

(Clinicopathologic Conference)

## Asphyxiating Thoracic Dysplasia<sup>†</sup> (SNUCH CPC-40)

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This newborn male infant was transferred to NICU of Sowha Children's Hospital from a private OB & Gyn Clinic. He was the first baby of this mother, and the gestational period was 41 weeks. No history of illness or drug administration during this pregnancy was elicited. There was no perinatal problems. His Apgar score was 7 at 1 minute and 8 at 5 minutes.

At the time of admission his body weight was 2,880 gm; body length 50cm; head circumference 35cm and chest circumference 27cm. Body temperature was 36.7°C, pulse rate 148 per minute and respiratory rate 70 per minute. He was cyanotic generally and the crying was weak.

Physical examination revealed a normocephalic head with open fontanelles. The chest was narrow and bell-shaped(Fig. 1). The respira-



Fig. 1. General appearance of the patient. The chest is markedly restricted to show narrow thorax.

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tory sound showed generalized decrease bilaterally, and the heart sound was regular without murmur. The abdomen was soft and

distended. The liver and spleen were not palpable. Four extremities were proportionally short. The hands and feet were chubby. There was no polydactyly. Chest X-ray film showed short ribs and narrow chest. Both lungs were poorly aerated and showed findings of aspiration pneumonia. The pelvis showed squared ilia.

After admission blood gas analysis showed pH 7.09, pCO<sub>2</sub> 63mmHg, pO<sub>2</sub> 119mmHg, HCO<sub>3</sub> 19.3mmol/l, BE -12.8 mmol/l, O<sub>2</sub> saturation 96.7%. The electrolytes were; Na 132 mmol/l, K 5.2mmol/l, Cl 101mmol/l. Complete blood counts revealed hemoglobin 18.0g/dl, hematocrit 50.9%. WBC 16,700/cmm (seg 63%, lymph 29%), platelet 194,000/cmm. Blood chemistry showed; AST 42 IU/l, ALT 13 IU/l, BUN 18 mg/dl, Creatinine 0.9 mg/dl, Ca 9.2 mg/dl, phosphorus 6.1 mg/dl, total protein 5.4 gm/dl and albumin 2.6 gm/dl/

He was managed with oxygen hood with FiO<sub>2</sub> 0.6. Blood gas findings improved slightly in oxygen hood. However, after 24 hours of life the respiratory movement became weak again and the blood gas data became worse, necessitating respirator assistance with IPPV. However, only a transient alleviation of symptoms and blood gas findings could be obtained. He suffered from intractable hypercarbia and acidosis, and became unresponsive to any drug or assisted ventilation. He died 66 hours after birth.

## DISCUSSION

Dr. Kim : The patient was a fullterm newborn male with 41 weeks of gestation and there was no remarkable problem during pregnancy or perinatal period. His APGAR score was 7 at 1 minute and 8 at 5 minutes. Body weight was 2880 gram, body length was 50cm, and head circumference was 35cm. His chest circumference was 27cm showing markedly reduced size of the thoracic cavity. Physical examination revealed normocephalic head and narrow, bell-shaped thorax and proportionately short extremities. He had chubby hands and feet, but

there was no polydactyly or abnormal fusion of the phalanges. His blood gas analysis revealed respiratory and metabolic acidosis. He was treated with O<sub>2</sub> therapy and showed improvement in blood gas findings, but after 24 hours respiratory movement became weak and he suffered from intractable hypercarbia and acidosis, and expired 66 hours after birth.

