Case Report

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An unusual *de novo* duplication 10p/deletion 10q syndrome: The first case in Korea

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We herein report an analysis of a female baby with a *de novo* dup(10p)/del(10q) chromosomal aberration. A prenatal cytogenetic analysis was performed owing to abnormal ultrasound findings including a choroid plexus cyst, prominent cisterna magna, and a slightly medially displaced stomach. The fetal karyotype showed additional material attached to the terminal region of chromosome 10q. Parental karyotypes were both normal. At birth, the baby showed hypotonia, upslanting palpebral fissures, a nodular back mass, respiratory distress, neonatal jaundice and a suspicious polycystic kidney. We ascertained that the karyotype of the baby was 46,XX,der(10)(pter→q26.3::p11.2→pter) by cytogenetic and molecular cytogenetic analyses including high resolution GTG-and RBG-banding, fluorescence *in situ* hybridization, comparative genomic hybridization, and short tandem repeat marker analyses. While almost all reported cases of 10p duplication originated from one of the parents with a pericentric inversion, our case is extraordinarily rare as the *de novo* dup(10p)/del(10q) presumably originated from a rearrangement at the premeiotic stage of the parental germ cell or from parental germline mosaicism.

Key words: Chromosome 10, trisomy 10p, Chromosome 10, monosomy 10q.

Introduction

Dup(10p)/del(10q) aberrations are usually generated from recombination between a parental pericentric inverted chromatid and its normal homologue during meiosis I. Cases of dup(10p) with small del(10q), with respect to clinical phenotypes, are regarded as trisomy 10p syndrome because they involve a large segment of chromosome 10p [1-3].

Most previous trisomy 10p cases originated from familial reciprocal translocations or familial pericentric inversions and *de novo* trisomy 10 is very rare [4-6]. The common clinical features of trisomy 10p are severe mental and growth retardation, craniofacial anomalies (high forehead, hypertelorism, upward slant of the palpebral fissures, and cleft lip and palate), and other organ malformations (cardiac and renal anomaly) and skeletal abnormalities (clubfoot and flexion abnormalities).

Since Lewandowsky et al. [7] reported the deletion in the long arm of chromosome 10, over 60 cases of interstitial or terminal 10q deletion ranging from 10q23-q26 resulting from familial translocations or other chromosomal anomalies have been identified. The main clinical manifestations of terminal deletion of 10q syndrome are typical facial dysmorphisms, mental and

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developmental retardation, postnatal growth retardation, anogenital anomalies, limb abnormalities or cardiac/renal anomalies.

Fourteen cases of rec(10), duplication 10p accompanied with deletion 10q, have been reported previously, and all were derived from a parental pericentric inversion.

Here, we report an exceptional *de novo* dup(10p)/del(10q) case characterized by high resolution (HR) GTG- and RBG-banding, fluorescence *in situ* hybridization (FISH), comparative genomic hybridization (CGH) techniques, and short tandem repeat (STR) marker analyses; a few common phenotypic features were observed based on a review of previous cases of rec(10). This study was approved by the Ethics Committee of Cheil General Hospital and Women's Health Care Center (#CGH-IRBGR-2007-02) and patient's consent was obtained.

Case

1. Case description

A 24-year-old, mother (G1 P0) was referred for cordocentesis owing to abnormal ultrasound findings including right choroid plexus cysts, prominent cisterna magna, slightly medially displaced stomach, and renal pelvis dilation at 26 weeks of gestation. After genetic counseling, parental chromosome analyses were also carried out. A female baby was born at 38.2 weeks of gestation by normal vaginal delivery. The birth weight was 2,910 g (50th percentile), length was 47 cm (25th percentile), and head circumference was 33 cm (25-50th percentile). The Apgar scores were 6 and 7 at 1 and 5 min, respectively. Four days after delivery, she was transferred to the newborn intensive care unit owing to hypotonia with decreased muscle tone and moderate feeding difficulties. General activity was good and neck and clavicles were normal. The urogenital tract and anus were normal, and extremity and motor activity was good. However, the newborn baby had a 1-cm nodular back mass, up-slanting palpebral fissures, asymmetric head shape, and suspicious multiple renal cortical cysts in both kidneys. In addition, the baby exhibited respiratory distress, neonatal jaundice, unspecified thrombocytopenia, and direct bilirubinemia. On day 25, the baby was discharged from the hospital.

