anus	/	/	/	/	/	/	/	/	/	/	/	/	/	/	+

M = male; F = female; + = present; - = absent; / = not reported; MCDK = multicystic dysplastic kidney.

region with genes involved in sacral agenesis was mapped at the end of 7q (7q36) [14]. According to Sawyer et al, the deletion of 7q36 is associated with holoprosencephaly [10]. Although our case had the same deletion at 7q36, the infant did not have holoprosencephaly. Different genes may be involved in developing the different anomalies in Sawyer et al's case and ours. Further molecular cytogenetic studies are indicated.

In conclusion, ring chromosome 7 is extremely rare and our case might be the 15th and youngest case in the medical literature. In addition, our case had MCDK and imperforate anus, which have not been previously reported. Prenatal diagnosis of ring chromosome 7 is very difficult. Chromosomal anomaly should be highly suspected when IUGR and multiple congenital anomalies are detected. Prenatal or perinatal cytogenetic study is warranted.

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