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Килевидная деформация грудной клетки по «верхнему» типу (синдром Куррарино–Сильвермана): клинический случай

D. Mannatrizio[1], G. Fascia[1], G. Guglielmi[1, 2]

[1] Department of Clinical and Experimental Medicine, Foggia University School of Medicine, Фоджа, Италия
[2] Radiology Unit, Barletta University Hospital, Барлетта, Италия

АННОТАЦИЯ

Преждевременное слияние некоторых центров оссификации грудины и сращение манубриостернального сочленения приводят к редкой форме деформации грудной клетки, называемую синдромом Куррарино–Сильвермана. У пациентов наблюдается аномально короткая грудина со смещением вперёд в области манубриостернального сочленения. Наиболее часто сочетается с сердечно-лёгочными заболеваниями и деформациями позвоночника. Подобную аномалию также ассоциируют с синдромами Нунан и Тернера.

В статье представлен случай 66-летней пациентки, обратившейся в клинику для прохождения повторной компьютерной томографии после операции и химиотерапии по поводу рака молочной железы, с жалобами на частые ежегодные эпизоды одышки, кашля, бронхита, более выраженные в детстве. Результаты компьютерной томографии показали отсутствие метастатических поражений и других сопутствующих заболеваний, за исключением редкой формы деформации передней грудной стенки, так называемой килевидной деформации верхней части грудной клетки (pectus carinatum — *верхний киль*) по хондроманубриальному типу. Угол в дорсальном направлении составлял 130°, длина грудины 9 см без вдавления в нижней трети.

Ключевые слова: килевидная деформация грудной клетки; компьютерная томография; грудина; деформация костей.

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"Superior Pectus Carinatum" (Currarino–Silverman Syndrome) in a 66-year-old woman: a case report

Domenico Mannatrizio[1], Giacomo Fascia[1], Giuseppe Guglielmi[1, 2]

[1] Department of Clinical and Experimental Medicine, Foggia University School of Medicine, Foggia, Italy
[2] Radiology Unit, Barletta University Hospital, Barletta, Italy

ABSTRACT

The premature fusion of some of the sternal ossification centers and the obliteration of the manubrio-sternal joint caused a rare deformity called Currarino–Silverman syndrome. Patients present an abnormally short sternum with a forward angulation at the manubrio-sternal junction. Cardiopulmonary diseases and spinal deformities are the most frequent related disorders. It was also described as a component of Turner's and Noonan's syndromes.

Herein, we present the case of a 66-year-old woman who presented to our clinic for follow-up computed tomography after surgery and chemotherapy for breast cancer with frequent episodes of dyspnea, wheezing, bronchitis, and mild dyspnea annually, which was more frequent during childhood. Computed tomography showed the absence of metastatic lesions and other accompanying diseases, except for a rare deformity of the anterior chest wall, the so-called, a "superior" pectus carinatum, a chondromanubrial deformity with a dorsal-open angle of 130º, and a sternum body length of 9 cm, which is not depressed in the lower third.

Keywords: pectus carinatum; computed tomography; sternum; bone deformity.

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一例66岁女性“上部鸡胸”（Currarino-Silverman综合征）病例

Domenico Mannatrizio[1], Giacomo Fascia[1], Giuseppe Guglielmi[1, 2]

[1] Department of Clinical and Experimental Medicine, Foggia University School of Medicine, Foggia, Italy
[2] Radiology Unit, Barletta University Hospital, Barletta, Italy

摘要

Currarino-Silverman综合征是一种罕见的畸形，由一些胸骨骨化中心过早融合和胸骨柄-胸骨关节闭塞导致。患者胸骨异常短小，胸骨柄-胸骨连接处成角向前突出。最常见的相关疾病有心肺疾病和脊柱畸形。还被描述为特纳综合征和努南综合征的组成部分。在此，我们介绍了1例66岁女性患者，其在因乳腺癌接受手术和化疗后到我们诊所进行计算机断层成像（CT）随访，其每年频繁出现呼吸困难、喘息、支气管炎和轻度呼吸困难，这些症状在儿童期更为频繁。CT显示无转移病灶和其他伴随疾病，仅发现罕见的前胸壁畸形，即“上部”鸡胸，其软骨柄畸形，背侧张开角130°，胸骨体长9cm，下三分之一处未凹陷。

关键词： 鸡胸；CT；胸骨；骨畸形。

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BACKGROUND

The most common congenital chest wall malformations (CWMs) are pectus excavatum (PE) and pectus carinatum (PC). These anomalies present during the first years of life and occur during childhood. PC is less frequent than PE, and it affects 5%–15% of all patients with CWMs and occurs in approximately 1:1000 to 1:10000 of all live births, with a male predominance (4:1) (1) (2). The mild forms are more frequent than the severe forms of PC (3). The chondromanubrial type was first described by Guido Currarino and Frederic Silverman in 1958 (4).