2. Cytogenetic and CGH analysis

Conventional GTG-banding analyses were carried out on cultured lymphocytes obtained from fetal cord blood and parental peripheral blood samples. After birth, HR GTG- and RBG-banding techniques were performed to determine the precise breakpoints of the abnormal chromosome.

The genomic DNAs for CGH analyses were extracted from peripheral blood samples of the infant and from a karyotypically normal female for use as a control (reference DNA). CGH procedures were performed as described by Kallioniemi et al. [8] and the manufacturer's instructions (Vysis Inc., Downers Grove, IL, USA). CGH signals were captured (ZEISS, Oberkochen, Germany) and analyzed by the CytoVision (version 3.52; Applied Imaging, Southend-on-Sea, UK).

All 30 metaphase chromosomes from the fetal cord blood sample indicated that the fetus had extra chromosomal material at the long arm of one chromosome 10. Initially, we reported the fetal karyotype as 46,XX,add(10)(q?25). The results of the parental chromosome study were both normal indicating the *de novo* origin of the abnormal fetal chromosome 10. After birth, we reexamined the newborn's chromosome obtained from the lymphocyte culture using HR GTG- and RBG-banding

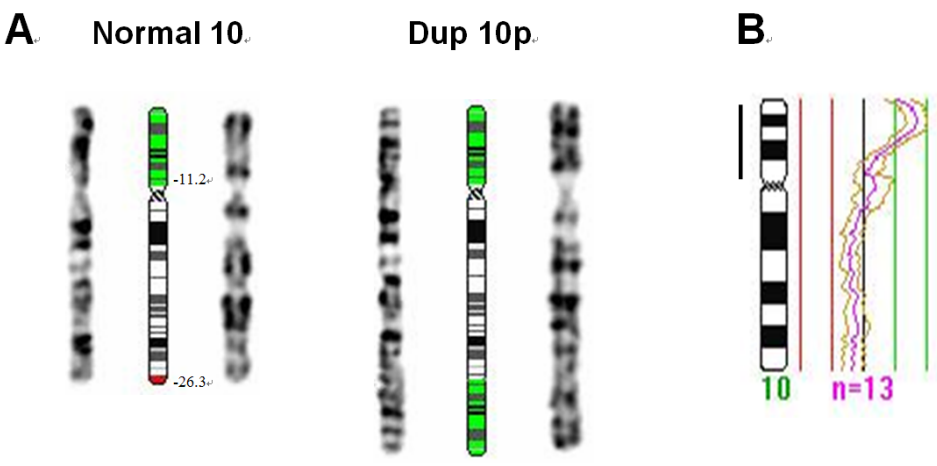

Fig. 1. Partial karyotype and ideogram and comparative genomic hybridization (CGH) analysis. (A) GTG- (left) and RBG- (right) banded partial karyotype and ideogram (middle) of the duplicated chromosome 10 of the infant showing the short arm of chromosome 10 attached to the terminal region of the long arm of chromosome 10. 46,XX,der(10)(pter→q26.3::p11.2→pter). (B) CGH analysis. Partial profile of dup(10p) shows amplification of the signal at the chromosome 10pter→p11.2 segment (black bar).

techniques, and revealed that the chromosome 10p was duplicated (Fig. 1A).

The CGH profile showed prominent amplification of the signal in the region of 10p11.2→15.3. However, signal loss in the region of 10q26.3 was not detected (Fig. 1B). The deleted region was likely too small to detect by CGH. After collecting the above data, we defined the newborn's karyotype as 46,XX,der(10)(pter→q26.3::p11.2→pter).

