

心脏右心房未分化肉瘤1例并文献回顾

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收稿日期: 2024年1月18日; 录用日期: 2024年2月11日; 发布日期: 2024年2月19日

摘要

目的: 探讨原发性心脏未分化肉瘤的临床症状、影像学检查及病理表现特点。方法: 对温州医科大学附属衢州医院收治的1例心脏未分化肉瘤患者的临床资料进行回顾性分析, 并以“心脏未分化肉瘤”为关键词, 检索中国知网、万方数据库和中华医学期刊全文数据库, 以“cardiac undifferentiated sarcoma”为关键词检索PubMed数据库, 检索时间自建库至2023年12月, 总结原发性心脏未分化肉瘤的临床症状、影像学检查及病理表现特点。结果: 检索国内外文献共收集到原发性心脏未分化肉瘤47例, 加上本例共48例。原发性心脏未分化肉瘤患者临床表现为进行性呼吸困难、双下肢水肿、乏力、咳嗽咳痰、胸痛。发病部位位于右心房5例, 右心室9例, 左心房19例, 左心室8例, 心包2例, 未明确3例。最小年龄为1岁, 最大发病年龄为87岁, 其中50~70岁发病23例。结论: 心脏未分化肉瘤患者的临床表现多样, 不具有典型性, 以进行性呼吸困难最多见; 超声心动图对于诊断具有重要意义, 最终确诊仍需病理检查。

关键词

心脏, 未分化肉瘤, 诊断, 影像检查, 临床病理

Undifferentiated Sarcoma of the Right Atrium: A Case Report and Literature Review

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Received: Jan. 18th, 2024; accepted: Feb. 11th, 2024; published: Feb. 19th, 2024

Abstract

Objective: To investigate the clinical symptoms, imaging and pathological features of primary car-

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diac undifferentiated sarcoma. Methods: The clinical data of one patient with cardiac undifferentiated sarcoma admitted to Quzhou Hospital Affiliated to Wenzhou Medical University were retrospectively analyzed. "Cardiac undifferentiated sarcoma" was used as the key word to search China National Knowledge Infrastructure (CNKI), Wanfang Database and Chinese Medical Journal Full-text Database. "Cardiac undifferentiated sarcoma" was used as the key word to search PubMed database. The search time was from the establishment of the database to December 2023. The clinical symptoms, imaging examination and pathological features of primary cardiac undifferentiated sarcoma were summarized. **Results:** A total of 47 cases of primary cardiac undifferentiated sarcoma were collected from domestic and foreign literatures; including this example, there are a total of 48 cases. The clinical manifestations of patients with primary cardiac undifferentiated sarcoma were progressive dyspnea, edema of both lower limbs, fatigue, cough and expectoration, and chest pain. The location of the disease was located in the right atrium in 5 cases, right ventricle in 9 cases, left atrium in 19 cases, left ventricle in 8 cases, pericardium in 2 cases, and unclear in 3 cases. The minimum age was 1 year old, and the maximum age of onset was 87 years old, including 23 cases of 50~70 years old. **Conclusions:** The clinical manifestations of patients with cardiac undifferentiated sarcoma are diverse and not typical. Progressive dyspnea was the most common symptom; echocardiography is of great significance for diagnosis, and final diagnosis still requires pathological examination.

Keywords

Cardiac, Undifferentiated Sarcoma, Diagnosis, Image Examination, Clinical Pathology

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1. 引言

原发性心脏肿瘤在临床中较为罕见，发生率仅为 0.02%，其中只有 25%为恶性[1]。该肿瘤最常见的恶性组织学类型是血管肉瘤，以未分化多形性肉瘤恶性程度最高[2] [3]。原发性心脏未分化肉瘤表现隐匿，不具有典型临床症状，临床认识不足，目前国内仅见 13 例报道。2023 年 12 月我院胸心外科收治 1 例心脏恶性肿瘤患者，病理结果显示为未分化型肉瘤。本文对国内外已报道的心脏未分化肉瘤文献进行总结，以提高临床医生对心脏未分化肉瘤的诊断及认识。

2. 病例资料

2.1. 本例资料

患者，男，67 岁，既往渗出性心包炎、阵发性房颤、心功能不全病史。因“胸闷气促 3 月”于当地医院就诊，查胸部增强 CT 提示：“纵隔内团块合并钙化”，建议转至上级医院治疗，遂于 2023-12-02 入住我院。患者入院后稍感头晕胸闷，家属诉患者曾有面部浮肿及双下肢水肿，无畏寒发热，无咳嗽咳痰等其他明显不适。体格检查：脉搏 94 次/分，血压 140/80 mmHg，颈静脉无怒张，双肺呼吸音粗，可闻及湿性啰音；心前区无隆起，心尖搏动范围正常，未触及震颤和心包摩擦音，心脏叩诊浊音界正常，律齐，各瓣膜听诊区未听及心脏杂音和额外心音，双下肢无水肿。入院完善相关辅助检查，B-型尿钠肽 182 pg/ml，血沉 17 mm/h，血常规、尿常规、血清生化、细胞免疫功能检测等未见异常。肿瘤标记物：AFP、CEA、CA199、CA72-4、鳞状上皮细胞癌抗原等均阴性。心脏彩超：心脏右房旁可见一混合回声

