Pectus excavatum and situs inversus totalis: A new combination or a coincidence? [1]

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LETTER TO THE EDITOR

PECTUS EXCAVATUM AND SITUS INVERSUS TOTALIS: A NEW COMBINATION OR A COINCIDENCE?

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In this report, we describe a 3-year-old boy with situs inversus totalis and pectus excavatum who underwent successful surgical correction. It is discussed whether this unusual condition is a new combination or coincidence.

Pectus excavatum, the most common congenital chest wall deformity, is manifested by deformity of the costal cartilages resulting in a depressed and often rotated sternum. Although the exact etiology is unknown, there are various suggestions. It was accepted as a developmental abnormality of the diaphragm in the past. Hecker and associates described histopathologic changes that can be seen in scoliosis and aseptic necrosis, at the costal cartilages of the pectus (2). Pectus excavatum is frequently associated with other musculoskeletal abnormalities such as scoliosis, Marfan syndrome, Prune Belly syndrome (4, 5). This deformity also occurs in chromosomal anomalies such as Turner syndrome (3). In addition there is a clear-cut increased family incidence, which suggested a genetic basis (4). In a review of 704 patients by Shamberger *et al.*, 37% had a family history of chest wall deformity (4).

Situs inversus totalis is defined as complete mirror-image transposition of the abdominal and thoracic viscera. Situs inversus totalis with cardiac, pulmonary, renal, esophageal, hepatic, pancreatic and vascular anomalies have been reported (1). The cause is believed to be a gene mutation involving either autosomal dominant or recessive inheritance with variable penetrance, although sporadic cases are reported to occur (1). Some authors suggest that the early embryonal process of the determination of the normal body situs is complex and probably controlled by several genes.

A-3-year old boy was referred to this hospital for the evaluation of deep pectus excavatum. The mother and father were second-degree relatives. He was the first child of the family and had a younger brother 15-months of age. There was no family history of congenital or other disease. Both children were born after a normal pregnancy and delivery. On examination growth was within normal limits. He had moderate respiratory distress with deep pectus excavatum. Urine and hematologic and biochemical tests were within normal range. Chest films revealed a deep pectus excavatum and dextrocardia. An abdominal ultrasound confirmed the diagnosis of situs inversus totalis but showed no additional malformation. A frontal view of the upper abdomen and chest on CT scan revealed dextrocardia and a left-sided liver shadow. An ECG showed the typical pattern of mirror image dextrocardia, and echocardiography was normal excluding dextrocardia. Chromosomal analysis performed on a peripheral blood lymphocyte culture showed a normal male karyotype. Pectus excavatum was corrected via a modificated Ravitch method. Postoperative period was unremarkable and the patient was discharged after 8 days. On follow up, he was in a good general clinical condition.

In conclusion, this is the first report of a patient with pectus excavatum and situs inversus totalis. It is unclear whether these findings were

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coincidental or a new combination. Further studies are needed to confirm this unusual combination.

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