The real etiopathogenesis of type 2 PC is still unknown, but the most plausible hypothesis is that the deformity is caused by an excessive growth of costal cartilage secondary to genetic factors, which results in a sternal deformity without sternal pathology (5).

A short, solid sternum with prominent outward protrusion and bilateral deformity of the second to fifth costal cartilages that form an acute intercostal angle is the pathognomonic aspect of this syndrome (5). The xiphoid process is usually directed forward, even if it can be also absent. In very rare cases, a "superior or chondromanubrial PC" has a normal length, and the sternum is not depressed in the lower third.

Degenerative changes in hyaline cartilages, atypical fibrils, reduced number of chondrocytes, and thin periosteum are observed microscopically (6).

DESCRIPTION OF THE CASE

A 66-year-old woman presented to our clinic to undergo a follow-up computed tomography (CT) after surgery and chemotherapy for breast cancer. She also reported 3–4 episodes of bronchitis, wheezing, and mild dyspnea annually, which was more frequent during her childhood.

She underwent CT of the chest and abdomen with contrast enhancement, which showed the absence of metastatic lesions and any other respiratory tract disorders but revealed a chondromanubrial deformity with a dorsal-open angle of 130° and a sternum body length of 9 cm, which was not depressed in the lower third. Dorsal kyphosis was also present.

During the anamnesis, her CWM was not evident because she was dressed up with a turtleneck sweater that covered her chest.

She reported having an unknown inborn CWM, which was first noted at the age of 3 and progressed until the age of 13 years, accompanied by dyspnea and wheezing that accentuated during exercise and recurrent respiratory infections.

Moreover, the aesthetic appearance of her chest had psychological implications in her childhood, such as having strange feeling among her schoolmates, insecurity, withdrawal, and avoidance of all sports activities that require exposure of the chest. She has passed her childhood hiding her health problem.

DISCUSSION

The Currarino–Silverman (CS) syndrome is an extremely rare congenital deformity of the sternum.

It is also known by the terms "pouter pigeon chest," "chondromanubrial deformity," "type 2 pectus carinatum," and "pectus arcuatum." According to the original paper of Currarino and Silverman (4), this rare developmental anomaly is characterized by prominent sternal angulation with a decrease in length as a result of congenital complete non-segmentation or premature synostosis of the sternum.

The theory of a congenital etiology by gene downregulation is predominant (7-11). Essentially, a family history of CWMs is present in approximately 25% of the patients (1).

An abnormality of the differentiation of anterior segment mesenchymal cells and abnormal migration of mesenchymal cardiac precursors to the endothelial heart tube at the time of cardiogenesis may result in defects of endocardial cushion, sternum, and aortic arch derivatives (7). This syndrome is often combined with congenital heart defects and spinal abnormalities (kyphosis, scoliosis, and kyphoscoliosis), and it was also described as a component of Turner's and Noonan's syndromes (8).

Frequent confusion of CS syndrome with PE deformity is still an issue since one-third of patients present with concomitant mild to moderate depression of the lower third of the sternum (9).

Indeed, CS syndrome can be easily mistaken with PE, as both deformities appear in an almost similar fashion, but surgical approaches are very different. A distinctive feature of a PE deformity is the beginning of the sternal depression at the angle of Louis that becomes progressively deeper toward the xiphoid process with distorted and elongated cartilages (10). The angle of Louis must be less than 110° to be considered a true depression (5). Thus, confusion about classification is still an actual issue.

A uniform classification is a basis for surgical treatment and assessment of its short- and long-term results.