3. FISH analysis

FISH studies on the metaphase chromosomes obtained from the newborn's lymphocyte culture were carried out to identify both ends of chromosome 10. Two probes for the subtelomeric region of 10p (TelVysion 10p Spectrum Green; Vysis Inc.) and 10q (TEL 10q DNA Probe Red; Qbiogene, Amsterdam, The Netherlands) were used according to the manufacturer's instructions. The FISH images were observed under an Axio Imager Z1 (ZEISS) and captured using Isis software (ver. 5.1.2; MetaSystems, Altlussheim, Germany).

FISH studies with the probe for subtelomeric regions of 10p and 10q showed two signals for 10p in both ends of the abnormal chromosome 10 (duplication of 10p) and loss of signal for 10q (deletion of 10q) (Fig. 2).

4. STR marker analysis

STR marker analyses were performed to examine the parental origin of the duplicated region on the infant's chromosome 10p by using 10 STR marker loci. The highly heterozygotic STR markers D10S526, D10S1145, D10S518, D10S504, D10S1154, D10S1142, D10S514, D10S1168, D10S527, and D10S2325 were selected residing from 10p15 through 10p12, from the Ensemble Genome Browser (http://www.ensemble.org) and the National Center for Biotechnology Information (NCBI) genome database (http://www.ncbi.nlm.nih.gov). A 20 ng sample of DNA from each family member was amplified in a 15 μL reaction containing the following: 10 mM Tris-HCl, 50 mM KCl, 1.5 mM $MgCl_2$, 10 mM dNTP (ABI, Walthanm, MA, USA), 0.2 U GoldTaq polymerase (Perkin Elmer, Walthanm, MA, USA), and 0.67 pmol each primer. Amplification was performed with the following conditions: 5 min at 95°C, 35 cycles of 30 s of denaturation at 95°C, 20 s of annealing at 58°C, and 40 s of extension at 72°C; and a 7 min final extension at 72°C. Polymerase chain reaction products were electrophoresed through a 7% polyacrylamide gel in 0.5× TBE buffer and the gel was stained in ethidium bromide (1 mg/mL) for 20 min. A photoimage was captured by using BiPS 3.0 (Biomedlab, Seoul, Korea).

Parental origin of abnormal 10p was confirmed by using

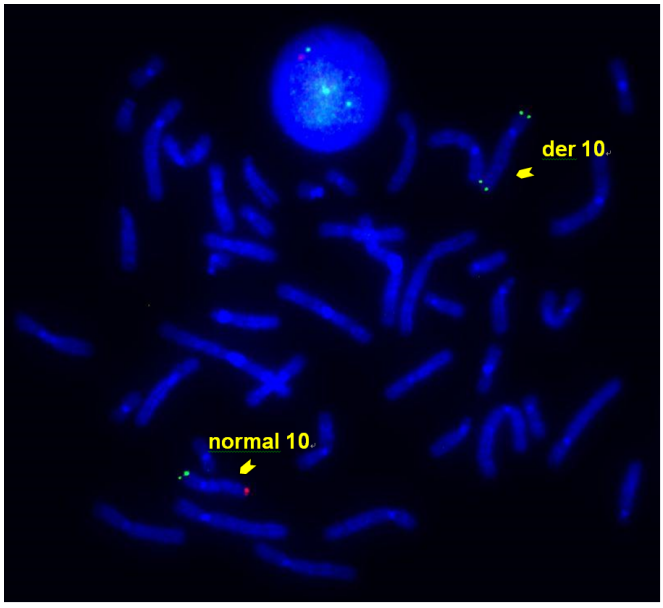

Fig. 2. Fluorescence *in situ* hybridization (FISH) analysis. The deletion of the terminal region of chromosome 10q (spectrum orange signal) and the duplicated signals for the terminal region of chromosome 10p on both ends of derivative (der) chromosome 10 (spectrum green signal).

