Congenital Herniation of the Large Bowel
Into the Pericardial Cavity*

Survival into Adulthood Despite a Generalized
Connective Tissue Disorder*

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An adult with an unusual combination of mesodermal defects is presented. A striking finding was the herniation of a major portion of the large bowel into the pericardial cavity through a foramen of Morgagni defect. In addition, the patient exhibited a variety of muscular skeletal anomalies, Monckeberg sclerosis of the muscular arteries, and complete heart block. Urinary mucopoly saccharide excretion and a chromosome count were normal. The most likely explanation of the observed findings is a biochemical defect in connective tissue.

Heritable disorders of connective tissue, such as the Marfan syndrome, Ehlers-Danlos syndrome, pseudoxanthoma elasticum and Hurler's syndrome may have musculoskeletal as well as cardiovascular manifestations. McKusick believes that the multiple and diverse manifestations of these syndromes indicate that the normal gene which underwent mutation determines a substance or process with widespread significance to the body's economy. An adult patient was recently seen with a major portion of the large bowel herniated into the pericardial cavity, striking skeletal anomalies, vascular calcification and complete heart block. To the authors' knowledge, this combination of connective tissue abnormalities has not been reported previously. The following report is a description of this patient together with the clinical and laboratory studies performed upon him.

CASE REPORT

The patient is a 36-year-old man with congenital bilateral hip dislocations and talipes equinovarus. The details of the pregnancy and delivery are not available. As a child he had multiple operations on his feet and hips but denied any history of rheumatic fever or diphtheria. During adolescence he had one episode of pneumonia but was otherwise in good health.

He was well until the age of 32 when he developed progressive abdominal swelling and ankle edema. At the age of 35 he was admitted to another hospital with radi- cal leg pains, ankle edema and ascites. Bradycardia was noted at that time and complete heart block was present as shown on the electrocardiogram. With diuretic therapy the patient improved. In the fall of 1963 he developed increasing difficulty in walking, associated with recurrent leg pains and ankle edema, and was admitted to the Strong Memorial Hospital in December, 1963, for evaluation.

Despite the physical limitations the patient completed high school and two years of college. Throughout his adult life he has remained gainfully employed as a bookkeeper. The patient is the only child of nonconsanguineous parents. He knows of no other relatives with similar skeletal abnormalities. The mother is alive and well, and the father died at age 33 of alcoholism and tuberculosis.

Physical Examination

Pulse, 50/min., respirations, 35/min., and blood pressure, 160/92 mm Hg. The patient was of short stature (5 feet) with a relatively large head, frontal bossing, short neck, scoliosis, lordosis, protuberant abdomen and a flared chest. The hands were deformed by ulnar deviation at the wrists and flexion contractions of the phalanges. Hip movements were limited in all directions and he had bilateral talipes equinovarus. Eye examination gave normal results without corneal clouding or blue sclerae; the optic fundi were unremarkable. Hearing, confirmed by audiometric examination, was normal. The teeth were unremarkable and in good repair. The jugular veins were distended to the angle of the mandible at 90 degrees with occasional "cannon waves." There was slight limitation in expansion of the chest, the expiratory phase was slightly prolonged and bowel sounds were audible over the entire thorax. Cardiac size could not be estimated clinically, and there were no thruts, heaves or thrills. The heart sounds were distant and no murmur were audible. The abdomen was distended and diastasis recti was present. The liver edge was palpable 2 cm below
the right costal margin; the spleen was not felt and shifting dullness was absent. The testes and penis were unremarkable; a normal male hair distribution was present and there were no inguinal herniae. Pitting edema to the knees of both lower extremities was present. The deep tendon reflexes were symmetrically hyperactive, bilateral ankle clonus and bilateral Babinski signs were present, but the sensory examination was normal. Mental testing showed an average intelligence (I.Q. 103).

**Laboratory Studies**

The hematocrit was 45 percent to 50 percent and the white blood count was 12,000 cells per cubic millimeter with a normal differential. There was no abnormality of the polymorphonuclear leukocytes. Results of urine analysis were negative except for trace protein; 150 mg of protein were present on quantitative 24-hour collection. The electrocardiogram revealed complete heart block with a ventricular rate of 50/min.; the QRS complex had a right bundle branch block configuration (Fig 1). Initial venous pressure was 17 cm H₂O, and it subsequently declined to below 10 cm H₂O after the administration of a vigorous cathartic. The following findings were present on skeletal x-ray survey: talipes equinovarus of the feet with preservation of the ankle joints; subluxation of the metacarpal joints of the thumb of the right hand; contracture deformity of the phalanges of the fourth and fifth fingers of both hands; ulnar deviation of both wrists; fragmentation and flattening of both hip joints with subluxation of the left femur on the hip; scoliosis of the lumbar spine with a concavity toward the right; normal vertebral bodies; narrowing of the cervical spinal canal in the A-P diameter with C-1 closely approximated to the cranial vault; the narrowing of the distance between the posterior arch of C-1 and the posterior surface of the odontoid process. Platybasia was present on skull x-rays and the mandibular condyles were broad and shallow. On myelography, there was severe narrowing of the spinal canal in the region of the foramen magnum in association with the platybasia; also, extradural compression of the opaque material was present at the L₂ to L₃ vertebral interspaces suggesting a herniated lumbar disc in this region. Multiple air-fluid levels in the thorax consisting of large bowel and portions of the small intestine (Fig 2) were demonstrated roentgenographically. Chest tomograms and barium enema studies (Fig 3) revealed that the thoracic intestines were located within the pericardium and the intestines were herniated through a foramen of Morgagni defect in the anterior portion of the diaphragm. In the radiologic evaluation of the skeleton, calcification of the blood vessels in the pelvis, wrist, and ankles was noted.