Radiological findings showed marked narrowing of the thoracic cage due to short horizontal ribs(Fig. 2). There was anterior flaring of the ribs and inferior ribs flared resulting in bell-shaped thorax. Also he had high positioned handle-bar shaped clavicle and his scapula was normal. Bony spine was normal, showing normal appearance of vertebral body

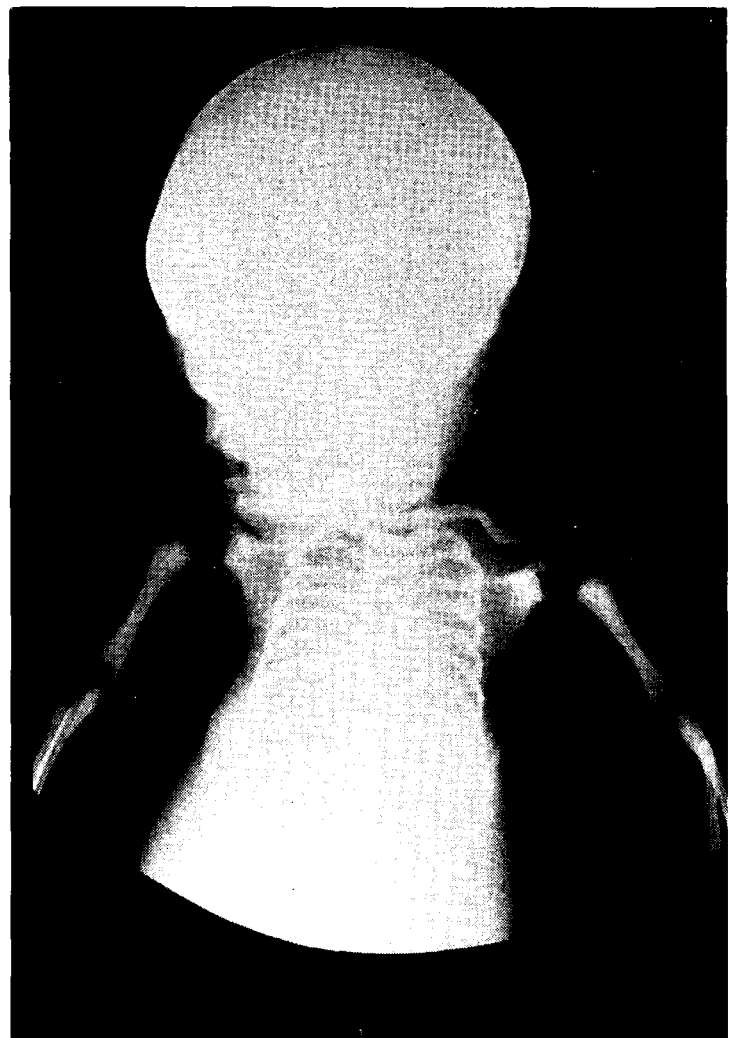


Fig. 2. Anteroposterior view of the chest. The thoracic cage is extremely narrow with rib ending at mid-axillary line. The ribs are short, thickened at the distal ends, and relatively horizontal with elevation of the clavicles. The spine is normal.

