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A Case of Tetrasomy 18p with Tracheomalacia

Ayako GOTO [1)], Goro SHIROTANI [1)], Hiroya KAKURA [1)],
Yoshio MORIYASU [1)], Yukiko IHARA [1)], Hitomi HAYASHI [1)],
Reimi TSURUSAWA [1)], Yukako YOSHIKANE [1)], Takahito INOUE [2)],
Takashi KODAMA [2)], Kouji IDE [2)], Shinichi HIROSE [2)],
and Atsushi OGAWA [1)]

[1)] *Department of Pediatrics, Chikushi Hospital, Fukuoka University*
[2)] *Department of Pediatrics, Faculty of Medicine, Fukuoka University*

Abstract

We present here a case of tetrasomy 18p with tracheomalacia. A three-month-old male child presenting with growth retardation and respiratory distress was admitted to our hospital. He also showed hypertonia, feeding difficulty, low set ears, high arched palate, micrognathia, unilateral single palmar crease, ventricular septal defect and hydronephrosis. A chromosomal analysis using blood lymphocytes stimulated by PHA showed 47,XY, + i (18). Three-dimensional CT and radioscopy revealed tracheomalacia in the upper airway. Children with tetrasomy 18p often exhibit growth retardation, feeding difficulty and respiratory distress. Tracheomalacia may cause these symptoms.

Key words: **Isochromosome 18p, Growth retardation, Tracheomalacia, 3 dimensional CT, Radioscopy of the upper airway**

Case Report

In 2009, a-3-month-old boy who presented growth retardation and respiratory distress was admitted to Fukuoka University Chikushi Hospital. He was born at 41 weeks' gestation, with a birth weight of 2616g. His family history was unremarkable. At 3 months his height was 55.5cm (-2.68SD), his weight 4.5kg (-2.62SD), and his head circumference 38cm (-1.27SD). He showed low set ears, high arched palate, micrognathia, wheeze, retraction, and unilateral single palmar crease. Neurological examination revealed hypertonia and head lag. Laboratory findings, brain CT and MRI, and chest roentgenography showed no abnormalities. Echocaediogram, however, revealed ventivular septal defect (0.8mm) and ultrasonography showed right hydronephrosis. The boy presented with wheezing and retraction, so we examined his upper airway using three dimensional CT and fluoroscopy. We found evidence of tracheomalacia. (Fig. 1 (a), (b))

Taken together, his problems included growth retardation, hereditary stigmata of degeneration, tracheomalacia, ventricular septal defect, right hydronephrosis, dacryocystitis, mental retardation and hypertonia. Since he had many degeneration stigmatas, we analyzed his chromosomes after informed consent was obtained from his parent.

Cytogenetic and fluorescent in situ hybridization analyses

Chromosome analysis was performed on peripheral PHA stimulated blood lymphocytes; G banding was used to analyze and illustrate 20 metaphase chromosomes and showed 47, XY,+mar karyotype. (Fig, 2 (a)) Fluorescent in situ hibridizasion (FISH) technique identified the extra marker and showed the boy's karyotype to be 47, XY, +i (18) (p10). (Fig. 2 (b)) Accordingly, we diagnosed the illness as a Tetrasomy 18p. No permission to examine

Correspondence to: Dr. Atsushi Ogawa, Department of Pediatrics, Chikushi Hospital Fukuoka University, 1-1, 1-chome Zokumyouinn, Tikushino city, Fukuoka 818-8502, Japan
Tel：+81-92-921-1011 Fax：+81-92-928-3890 E-mail：a-ogawa@fukuoka-u.ac.jp