Muscle evaluation, including electromyography and a deltoid muscle biopsy, was normal. Pulmonary function tests (Table 1) revealed markedly reduced total lung capacity and vital capacity and an increase in the residual volume to total lung capacity ratio. The timed vital capacity and the maximum expiratory flow rate were reduced. Mild arterial oxygen desaturation was present without carbon dioxide retention or polycythemia. The pulmonary function tests were consistent with severe restrictive and moderate obstructive respiratory impairment.

Serum protein fractionation and protein electrophoresis were normal. Results of Wassermann test and a rheumatoid latex fixation test were negative. Blood serum chemistries including those of glucose, urea nitrogen, electrolytes, alkaline phosphatase, transaminase, calcium, phosphorous, bilirubin and cephalin flocculation were normal. The serum protein bound iodine and urinary 17-ketosteroid excretion were also normal. Bovine albumin and toluidine blue screening tests for increased urinary mucopolysaccharides were negative. Chemical analyses of two 24-hour urine collections for mucopolysaccharide excretion were normal. The mucopolysaccharide excretion expressed in terms of uronic acid was 8 mg/day. Paper chromatography and electrophoresis of the patient's urine failed to reveal any abnormal mucopolysaccharide constituents. On bone marrow chromosome analysis, a normal male karyotype was present. Results of slit lamp examination of the eyes were normal.

During the four years since the aforementioned studies were carried out, the patient has remained active and free of significant symptoms of cardiac or respiratory embarrassment.

**Discussion**

This patient has a number of striking abnormalities involving the diaphragm, bones, joint, blood vessels and heart. All of the abnormalities involve mesodermal structures and we may conclude that this patient is a dwarf with generalized connective tissue disorder. We have been unable to assign a specific diagnostic label to this patient's condition. The skeletal defects conform closely to the syndrome of diastrophic dwarfism which has certain overlap with Hurler's syndrome and Morquio's disease. Hurler's syndrome is excluded by the absence of mucopolysaccharides in the urine. Complete heart block and vascular calcification have not been described previously in diastrophic dwarfism, Hurler's syndrome, or Morquio's disease, and these findings are unique features of the case. The patient is karyotypically normal and the most likely...
explanation for the observed findings is a biochemical defect involving skeletal and visceral connective tissue. In view of the limited family history, the heritable aspects of this patient's disease cannot be determined at the present time.

The age of onset of the heart block in this patient is not clear. Records from the hospitals in which the patient was operated on for the multiple orthopedic procedures during childhood did not reveal any information about heart rate. Heart block was first documented at age 35, and it is only conjecture whether this conduction disturbance was present since birth. Although many of the patients with connective tissue disorders have diffuse cardiac involvement, heart block has been described only in Marfan's syndrome. Recently, James and coworkers investigated the cardiac conducting system in Marfan's syndrome and they described two patients in whom first degree heart block was associated with occlusion of the atrioventricular nodal artery by medial hyperplasia, and hemorrhage and fibrosis in the region of the node. In addition, many other atrial and ventricular myocardial vessels were involved in their two patients. In the patient under discussion, the vascular calcification suggests that the heart block may be secondary to nodal artery disease in a manner similar to that described by James in Marfan's syndrome.

The calcification of the muscular arteries of the extremities in the patient is characteristic of Monckeberg's sclerosis. The etiology of this condition is unknown but the first changes observed histologically are a loss of undulation and pliability of the internal elastic membrane. Thereafter, the membrane becomes fragmented and acquires calcium on its surface and in its intrinsic structure. These degenerative changes are subsequently followed by mesenchymal reactions of a reparative nature. The disease process begins in early life and gradually progresses with advancing age. In Monckeberg's original article, the vascular calcification was observed primarily in the older age group, and this disease process is considered to be a variant of the generalized arteriosclerotic process. Premature calcification of the arterial walls has been noted in pseudoxanthoma elasticum but not in other connective tissue disorders. The similar vascular calcification in the case under discussion suggests that the underlying connective tissue abnormality may have certain features in common with pseudoxanthoma elasticum. Elucidation of the biochemical disorder in these disease processes may provide
insight into the etiology of arteriosclerosis.