团，位置较深，无法完全测量，内可见多发强回声，与房壁分界欠清；主动脉窦部增宽(图 1)。腹部彩超：肝回声增粗；脾周积液；腹水，较宽处位于脾周，深约 1.2 cm。冠脉 CTA：左前降支近段钙化斑块，管腔轻-中度狭窄；回旋支近段混合斑块，管腔轻度狭窄；纵隔肿块伴钙化(图 2)。患者于 2023-12-06 在全身静脉麻醉下行纵隔肿物穿刺活检，送病理，结果示：少量间叶源性恶性肿瘤组织，可参照梭形细胞/多型未分化肉瘤处理(图 3)。免疫组化结果：CDK4 (-)，P16 (+)，MDM2 (-)，Desmin (少量+)，SMA (部分+)，CD34 (-)，SATB2 (-)，H3K27Me3 (+)。结合患者症状体征、相关辅助检查结果，初步考虑为“右心房原发性未分化肉瘤”。家属考虑患者高龄，一般情况较差，已放弃治疗并自动出院。



Figure 1. Strong echogenic mass adjacent to the right atrium
图 1. 右房旁强回声团

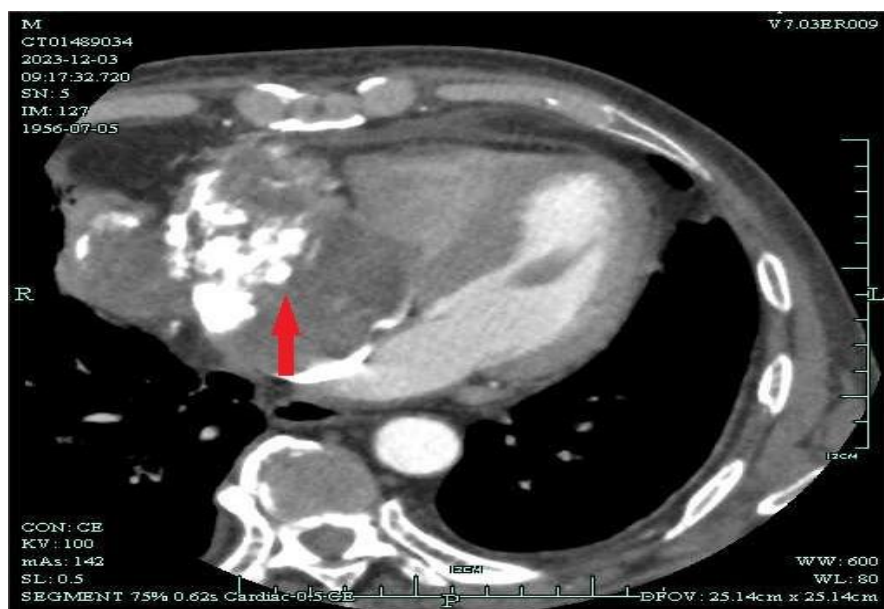


Figure 2. A mass with calcification can be seen in the mediastinum
图 2. 纵隔内可见肿块伴钙化

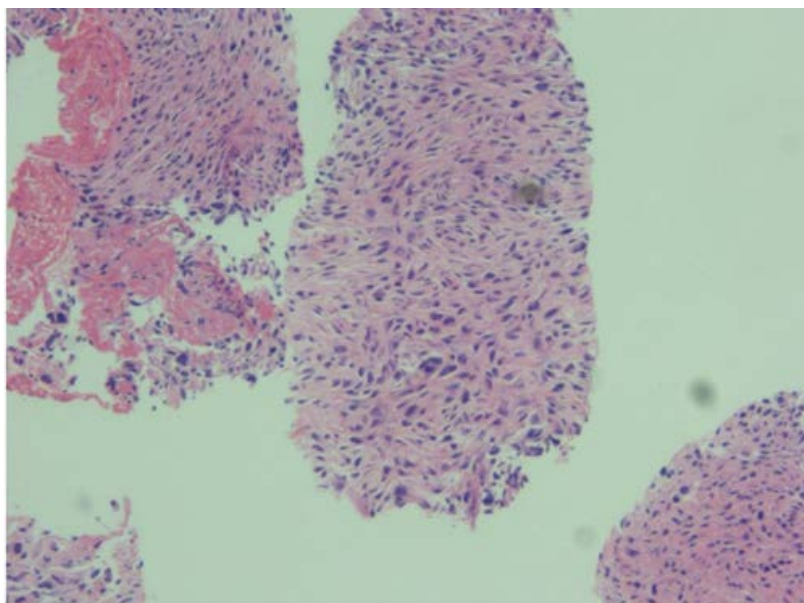


Figure 3. Pathological biopsy of tumors
图 3. 肿物病理活检

2.2. 文献复习

以“cardiac undifferentiated sarcoma”为关键词检索 PubMed 数据库收录的文献，以“心脏未分化肉瘤”为关键词在中国期刊全文数据库，万方数据库和中国维普书库检索建库至 2023 年 12 月收录的文献。共检索到原发性心脏未分化肉瘤 43 篇[1] [2] [4]-[44]，其中中文文献 10 篇(13 例)，外文文献 33 篇(34 例)，加上本例共 48 例。主要临床症状包括进行性呼吸困难 23 例，双下肢水肿 9 例，乏力 7 例，咳嗽咳痰 6 例，胸痛 4 例。发病年龄在各年龄段均有分布，最小年龄为 1 岁，最大发病年龄为 87 岁，其中 50~70 岁发病 23 例。发病部位位于右心房 5 例，右心室 9 例，左心房 19 例，左心室 8 例，心包 2 例，未明确 3 例。

