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

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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

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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

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简评

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9 cm，下三分之一处未凹陷。

关键词：鸡胸；CT；胸骨；骨畸形。

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绪论

最常见的先天性胸壁畸形类型是漏斗形（pectus excavatum）和龙骨形(pectus carinatum)畸形。这些异常发生在儿童时期，并在生命早期表现出来。龙骨畸形比漏斗胸少见，在所有胸壁畸形患者中约占5-15%，在所有新生儿中为1:1000-1:10,000，主要是男孩（4:1)[1,2]。同时，轻度龙骨畸形比重度龙骨畸形更常见[3]。G. Currarino和F. Silverman在1958年首次描述了软骨管型龙骨畸形[4]。

2型龙骨畸形的病因尚不清楚，但根据最似乎可信的假设，在遗传因素的影响下肋软骨过度生长导致胸骨畸形，导致此类疾病没有严重的病理[5]。

Currarino-Silverman综合征的特征是胸骨短而大，有明显的向外突出和II-V肋软骨的双侧畸形，形成急性肋间角[5]。剑突通常是向前的，但也可能没有剑突。在非常罕见的情况下，对于“上”或软骨型龙骨畸形，胸骨尺寸正常，下三分之一没有凹陷。

显微镜检查显示透明软骨退行性变化、非典型原纤维、软骨细胞数量减少和骨膜变薄[6]。

病例报告

患者，女性，66岁，在乳腺癌手术和化疗后入院进行重复计算机断层扫描。主诉咳嗽发作、支气管炎和中度喘息(每年3-4次），这些症状在儿童时期更为明显。

胸部和腹部对比增强计算机断层扫描结果显示没有转移性病变和任何其他呼吸道疾病，但显示软骨瘤类型的胸部龙骨畸形。背侧角度为130度，胸骨长为9 cm，下三分之一无凹陷。还注意到背侧后凸（图1-4）。

在收集病史时，胸壁的变形在视觉上并不明显，因为患者的胸部被一件带有大领子的毛衣覆盖。该患者报告了病因不明的先天性病变，3岁时首次发现，并一直发展到13岁，伴有气短、喘鸣，运动时加重，并反复出现呼吸道感染。此外，儿童时期，胸部的外观对患者有负面的心里影响（在同学中感到尴尬、独立、避免所有需要暴露上身的运动）。患者在整个童年时期都在隐藏自己的健康问题。

讨论

Currarino-Silverman综合征是一种极为罕见的先天性胸部畸形，又称“鸽胸”、“软骨瘤畸形”、“2型龙骨畸形”、“漏斗胸”。据G. Currarino和F. Silverman[4]，这种先天性异常的特征是由于完全没有分割或过早的骨结合，胸骨的长度明显弯曲和缩短。在关于疾病起源的假设中，基因先天性下调的假设占主导地位[7-11]。例如，大约25%的患者具有胸壁畸形的遗传易感性[1]。

在心脏发生过程中，前段间充质细胞分化受损和间充质心脏祖细胞异常迁移到内皮心管可导致心房间隔、胸骨和主动脉弓缺损[7]。Currarino-Silverman综合征通常与先天性心脏缺陷和脊柱异常（脊柱后凸、脊柱侧凸、脊椎后侧凸）合并，并与努南氏综合征和特纳氏综合征有关[8]。

Currarino-Silverman综合征经常与漏斗胸混淆，因为1/3的患者伴有胸骨下三分之一的轻度或中度凹陷[9]。实际上，漏斗胸可能被误认为是Currarino-Silverman综合征，因为这两种类型的畸形外观几乎相同，但需要完全不同的手术方法。漏斗形畸形的一个明显特征是胸骨在刘易斯角水平的凹陷，逐渐向剑突进展，肋骨软骨变形和拉长[10]。刘易斯角必须至少为110º，胸骨凹陷才算真实[5]。因此，对畸形分类的混淆仍然是目前的一个紧迫的问题。