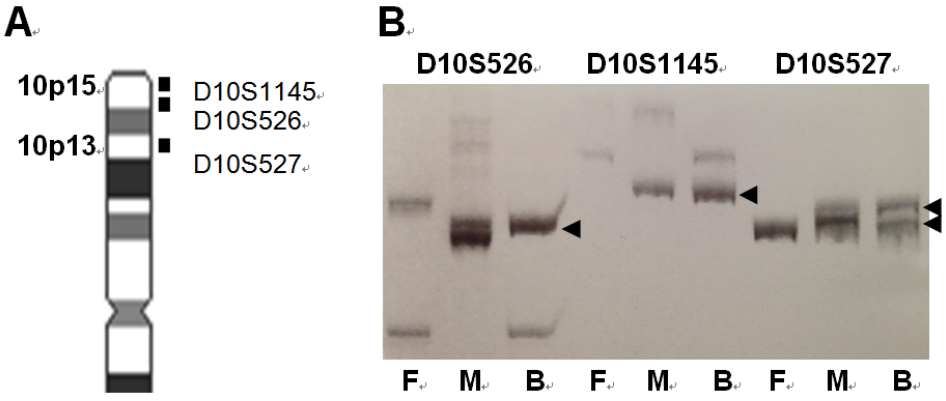

Fig. 3. (A) An ideogram of chromosome 10 depicting three informative short tandem repeat (STR) markers of the selected markers residing on the short arm of chromosome 10. (B) Polymerase chain reaction products of STR marker analysis for the parents and newborn were electrophoresed on a 7% polyacrylamide gel. For D10S526 and D10S1145, the newborn's bands from the maternal allele are more intense (arrowhead) than bands from the paternal allele. For D10S527, the newborn's bands are from maternal alleles (arrowheads). F, father; M, mother; B, baby.

STR markers between 10p15 and 10p12. Of them, three tetranucleotide markers, D10S526 (10p15), D10S1145 (10p15) and D10S527 (10p13), were informative alleles (Fig. 3A). Each maternally derived allele for the STR markers D10S526 and D10S1145 had higher intensely than that of the paternally derived alleles. For D10S527, the newborn's bands were both from maternal alleles. This indicated that the *de novo* duplicated 10p segment from 10p15-p13 was from the maternal allele (Fig. 3B).

Discussion

The *de novo* chromosomal aberration in this case showed a typical recombinant chromosome comprising a duplication of a large segment of 10p and terminal deletion of 10q. Although soem STR markers showed informative results and the region might have additional segmental crossing-over, the duplicated segment of at least the 10p15-p13 region appears to be derived from the maternal chromosome.

A duplication-deletion chromosome is usually assumed to be the product of crossing-over between an inherited pericentric inverted chromosome and its normal homologue during meiosis I. We reviewed all 14 cases of dup(10)/del(10) cases reported previously and summarized the cytogenetic and clinical findings of those and the present case (Table 1) [1-3,9-15]. Based on STR marker analyses, the *de novo* duplicated chromosomal segment of this case was of maternal origin. However, we could not determine whether the duplication-deletion chromosome was generated from successive rearrangement events or a distinctive independent rearrangement in a maternal gamete. We assumed two possible mechanisms for the present case: 1) a pericentric inversion and recombinant homologous might have occurred by successive cross-over at maternal meiosis I during gametogenesis, or 2) the mother may have a premeiotic pericentric inversion by maternal germline mosaicism and duplication-deletion rearrangement might have been generated by cross-over at maternal meiosis I during gametogenesis. In cases involving other autosomes, there were two reports of *de novo* recombinant duplication derived from premeiotic pericentric inversion on chromosome 1 and 13, respectively [16,17].

