Currarino-Silverman Syndrome (Pectus Carinatum Type 2 Deformity) and Mitral Valve Disease*

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Currarino-Silverman syndrome is a rare disorder characterized by premature fusion of manubrio-sternal joint and the sternal segments, resulting in a high carinate chest deformity; it is frequently associated with congenital heart disease. Among the various heart lesions reported in this syndrome, mitral valve disease and coarctation of the aorta have not yet been described (to our knowledge). Our report consists of five children with this syndrome, four of whom had mitral valve disease, with an associated coarctation of the aorta in one patient. The fifth patient had an innocent heart murmur. (Chest 1992; 102:790-82)

Currarino-Silverman (C-S) syndrome, also known as pectus carinatum type 2 or pouter pigeon breast, is a rare deformity and is probably caused by premature fusion of some of the sternal ossification centers and by obliteration of the manubrio-sternal joint. This results in an abnormally short sternum with forward angulation at the manubrio-sternal junction. Different types of congenital heart diseases (CHD) have been reported in connection with this disorder, although, to our knowledge, mitral valve disease and coarctation of the aorta have not been described. Our report consists of five children who were identified with C-S syndrome in the course of the last four years. Four of them also had mitral valve abnormalities, with an associated coarctation of the aorta in one patient. The fifth patient had an innocent heart murmur. In the mirror of these cases, the possible pathogenesis of CHD and C-S syndrome is discussed.

Material and Methods

The records of all patients at the Pediatric Cardiology Department of the James H. Quillen College of Medicine, from December 1986 to the present were reviewed. Of a total 2,113 patients reviewed, five patients (all female) ranging in age from newborn to nine years were identified with C-S syndrome. Patients with associated congenital mesenchymal syndromes such as Down's, Marfan's, or Noonan's syndrome or any recognized chromosomal abnormality were excluded from the study. All patients had a complete history and physical examination by a pediatric cardiologist (A.V.M.), 12-lead surface electrocardiogram, chest roentgenograms (anteroposterior and lateral views), and complete color-flow echocardiograms. Three patients had several serial chest roentgenograms, and the same number underwent cardiac catheterization and angiography. The diagnosis of C-S syndrome was made both clinically and roentgenographically (Fig 1 and 2). Each patient's lateral chest roentgenograms were reviewed for the length and configuration of the sternum, segmentation, and state of fusion of the manubrio-sternal joint.

Results

The clinical and laboratory findings are summarized in Table 1. All five patients had short, solidly synostosed sterna with one or two segments in the sternal body and prominent angulation at the manubrio-sternal junction (Fig 1 and 2). Four of these five had organic heart disease. Mitral valve abnormality was present in all four; one child had a parachute mitral valve with mild mitral stenosis (MS) and regurgitation (MR), one had endocardial cushion type of atrioventricular valve with mild to moderate MR, and two had mitral valve prolapse (MVP) with mild MR. Additionally, one patient had coarctation of the aorta that was successfully treated by percutaneous balloon angioplasty and one patient had ostium secundum type of atrial septal defect that closed spontaneously.

Figure 1. Clinical photograph of patient with Currarino-Silverman syndrome (pectus carinatum type 2).
by premature fusion of the sternal ossification centers and obliteration of the manubrio-sternal joint, is much rarer. This causes a high carinate deformity and is also named “pouter pigeon deformity.” Currario and Silverman were the first to mention the high association of this deformity with CHD.

Shamberger and Welch compiled and analyzed patients with chondro-manubrial deformity and CHD from the literature. Included in their summarization was the report by Lees and Caldicott who reviewed consecutive roentgenograms of 1,915 children, and found anomalies of sternal fusion in 135 (7 percent), and 24 of these had proven CHD. It is possible, however, that Lees and Caldicott may not have referred to the C-S syndrome in their series, but to various anomalies of fusion of the sternal body segments (sternabrae). Some of the patients whose cases were reported by Steiner et al and Gabrielsen and Ladyman also may not be typical cases of C-S syndrome. Excluding the patients of Lees and Caldicott in the summarization of Shamberger and Welch, it appears that of 58 patients, 32 patients had various congenital heart lesions associated with C-S syndrome. Of these 32 patients, the most frequent lesions found were ventricular septal defect (18 cases), patent ductus arteriosus (12 cases), atrial septal defect (four cases), tetralogy of Fallot (three cases each), and transposition of the great arteries (two cases). Our report describes, for the first time, mitral valve abnormalities and coarctation of aorta in C-S syndrome.

The embryology of the sternum has been dealt with in detail by several authors. Briefly, the sternum and the endocardial cushions of the heart are formed between the fourth and sixth weeks from mesenchyme from prebranchial and lateral body wall regions. The atrioventricular valves and at a later stage, the membranous part of the interventricular septum, are developed from the endocardial cushions. Gabrielsen et al suggested that an environmental insult or a genetically predisposed injury to the anterior segment mesenchyma at the time of cardiogenesis may result in abnormalities of endocardial cushion, sternum, and aortic arch derivatives. Abnormal migration of prebranchial mesenchymal cells to the endothelial heart

**Table 1—Summary of Patients with Currario-Silverman Syndrome***

<table>
<thead>
<tr>
<th>No./Sex</th>
<th>Age at Diagnosis</th>
<th>Roentgenographic Appearance of Sternum</th>
<th>Cardiac Abnormalities, Clinical and Echo</th>
</tr>
</thead>
<tbody>
<tr>
<td>1/F</td>
<td>8 yr</td>
<td>Short, 1 segment, fused MSJ</td>
<td>MVP, moderate MR</td>
</tr>
<tr>
<td>2/F</td>
<td>6 yr</td>
<td>Short, 1 segment, fused MSJ</td>
<td>PMV, MS, coarctation aorta</td>
</tr>
<tr>
<td>3/F</td>
<td>Newborn</td>
<td>Short, 2 segments, fused MSJ</td>
<td>ECD† type VSD, small PDA</td>
</tr>
<tr>
<td>4/F</td>
<td>9 yr</td>
<td>Short, 1 segment, fused MSJ</td>
<td>MVP, mild MR</td>
</tr>
<tr>
<td>5/F</td>
<td>7 yr</td>
<td>Short, 1 segment, fused MSJ</td>
<td>Innocent heart murmur</td>
</tr>
</tbody>
</table>

*ECD = endocardial cushion defect; MR = mitral regurgitation; MS = mitral stenosis; MSJ = manubrio-sternal joint; MVP = mitral valve prolapse; PDA = patent ductus arteriosus; PMV = parachute mitral valve; VSD = ventricular septal defect.

†Spontaneous closure of atrial septal defect, ostium secundum type.
tube caused by genetic or environmental factors has also been proposed as a mechanism for defective cardiogenesis.11 Ravitch4 further pointed out that the compression of the early development of the appendicular and axial skeleton and the heart into the same short period of time may provide ample opportunity for the production of associated anomalies of these structures.

A few chromosomal duplication (eg, trisomy 18) and interstitial deletion syndromes12 list a short sternum and/or pectus carinatum and CHD among other typical features, suggesting the possibility of a genetic etiology. The finding of pectus carinatum type 2 in two siblings and their mother, and MVP and MR in the mother and one daughter further strengthens this possibility. Also, interestingly, all the patients in our series were female.

Though the number of patients in this series and in the literature is small, we would like to reemphasize that the association of CHD and C-S syndrome is more than just chance occurrence. Therefore, patients found to have C-S syndrome should be further studied for cardiac anomalies.

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