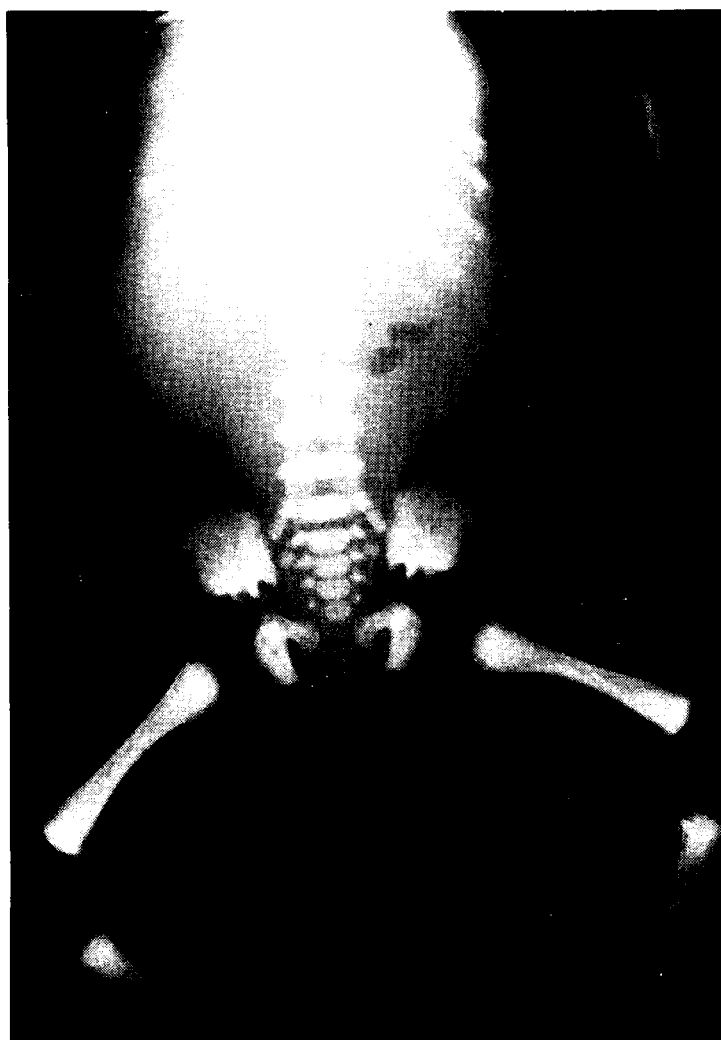


Fig. 3. Anteroposterior view of the pelvis. The iliac bones are short in vertical dimensions. The margins of acetabular roofs are irregular with prominent spurs.

and normal interpedicular distance from cervical to lumbar spine level (Fig. 3). The pelvic bones showed square-shaped, small sized ilia with flattened acetabular roof and there was inferior hook like bony projection in the mid portion of the acetabular roof showing trident acetabulum. Lower extremity showed mild shortening but the bony density or architecture of metaphyseal end were normal. The hand showed short metacarpal and phalangeal bones and marked shortening of distal phalanx was seen (Fig. 4). The fingers showed splayed appearance. There was no evidence of polydactyly or metacarpal bony fusion anomaly. Brain ultrasonography showed left side germinal matrix hemorrhage shown as echogenic nodular mass in the lateral aspect of the frontal

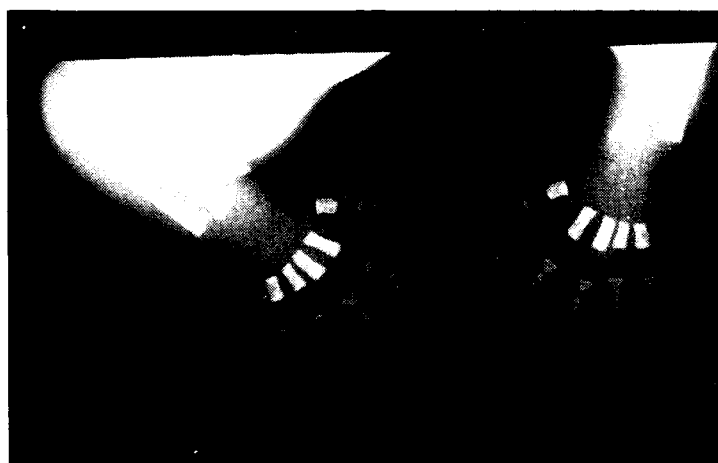


Fig. 4. Radiograph of hand bones in this patient.

horn and sagittal scan of the lateral ventricle showed small cystic lesion at the glomus of the choroid plexus suspecting true choroid plexus cyst or loculated fluid collection due to previous germinal matrix hemorrhage and intraventricular extension. The abdominal ultrasound showed no remarkable abnormality in the liver and right kidney.

In summary, the patient had respiratory difficulty and narrow chest cage and short extremity, and died in neonatal period due to respiratory failure. Radiological abnormalities were short ribs and narrow thorax and mild shortening of the extremity and hypoplastic ilia and short phalanges. The spine, skull and bony densities were normal. The most striking findings were short ribs and narrow thorax and respiratory failure resulting in neonatal death. The differential diagnosis of short rib with neonatal death can be lined as below:

**1. Achondrogenesis :** Achondrogenesis usually shows large head due to hydrocephalus and extreme short stature with severe micromelia. The skeletal ossifications are remarkably decreased, even vertebral bodies are invisible due to poor ossification. And also there are extreme shortening and cuboidal appearance of the tubular bone and the distal end of metaphysis flared and metaphyseal scallopings are characteristic. This patient is different from

achondrogenesis in the point of normal bony densities, minimal long bone changes and normal spine.