In 2006, Acastello classified CWMs based on the site of the defect's origin (type 1, cartilaginous; type 2, costal; type 3, chondrocostal; type 4, sternal; type 5, clavicle-scapular) and attributed CS syndrome to the cartilaginous type 2 (superior) PC. Further, Torre *et al.* distinguished superior PC into two types:

- Type 1: The "inferior" or "chondrogladiolar" is the most frequent type, in which the sternal protrusion is located in the inferior or mid sternum and the last ribs can be slightly or severely depressed on lateral aspects.
- Type 2: The "superior" or "chondromanubrial" is the less frequent type, which has been further divided in

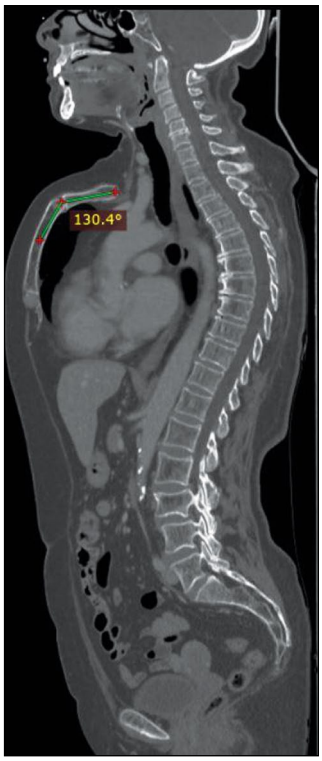

Fig 1. Sagittal whole body computed tomography scan showing an arching sternum and a chondromanubrial deformity with a dorsal-open angle of 130°.

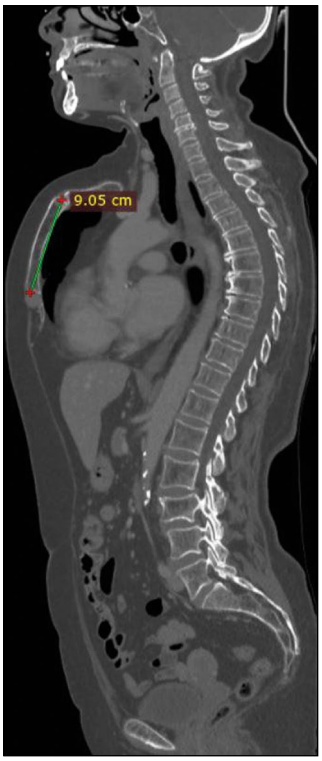

Fig 2. Sagittal whole body computed tomography scan showing a sternum body length of 9 cm and dorsal kyphosis.

two types depending on the external aspect. The first type is characterized by a superior PC with an inferior PE in which the sternum is S-shaped on a lateral view. This anomaly is classified as cartilaginous anomalies and called type II. Although this anomaly is considered a cartilaginous anomaly and a type II PC in the Acastello classification, Torre *et al.* classified it as part of sternal anomalies because of the sternal origin of the anomaly. The second type is a "superior PC" without the typical features of the CS syndrome. The sternum has a normal length and is not depressed in the lower third. This anomaly is probably due to a cartilage anomaly similarly to inferior PC. Torre *et al.* proposed to use the term "superior PC" only for this type and to include this anomaly in the first category (cartilaginous anomalies) of CWM classification. This entity is extremely rare.

Most patients with CS syndrome are asymptomatic; therefore, surgical correction is optional, and different opinions exist about the ideal age for surgical correction. Asthma and chronic bronchitis, which occur in 16% of the patients, are the most common associated concomitant diseases, responsible for bronchial and pulmonary symptoms (11).

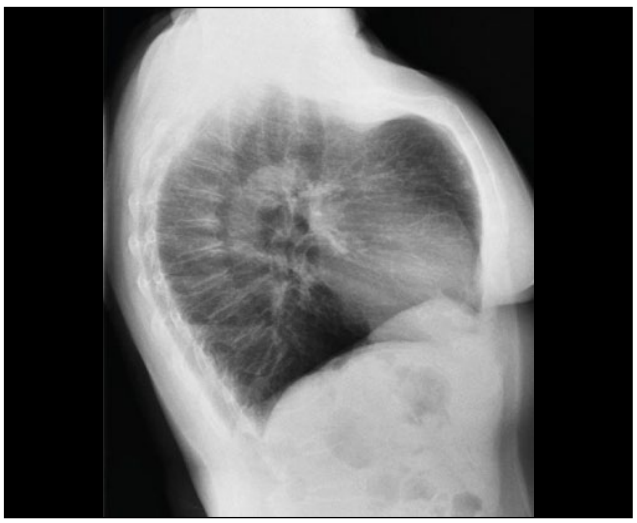

Fig 3. Lateral plain radiograph appearance of a type 2 pectus carinatum.