Large and small bowel were herniated into the pericardial cavity through a probable foramen of Morgagni defect in the diaphragm. Harrington, in a description of the various types of diaphragmatic hernias, comments that foramen of Morgagni defects are not due to faulty fusion or improper deposition of the embryonic mesodermal elements, since the anterior portion of the diaphragm is derived only from the septum transversum. This type of hernia is more likely to result from faulty attachment of the diaphragm to the sternum and this explanation is in accordance with the generalized connective tissue disorder exhibited by the patient. In an analysis of seven cases with congenital diaphragmatic hernias, Gross described only one patient with herniation of the bowel into the pericardial cavity. Chest roentgenograms in that patient were similar to the ones presented in the case under discussion.

Pulmonary function was severely compromised with restrictive and obstructive respiratory impairment. The restrictive defect probably resulted from compression of the lung by the bowel in the chest as well as a reduction in the thoracic cage volume by a foreshortened torso structure. The obstructive disease may be secondary to chronic subclinical respiratory infections which frequently complicate restrictive ventilatory abnormalities. Cardiac function was also impaired. Although the right-sided heart failure may have been related in part to the heart block and associated bradycardia, the massive herniation of the bowel into the pericardial cavity certainly may impede venous return to the heart.

Table 1—Pulmonary Function Tests

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<tr>
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<tbody>
<tr>
<td>Vital capacity (liter)</td>
<td>1.28</td>
<td>1.85</td>
<td>3.41</td>
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<tr>
<td>Residual volume (liter)</td>
<td>0.76</td>
<td>1.09</td>
<td>1.0</td>
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<tr>
<td>Total lung capacity (liter)</td>
<td>2.04</td>
<td>2.94</td>
<td>4.45</td>
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<td>RV/TLC (%)</td>
<td>37.5</td>
<td>37.1</td>
<td>23.0</td>
</tr>
<tr>
<td>Minute ventilation (liter/min.)</td>
<td>7.18</td>
<td>8.55</td>
<td>4.68</td>
</tr>
<tr>
<td>Respiratory rate</td>
<td>25.0</td>
<td>19.0</td>
<td>12.0</td>
</tr>
<tr>
<td>O2 consumption (ml/min.)</td>
<td>159.0</td>
<td>119.0</td>
<td>—</td>
</tr>
<tr>
<td>Timed vital capacity 1 sec. (%)</td>
<td>70.0</td>
<td>70.0</td>
<td>75.0</td>
</tr>
<tr>
<td>3 sec. (%)</td>
<td>88.0</td>
<td>90.0</td>
<td>95.0</td>
</tr>
<tr>
<td>Maximum expiratory flow rate (liter/min.)</td>
<td>77.0</td>
<td>66.0</td>
<td>7300.</td>
</tr>
<tr>
<td>Diffusion capacity (ml CO/min./mm Hg)</td>
<td>—</td>
<td>22.7</td>
<td>20.0</td>
</tr>
<tr>
<td>Arterial blood gases</td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>O2 content (vol. %)</td>
<td>17.3</td>
<td>—</td>
<td>17.5</td>
</tr>
<tr>
<td>O2 saturation (%)</td>
<td>92.9</td>
<td>—</td>
<td>&gt;93.0</td>
</tr>
<tr>
<td>CO2 content (vol. %)</td>
<td>42.9</td>
<td>—</td>
<td>40-50</td>
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<tr>
<td>Hgb (gm %)</td>
<td>13.9</td>
<td>—</td>
<td>13.5-15.0</td>
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FIGURE 3. Barium enema examination with PA and lateral roentgenograms of the chest. Almost the entire large bowel, appendix and a part of the small intestine are present within the pericardial cavity.
CONGENITAL HERNIATION OF LARGE BOWEL

The venous pressure was directly related to the degree of constipation; with cathartics the venous pressure diminished. Fecal cardiac tamponade may develop in this patient if obstruction becomes severe.

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REFERENCES

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FIRST ASIA PACIFIC CONGRESS ON DISEASES OF THE CHEST
Kyoto, Japan, July 2-4, 1969

The First Asia Pacific Congress on Diseases of the Chest, to be held at the Kyoto Assembly Hall, will be sponsored by the Japan Chapter of the College in cooperation with the Japan Society of Chest Diseases and the Japanese Association of Tuberculosis, with assistance from College chapters in the Asia-Pacific area. Deadline for papers and applications is February 28, 1969 (registration fee — $5). The registration fee includes admission to all scientific sessions and exhibits, as well as the Ninth General Meeting of the Japan Society of Chest Diseases and the 44th General Meeting of the Japanese Association for Tuberculosis.

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