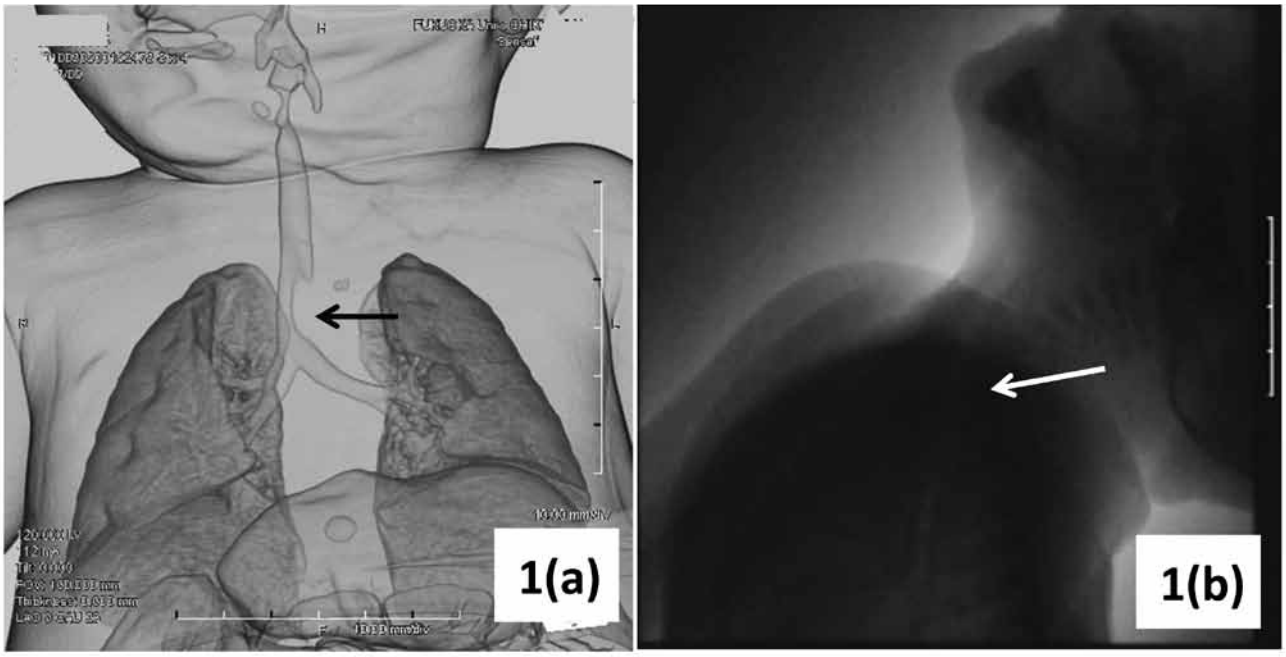

Fig. 1 (a) Three dimensional CT shows a section of the patient's narrow trachea.
(b) Fluoroscopy shows his trachea narrowing at its upper end.

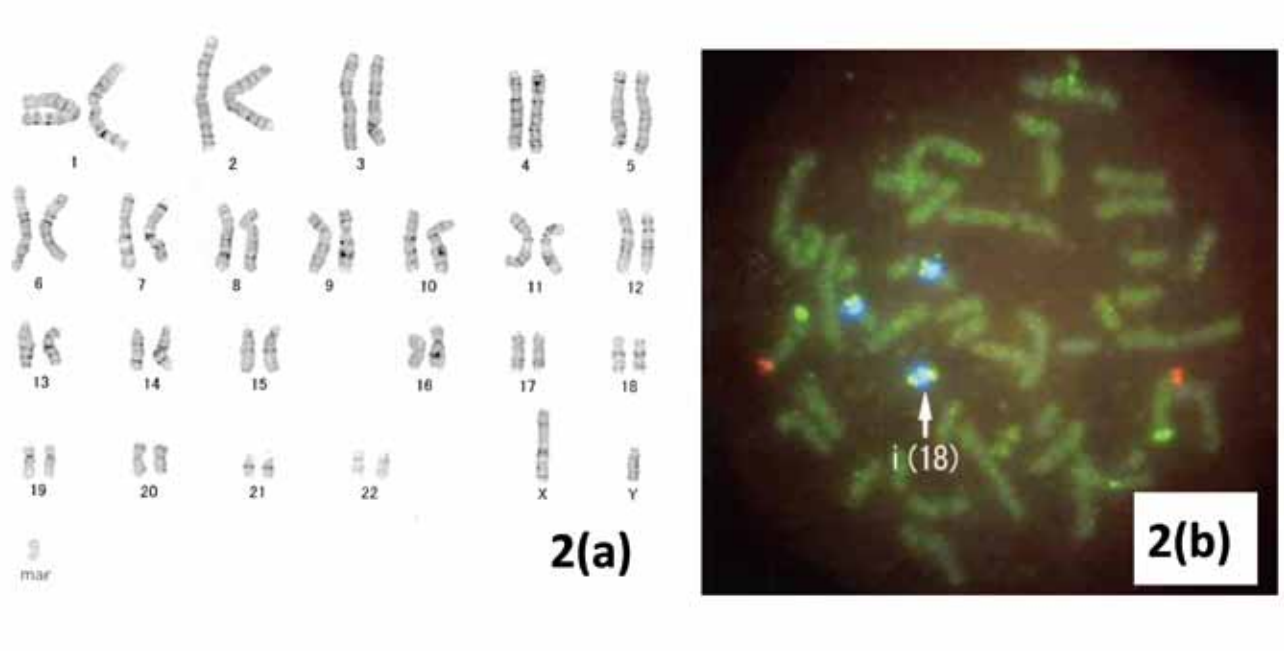

Fig. 2 (a) Karyotype of the patient, including a marker chromosome.
(b) FISH plate showing the subtelomeric region of chromosome 18p on the metaphase chromosome of our tetrasomy 18p patient. FISH probe identifies the short arm of chromosome 18p (spectrum yellow), centrometric 18 (spectrum aqua), chromosome 11p (spectrum green), and chromosome 11q (spectrum red). FISH also shows the extra marker is an ischromosome of short arm of chromosome 18.

his parent's chromosomes was obtained.

Discussion

Tetrasomy 18p appears to be one of the most commonly observed isochromosomes.[1] The first case of tetrasomy 18p was reported by Tangheroni et al in 1973 [2] and 50 cases have been reported since then. In Japan, Ogata et al first reported a case of tetrasomy 18p in Japan in 1977.[3] The majority of these were due to *de novo* formation, although Takeda et al reported a case of family with tetrasomy 18p[4]. Tetrasomy 18p is found once in every 140,000 live births [5], and affects males and females equally. Clinical findings for tetrasomy 18p include mental retardation（100%）, feeding difficulties（56%）, respiratory distress（15%）, microsephary（53%）abnormal muscle tone（73%）, ENT abnomalities（13%）, microcephaly（74%）, palatal abnormalities（6%）, congenital heart disease（24%）, cryptochidism（39%）.[6]

Our patient showed nearly all these symptoms. Although the literature reveals no cases of tetrasomy 18p with tracheomalacia, we believe the patient's tracheomalacia and hypertonia may have been one of the causes of his growth retardation and respiratory distress. The patient is now two years old and since age one, his tracheomalacia and hypertonia have improved, and his growth rate has increased.

Our conclusion is that children with Tetrasomy 18p often exhibit growth retardation, feeding difficulty and respiratory distress. Tracheomalacia may cause those symptoms.

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