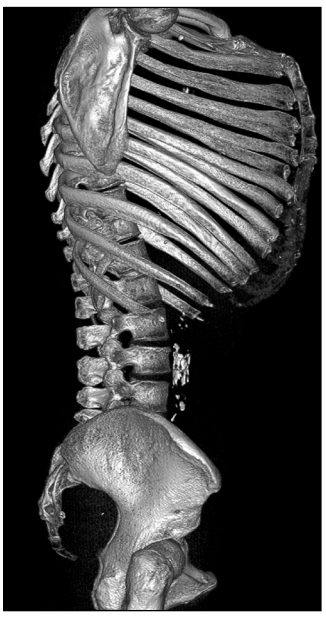

Fig 4. Computed tomography reconstruction of the rib cage showing that the sternum has a normal length and is not depressed in the lower third.

Kyphosis is present, to a greater or lesser degree, in almost all patients. Pain or tenderness at the protrusion site, decreased endurance, or palpitations can be present. Limitations at work and in sports and underachievement in school, in the absence of respiratory and heart diseases, should be attributed to emotional alterations.

A strong correlation of inborn cardiac pathology with CS syndrome has been reported, such as VSD, patent ductus arteriosus, atrial septal defect, tetralogy of Fallot, transposition of the great arteries, and coarctation of the aorta.

Chest CT with three-dimensional reconstruction, or magnetic resonance imaging in children with altered tissue density and radiation concern, is the best preoperative imaging for the evaluation of patients with CS. It allows the differential diagnosis with other pectus deformities, determination of the exact angles of the costal cartilages to the sternum, and enhances surgical planning.

In comparison to PE, several less invasive techniques have been developed for the correction of PC, including the Abramson procedure and its modifications, along with nonsurgical options such as observation, orthotic bracing, and dynamic compression (2) (12). However, due to the extreme rarity of the disease, challenging deformity, and variable anatomy of a fused sternum, there are no clear guidelines in the treatment approaches.

This unique growth pattern contributes to an almost universal failure of conservative treatment options, such as the vacuum bell or a compressive orthosis. The best surgical option remains the relatively aggressive Ravitch-type procedure with multi-level wedge osteotomy (12) (13), allowing for the achievement of a satisfactory outcome.

Given the rarity of the deformity, surgical correction should be completed by a multidisciplinary team, including thoracic reconstructive surgeons with experience in pectus deformities. The preferred age for correction is late puberty or adulthood, as cartilage resection will be performed when the rib growth ends (1) (14) (15). It is important to keep in mind the potential for thoracic dystrophy should cartilage resection be performed at a young age or too extensively. Thus, other studies have reported the preferable age of 5–7 years or early adolescence (5) (16). Some patients who refused to undergo surgery turned to bodybuilding to improve and define the musculature around the CWM to minimize the appearance of protrusion. Even if this approach does not correct the abnormality, it can improve self-esteem and confidence. For female patients, one possibility to make protrusion less noticeable and improve the appearance of the chest is breast augmentation (17).

CONCLUSION

Although a rare condition, CS can be easily diagnosed through its typical radiological findings. Therefore, clinicians must be aware of and recognize this imaging pattern to make an accurate diagnosis and prevent further examination and aggressive treatment.

ADDITIONAL INFORMATION

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Consent for publication. Written consent was obtained from the patient for publication of relevant medical information within the manuscript.

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AUTHORS' INFO

*** Guseppe Guglielmi,** MD, Professor;
address: Viale L. Pinto 1, 71121 Foggia, Italy;
ORCID: http://orcid.org/0000-0002-4325-8330;
e-mail: giuseppe.guglielmi@unifg.it

Domenico Mannatrizio, MD;
ORCID: http://orcid.org/ 0000-0003-3365-7132;
e-mail: dr.mannatrizio@gmail.com

Giacomo Fascia, MD;
ORCID: http://orcid.org/ 0000-0001-5244-5093;
e-mail giacomo.fascia@unifg.it

ОБ АВТОРАХ

*** Guglielmi Giuseppe,** MD, Professor;
адрес: Viale L. Pinto 1, 71121, Фоджа, Италия;
ORCID: http://orcid.org/0000-0002-4325-8330;
e-mail: giuseppe.guglielmi@unifg.it

Mannatrizio Domenico, MD;
ORCID: http://orcid.org/ 0000-0003-3365-7132;
e-mail: dr.mannatrizio@gmail.com

Fascia Giacomo, MD;
ORCID: http://orcid.org/ 0000-0001-5244-5093;
e-mail giacomo.fascia@unifg.it

* Corresponding author / Автор, ответственный за переписку