**2. Congenital hypophosphatasia** : Congenital hypophosphatasia usually shows markedly defective calvarial ossification and tubular bones. The ribs are short and thin, and metaphyseal end of the tubular bones show irregular radiolucency due to the rachitic changes and usually there is hypercalcemia.

**3. Thanatophoric dysplasia** : Thanatophoric dysplasia usually shows severe growth deficiency and clover-leaf large skull due to premature craniosynostosis and hydrocephalus. The ribs are very short and thorax are narrow. The spines show markedly decreased height of the vertebral body showing universal platyspondyly and the frontal view of the spine shows H-shaped vertebrae due to severe thinning of the vertebral body. The long bones show flared metaphyseal end and shortening and bowing, so deformity of the femur characteristically resembles “telephone receiver” appearance. There is decrease in the the interpedicular distance of the spine craniocaudally. This case is different from thanatophoric dysplasia that he showed normal spine and no radiological evidence of long bone deformity.

**4. Short-rib polydactyly syndrome** : Short-rib polydactyly syndrome usually shows short horizontal ribs and narrow thorax. There is polydactyly usually in the ulnar side but sometimes polydactyly can be absent. Iliac bones are hypoplastic and trident acetabulum can be shown. The short tubular bones show pointing appearance in the type I short-rib polydactyly syndrome (Saldino-Noonan) such as pointed end of the femur and there is more pronounced limb shortening. Type II short-rib polydactyly syndrome (Majewski) shows characteristic short, oval shaped tibia and cardiac anomalies or cleft palate are frequently associated.

**5. Chondroectodermal dysplasia** : Chondroec-

todermal dysplasia shows short rib with narrow thorax and mesomelic dysplasia. Polydactyly of ulnar side can be present and carpal anomaly such as carpal fusion is also associated. The pelvis shows hypoplastic trident appearance and the proximal tibial epiphysis is small when visible. Ectodermal dysplasia such as hypoplastic nail or fine hair or cardiac anomaly are associated and short upper lip with multiple frenulum can be associated. This case is quite difficult to differentiate from chondroectodermal dysplasia, but the absence of ectodermal dysplasia or polydactyly can be differential points.

**6. Asphyxiating thoracic dystrophy** : The asphyxiating thoracic dystrophy shows short horizontal ribs, narrow thorax, brachydactyly and hypoplastic trident pelvis. They usually suffer from tachypnea or aspiration pneumonia and due to hypoplasia of the lung, respiratory failure is common. The polydactyly can be associated in the radial side, but the frequency is quite low. The characteristic finding is that the asphyxiating thoracic dystrophy does not show abnormality in skull and spine.

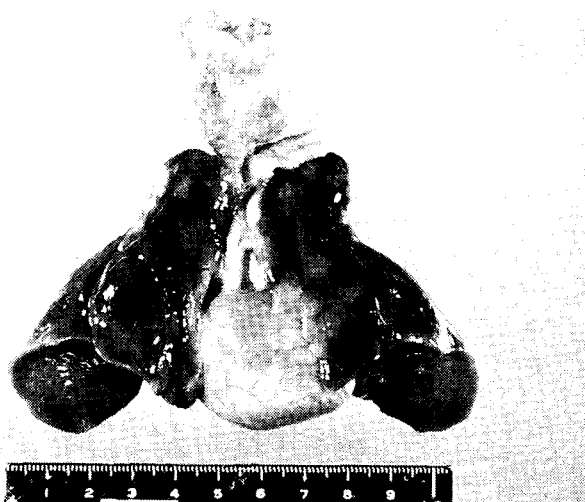
So when the infants with dystrophic thorax and skeletal deformity resulting from the disturbance of in utero endochondral bone formation without evidence of skull and spine abnormality, asphyxiating thoracic dystrophy is the most probable diagnosis. The most difficult differential diagnoses are chondroectodermal dysplasia and short-rib polydactyly syndrome. But due to the fact that there were no metaphyseal abnormalities, abnormal tibia, polydactyly or cleft palate, short rib polydactyly syndrome can be ruled out. And also there is no evidence of abnormal hair, nail or fused lip or polydactyly, chondroectodermal dysplasia is not likely, but radiologically finding alone can not exclude chondroectodermal dysplasia.