统一的分类是手术治疗成功和评价其近期和远期效果的基础。

Acastello在2006年提出根据最大畸形的位置（软骨、骨骼、软骨-肋骨、胸骨、锁骨-肩胛骨）将胸壁畸形分为5种类型。在这个分类中，作者认为Currarino-Silvermann综合征是一种软骨发育障碍，并将其归为2型（“上”）龙骨畸形。随后，M. Torre及其合著者将龙骨畸形分为两种类型：

- 1型——“下部”或“软骨瘤”——最常见的，其中胸骨的下部或中部出现突出，同时肋骨以轻度或更明显的形式横向凹陷;
- 2型——“上部”——不不太常见，根据畸形的外观又分为两种类型。前者的特征是存在“上”型龙骨畸形与“下”型漏斗形畸形相结合，其中胸骨在横向透视中具有S形。根据Acastello的分类，这种畸形属于软骨（2 型），而M. Torre及其合著者将此类疾病归因于由于畸形发生部位而导致的胸骨发育异常。第二种是“上”型的龙骨状畸形，没有典型的Currarino-Silverman综合征的症状，即，胸骨长度正常且下三分之一处没有凹陷。这种异常可能与软骨组织的发育障碍有关，类似于“下”型的龙骨畸形。M.Torre及其合著者建议将此类畸形称为“上型龙骨畸形”，并将其称为第1类（软骨性）胸壁畸形，这种畸形极为罕见。

在大多数患者中，该疾病是无症状的，因此并不总是需要手术。此外，专家对手术最佳年龄的意见各不相同。肺部和支气管最常见的伴随疾病（16%）是哮喘和慢性支气管炎[11]。几乎所有患者都或多或少有驼背。突出部位可能有疼痛或酸痛、耐力降低或心率加快。在没有呼吸和心脏病的情况下工作和运动的限制，在学校的表现不佳，更可能是由于心理因素造成的。Currarino-Silverman综合征与心血管系统病理学（室间隔缺损或房间隔缺损、动脉导管未闭、法洛四联症、大动脉转位、主动脉缩窄）的强相关性也有报道。

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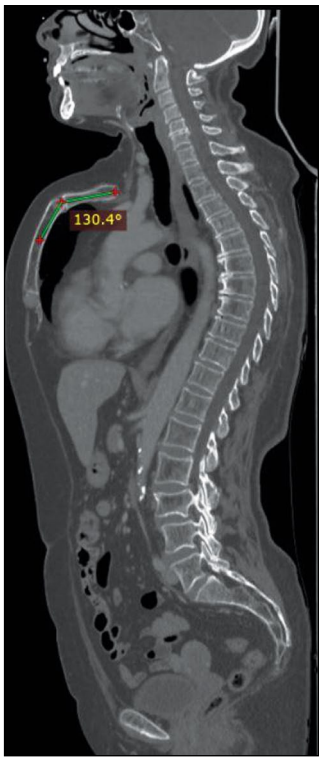

图1. 全身计算机断层扫描，矢状切面：弓形胸骨，软骨房畸形，背角130º。

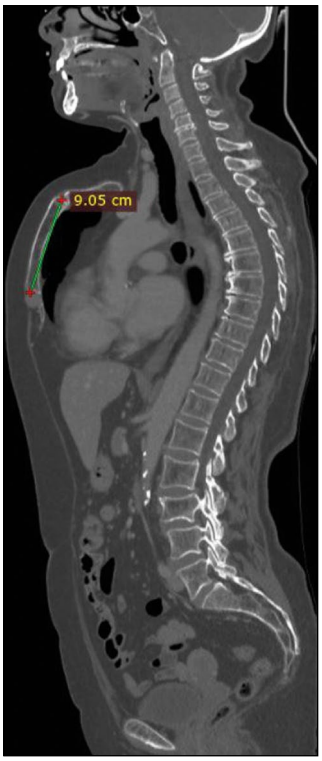

图2. 全身计算机断层扫描，矢状切面：胸骨长9 cm，背侧后凸。

评估Currarino-Silverman综合征患者的最佳术前成像方式是3D重建胸部计算机断层扫描或磁共振成像，特别是在组织密度改变和有辐射负载危险的儿童中。这些方法可以与其他畸形进行鉴别诊断，确定肋软骨与胸骨的准确倾斜角度，并决定进一步的治疗。