我国患者 13 例[2] [36]-[44]，其中有 8 例临床症状伴有胸闷气短，3 例患者出现咳嗽咳痰症状，尚未见临床症状为胸痛的报道。

3. 讨论

心脏未分化型肉瘤既往病例报道中的患者临床表现不具有特异性，其症状与肿瘤发生位置、大小、活动度、与周围组织关系相关，常见表现为胸痛、呼吸困难和头晕等血流梗阻症状[32]。Bouma [45]等人研究表明，7%以上的患者诊断时已出现全身转移。当肿瘤侵及肺部时可有咳嗽咳痰等表现，若同时合并胸腔积液，可伴有胸闷心悸等呼吸困难表现，偶有患者出现痰中带血[36] [41]。

影像学检查方面，超声心动图对于心腔的占位性病变具有重要意义。超声首次鉴别诊断原发性心脏恶性肿瘤具有一定的困难[46]。由于心脏肿瘤发病率较低，有研究指出有超过 30%的患者于体检中偶然发现，且部分心脏肿瘤来源复杂，如右心肿瘤可于静脉系统转移而来，可为原发性或转移性。对于心脏的转移性肿瘤，其回声表现丰富多样，仅依靠超声难以做出准确诊断[47]。超声表现需与心脏黏液瘤等良性肿物相鉴别：黏液瘤 20%发生在右心房，多为单发，常见表现为心腔内中等回声团块，带蒂，肿瘤在心房室之间具有往返活动。恶性未分化型肉瘤超声表现主要为具有宽阔基底，活动度差，与房室面接触范围广的实质性包块，其回声不规则，易合并胸腔积液或心包积液[37] [48]。根据肿瘤的生长速度、基底

情况、活动度、侵及心肌层次及周围组织情况，超声可对肿瘤良恶性程度做出初步的诊断。但由于超声心动图为二维图像，缺乏立体感，往往需要同 CT 或 MRI 影像结合来确定肿瘤与周围组织的毗邻关系。CT 或 MRI 除可直观显示肿瘤的起始部位、肿瘤密度、心脏的灌注情况外，又可显示全身情况，排除其他部位转移性肿瘤的可能[49]。未分化型肉瘤的 CT 表现不具有特征性，多表现为不均匀密度影。心脏 MRI 是区分恶性病变和评估肿瘤可切除性的金标准补充检查，主要表现为混杂信号，T1WI 以等或低信号多见，T2WI 上以高信号为主[50]。最终确诊仍需组织病理检查。

心脏未分化型肉瘤多发生于心腔内，有报道称 81% 的未分化肉瘤位于右心房[41]。肿瘤呈息肉状生长，基底部呈广基态伴浸润；大体观呈现灰白色，伴出血时可呈现灰红色，部分可见坏死及囊性变区域，质韧；镜下突出特征为肿瘤细胞具有明显的多形性，核仁突出，可见梭形细胞或肌纤维细胞呈车轮状排列，偶见鱼骨样或片状图案，未分化多形性肉瘤可由肌纤维细胞、间叶细胞及组织细胞混合构成[33]。未分化型肉瘤免疫组化表现无明显特征性，部分病例可表现 vimentin、 α 1-AT、actin 和 lysozyme 阳性[2]。MDM2 是一种原癌基因，是心脏内膜肉瘤中最常见的突变基因之一，史玉洁[43]等人报道了 4 例心脏未分化型肉瘤患者，有 3 例存在 MDM2 免疫组化阳性。此外，法国 Neuville 通过 FISH 和实时定量 PCR 检测证实 MDM2 基因扩增仅存在于未分化肉瘤中。因此对于高度疑似心脏未分化型肉瘤的患者，可进一步行探针扩增技术检测。

综上，本研究对原发性心脏未分化型肉瘤的临床表现、影像学检查及病理诊断方面做出报道并进行文献复习。心脏未分化型肉瘤发病率较低，临床缺乏典型表现，早期诊断十分困难，误诊率较高，如发现不明原因的气促、胸闷等血流梗阻症状，应高度怀疑心脏恶性肿瘤的存在。心脏超声、MRI 等影像学检查是诊断心脏未分化型肉瘤的重要方法，病理活检和免疫组化检查对于心脏未分化型肉瘤的诊断是十分有必要的。

基金项目

衢州市指导性科技项目(2018071)。

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