In conclusion, with the findings of short rib, narrow thorax, hypoplastic ilia, respiratory distress, and normal skull and spine, the most probable diagnosis is asphyxiating thoracic dystrophy. The possibility of chondroectodermal

dysplasia and short-rib polydactyly syndrome is low, but these two conditions should be included in the differential diagnoses.

**Pathologic findings (Dr. Chi) :** Postmortem examination confirmed the clinical diagnosis of asphyxiating thoracic dysplasia. The gross features were exactly same as described by the discussant. The chest organ particularly lungs were compressed by the narrow thorax(Fig. 5) Microscopically the epiphyseal plate showed growth plate consisting of irregular proliferation of chondroblasts with poor osteoid deposit. Endochondral ossification, however, is well-organized without giant chondrocytes(Fig. 6). There was no polydactyly. There was pneumonia which was the direct cause of death.

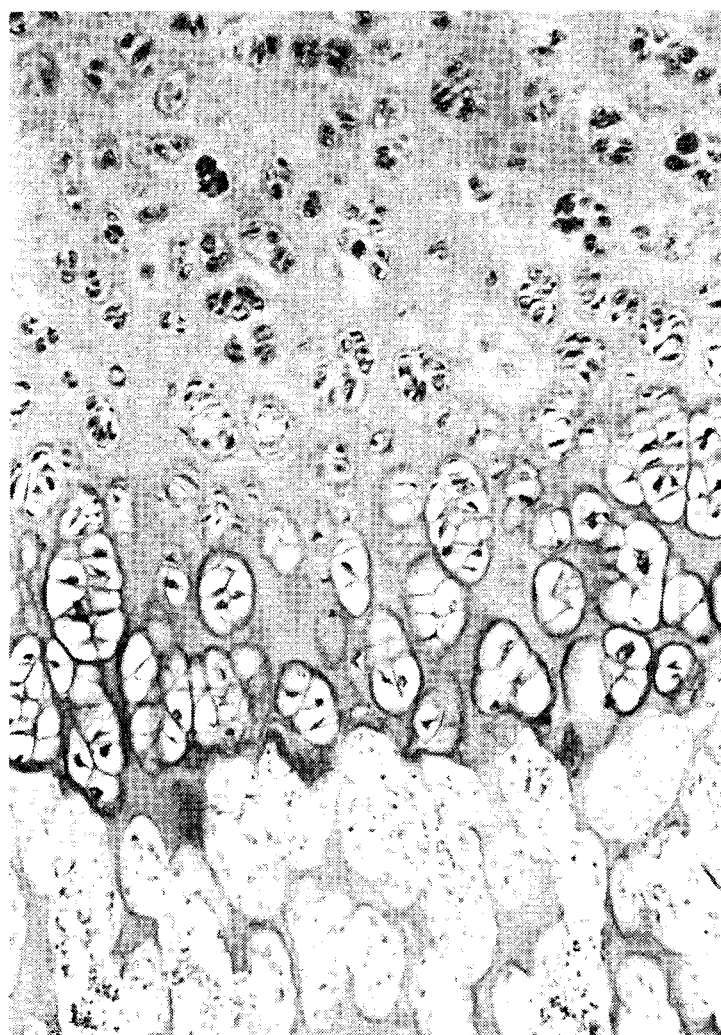
Pathological diagnosis: Asphyxiating thoracic dysplasia.



**Fig. 5.** The chest organs of the patient. The lungs are unusual in shape to show compressed and atelectatic appearance.

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**Fig. 6.** Photomicrograph of epiphyseal plate showing ossification process per se is not drastically disturbed. H & E X200

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