From a clinical perspective, the dup(10p)/del(10q) syndrome was characterized by hypotonia, poor feeding, moderate to severe mental retardation, microcephaly, wide sutures, frontal bossing, short nose, cleft lip and palate, camptodactyly, clinodactyly, club feet, and urogenital system [3]. According to the size of the duplicated or deleted segment, dup(10p)/del(10q) cases share similar clinical manifestation with trisomy 10p or deletion 10q syndrome (Table 2) [3]. The infant we described had a few common clinical anomalies of dup(10p) and del(10q) that are parallel to those of other reported cases with similar break points [1,3,9]. Deletion of 10q26 is more common compared to del(10)(q25), but demonstrates a less severe phenotype. Winsor et al. [18] and Yunis and Torres de Caballero [10] suggested that the distance between the break points and the centromere determine the lengths of the duplication or deletion and affect the viability of embryos. Additionally, genital anomalies in males, such as cryptorchidism are a highly frequent phenotype associated with the deletion 10q26. Chung et al. [19] and Tanabe et al. [20] suggested that genes in the region of 10q25 or 10q26 are related to the regulation of the testis-determining gene, *SRY*, or male sex differentiation.

Typical clinical manifestations are difficult to discriminate among syndromes of trisomy 10p, deletion 10q, and dup(10p)/del(10q). Upslanting palpebral fissures in the current infant seem to be related to trisomy 10p. High arched/cleft palate and retrognathia, reported as major anomalies of trisomy 10p, were excluded in this case, while the 10p11.2-p12.2 breakpoint region is thought to be related to these anomalies [21]. Hypotonia and a suspicious polycystic kidney are related to common symptoms of dup(10p)/del(10q) syndrome. Moreover, the 10q26 region is associated with obstructive uropathy due to anomalous ureterovesical junction [22]. Various reports suggest that the 10q terminal deletion is related to renal and urinary tract malformation in many female cases [3,23]. We inferred that the deletion of the 10q26.3 region in the present case is related to the suspicious polycystic kidney in our patient. Idiopathic jaundice was observed in the case of Lansky-Shafer et al. [1]. Thrombocytopenia was also observed in the cases of Ohba et al. [2] and Roberts et al. [11]. Neonatal respiratory distress has been reported in cases of del(10q) [24]. Lozić et al. [25] examined complete trisomy 10p syndrome and described a patient's physical defects such as dolichocepahly, cleft lip and palate, microcephaly, hypotonia, flat nasal bridge, clubfoot anomaly, marbled skin, and atrial septal defect.

In conclusion, this is *de novo* dup(10p)/del(10q) case is rarely observed; it witll facilitate the diagnosis of similar cases of recombinant dup(10p)/del(10q) and provides clinical information for genetic counseling.

Table 1. Cytogenetic and clinical findings in previous 14 cases of recombinant duplication chromosome 10 resulting from parental pericentric inversion compared to the present case