与漏斗胸相比，已经开发了几种微创技术来矫正龙骨畸形，包括Abramson手术及其修改，以及非手术选择，如观察、骨科固定和动态压缩 [2,12]。但由于该病极为罕见和复杂，以及变形胸骨的解剖结构多变，目前对于选择最佳治疗方法尚无统一建议。由于疾病的独特表现，诸如真空杯和压缩矫形器之类的保守疗法往往没有积极的效果。

最好的手术治疗方案仍然是相对激进的Ravitch手术，采用多级楔形截骨术，可以取得满意的效果[12，13]。鉴于该疾病的罕见性，手术应由具有胸部畸形治疗经验的专门从事胸部重建手术的多学科团队进行。矫正的首选年龄是青春期后期或成熟期，因为软骨切除术仅在肋骨生长完成后进行 [1，14，15]。同样重要的是要记住，年轻时的手术或过于广泛的软骨切除会导致胸廓营养不良的发生。因此，根据一些研究，手术的首选年龄是5-7岁或青春期早期[5，16]。一

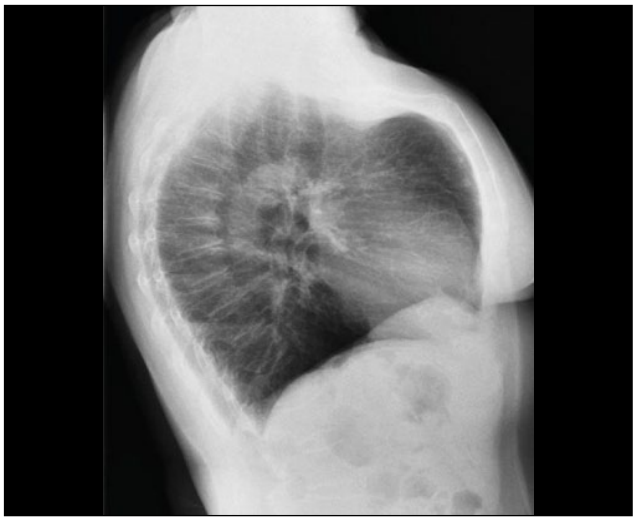

图3. X线平片，侧位片：第2型胸部龙骨畸形。

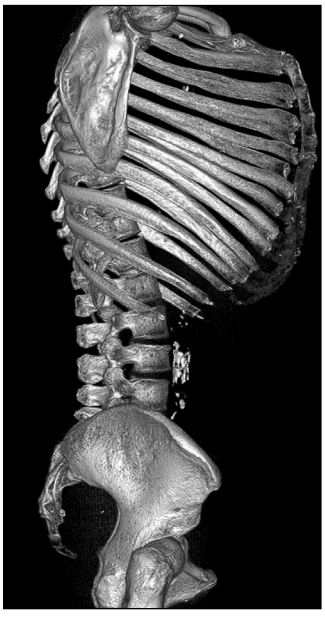

图4. 胸部计算机断层扫描：胸骨长度正常，下三分之一无凹陷。

些拒绝手术的患者转向健美运动，这使他们能够在畸形区域锻炼肌肉，从而最大限度地减少突出的外部表现。无论如何，这种方法可以增加自尊和自信，即使它并不能消除缺陷本身。对于女性患者，使突出不那么明显和改善乳房外观的选择之一是乳房整型[17–22]。

结论

虽然Currarino-Silvermann综合征是一种罕见的疾病，但根据典型的放射学图片，它很容易被诊断。临床医生必须了解并能够识别疾病的图片，以便准确诊断，从而避免进一步检查和侵袭性疗法。

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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