Variable	[12], 1973	[13], 1978	[14], 1979	[1], 1981	[10], 1981	[15], 1984	[11], 1989	[11], 1989	[2], 1990	[3], 1993	[3], 1993	[3], 1993	[9], 1994	[9], 1994	Present case
Breakpoints	p15-q25	N/A	p13-q26	p11-q26	p11-q25	p15-q24	p15.1-q25.2	p15.1-q25.2	p11.2-q25	p11.2-q26	p11.2-q26	p11.2-q26	p11.2-q26	p11.2-q26	p11.2-q26.3
Inheritance	Mat	Pat	Pat	Mat	Mat	Mat	Pat	Pat	Mat	Mat	Mat	Mat	Mat	Mat	De novo
Sex	M	NA	M	M	M	M	M	F	F	M	M	F	F	M	F
Trisomic segment															
Monosomic segment															
Age of at examination		NA		Newborn	10 yr		14 mo	9 yr	2 mo	4+1/2 mo	3+1/2 yr	Stillborn at 20 wk	4 yr	36 yr	2 yr
Phenotype	Trisomy 10q	Trisomy 10p	Trisomy 10p	Trisomy 10p	Trisomy 10p	Trisomy 10q	dup(10p)/ del(10q)	dup(10p)/ del(10q)	Trisomy 10p	Trisomy 10p	Trisomy 10p	Trisomy 10p	Trisomy 10p	Trisomy 10p	
Birth weight	3,170 g	ND	NA	1,640 g	2,750 g	2,000 g	3,500 g	3,400 g	2,322 g	2,950 g	2,800 g	normal*	2,700 g	NA	2,910 g
Hypotonia	+		+										+		Moderate
Growth failure														+	
Developmental/speech delay					+		+	+			+		+	+	
Mental retardation											+		+	+	
Microcephaly		+													
Dolichocephaly			+			+							+		
Wide sutures/fontanelle										+	+				
Absence of corpus callosum		+													
Asymmetric head/face		+			+	+					+				
Frontal bossing			+		+	+			+	+			+		
Hypertelorism		+			+	+								+	
Palpebral fissure abnormal					Upward	Downward			Horizontal						Upward
Epicanthic folds						+									
Strabismus							+				+				
Narrow palpebral fissure	+				+	+									
Broad/protruding nasal bridge	+	+	+		+	+	+		+	+				+	
Round sagging cheeks			+						+					+	
High and/or cleft lip/palate		+			+	+							+	+	
Micro/retrognathia				+		+			+	+					
Down-turned/turtle beak mouth			+						+	+					
Thin upper lip									+						
Low-set/malformed ears		+	+	+		+	+		+	+				+	

Pat, paternal; Mat, maternal; M, male; F, female; NA, not available; ND, not described.
*Limited autopsy.

Table 1. Continued

Variable	[12], 1973	[13], 1978	[14], 1979	[1], 1981	[10], 1981	[15], 1984	[11], 1989	[11], 1989	[2], 1990	[3], 1993	[3], 1993	[3], 1993	[9], 1994	[9], 1994	Present case
Hearing loss														+	
Short/webbed neck		+		+									+		
Microphthalmia	+					+									
Cardiac defect		+		Cysts										Heart murmur	
Renal defect		+			+							+			+
Genital defect			+	+											
Cryptorchidism		+					+								
Limb defects			+	+					+				+		
Clinodactyly	+					+			+						
Syndactyly							+	+							
Club foot		+							+	+					
Wide sandal gap						+								+	
Nail dysplasia		+													
Anemia													+		
Extra / single palmar creases				+					+	+	+		+	+	
Narrow thorax and pelvis									+/+						
Ocular malformation	+														
Hyperactivity								+							
Spina bifida occulta					+										
Umbilical hernia							+	+							
Respiratory distress								+							+
Thrombocytopenia								+	+						+
Absence of light 12th rib		+													
Feeding problem							+	+					+		Moderate
Jaundice				+											+
Back nodule mass															+
Direct bilirubinemia															+

Pat, paternal; Mat, maternal; M, male; F, female; NA, not available; ND, not described.
*Limited autopsy.

Table 2. Summary of major phenotypic anomalies of dup(10p), dup(10p)/del(10q) and del (10q) syndromes

Manifestation	Dup(10p)	Dup(10p)/ del(10q)	Del(10q)
Hypotonia	+	+	+
Microcephaly	+		+
Dolichocephaly	+		
Wide sutures	+	+	
Frontal bossing	+	+	
Broad/short nose	+	+	+
Palpebral fissures			
Up-slanting	+		
Horizontal			+
Strabismus			+
Cleft lip/palate	+		
Micro/retrognathia		+	
Down-turned mouth	+		+
Malformed ears	+	+	+
Short/webbed neck		+	+
Cardiac defect	+		+
Renal defect	+	+	+
Neonatal respiratory problem			+
Genital defect			
Cryptorchidism			+
Limb defects	+	+	
Growth failure	+	+	+

Acknowledgements

This study was approved by the Ethics Committee of Cheil General Hospital and Women's Health Care Center (#CGH-IRBGR-2007-02) and patient's consent was obtained.

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