

·CASE ANALYSIS·

·临床病例讨论·

巨块型肝血管周上皮样细胞肿瘤误诊1例

晏斯婕, 张立媛, 杨翼腴, 谢家媛, 张波

(中南大学湘雅医院超声影像科, 长沙 410008)

[摘要] 血管周上皮样细胞肿瘤(perivascular epithelioid cell tumor, PEComa)是一种罕见的间叶源性肿瘤, 巨块型(最大径 ≥ 10 cm)肝PEComa则更为罕见。本文报告1例被误诊为纤维板层型肝细胞癌的巨块型肝PEComa病例。患者为中年女性, 因上腹不适就诊, 2025年8月21日以“肝占位性病变”收入中南大学湘雅医院。患者既往无慢性肝病病史, 完善相关实验室检查无明显异常, 术前多种影像学检查提示肝巨大肿块, 误诊为纤维板层型肝细胞癌。手术完整切除(肿块大小为14.3 cm $\times$ 13.5 cm $\times$ 10.0 cm), 最终经病理及免疫组织化学确诊为肝PEComa。术后随访6个月, 患者无明显不适, 未见复发征象。肝PEComa因缺乏特异性的临床表现及影像学特征, 诊断较为困难。对病例的回顾性进行分析, 探讨误诊原因, 总结鉴别诊断要点, 可在加深临床及影像科医师对该病的认识, 为术前影像学诊断提供更多参考依据, 从而降低误诊率、优化治疗决策, 最终改善患者预后。

[关键词] 肝血管周上皮样细胞肿瘤; 纤维板层型肝细胞癌; 误诊; 影像学特征; 病理诊断

Misdiagnosis of a giant hepatic perivascular epithelioid cell tumor: A case report

YAN Sijie, ZHANG Liyuan, YANG Yitian, XIE Jiayuan, ZHANG Bo

(Department of Ultrasound Imaging, Xiangya Hospital, Central South University, Changsha 410008, China)

ABSTRACT

Perivascular epithelioid cell tumor (PEComa) is a rare mesenchymal neoplasm, and giant (maximum diameter is ≥ 10 cm) hepatic PEComa is even more uncommon. We report a case of giant hepatic PEComa that was preoperatively misdiagnosed as fibrolamellar hepatocellular carcinoma. The patient was a middle-aged woman who presented with upper abdominal discomfort and was admitted to Xiangya Hospital of Central South University on August 21, 2025, for evaluation of a hepatic space-occupying lesion. She had no history of chronic liver disease, and relevant laboratory tests showed no significant abnormalities. Multiple preoperative imaging modalities revealed a giant hepatic mass, which was initially diagnosed as fibrolamellar hepatocellular carcinoma. The tumor was completely resected

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第一作者(First author): 晏斯婕, Email: yansijie2624@163.com, ORCID: 0009-0007-2951-1006; 张立媛, Email: zhangliyuan0911@163.com, ORCID: 0009-0009-7157-382X

通信作者(Corresponding author): 张波, 主任医师, Email: zhangbo8095@csu.edu.cn, ORCID: 0000-0001-8012-0560

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(the size of mass was 14.3 cm×13.5 cm×10.0 cm) and the final diagnosis of hepatic PEComa was established by histopathological examination and immunohistochemical analysis. During a 6-month postoperative follow-up period, the patient remained asymptomatic and showed no evidence of recurrence. The diagnosis of hepatic PEComa is challenging because of its lack of specific clinical manifestations and characteristic imaging findings. Through retrospective analysis of this case, the causes of misdiagnosis were explored and the key points for differential diagnosis were summarized. This report aims to improve awareness of hepatic PEComa among clinicians and imaging specialists, provide additional references for preoperative imaging diagnosis, reduce the rate of misdiagnosis, optimize treatment decision-making, and ultimately improve patient outcomes.

KEY WORDS hepatic perivascular epithelioid cell tumor; fibrolamellar hepatocellular carcinoma; misdiagnosis; imaging characteristics; pathological diagnosis

血管周上皮样细胞肿瘤(perivascular epithelioid cell tumor, PEComa)是一类罕见的间叶源性肿瘤,由血管、平滑肌和上皮样细胞以不同比例混合构成,具有恶性潜能。该肿瘤主要发生于子宫、肾等器官^[1],而肝脏PEComa极为罕见,具有独特的组织学形态和免疫表型特征^[2]。肝PEComa影像学及临床表现多样,术前极易误诊为其他肝富血供肿瘤。当影像学表现为边界清晰、无肝硬化背景的较大富血供肿块时,该病尤其易与纤维板层型肝细胞癌相混淆,但目前国内外针对二者鉴别诊断的文献较少,亦缺乏系统的临床病理特征总结。现报告1例巨块型肝PEComa误诊为纤维板层型肝细胞癌的病例,并结合文献分析误诊原因,以期提高临床医师对肝PEComa的认识,减少术前误诊。

1 病例资料

患者,女,52岁,因“上腹不适5 d”就诊于当地医院,CT发现肝右叶巨大肿块,考虑巨块型肝癌伴液化坏死,为进一步诊治,2025年8月21日于中南大学湘雅医院就诊。既往无肝炎病史。体格检查:全腹柔软,无压痛及反跳痛,肝脾肋下未触及,肝区无叩痛。

实验室检查:乙型肝炎表面抗原(hepatitis B surface antigen, HBsAg)为阴性;肿瘤标志物神经内分泌特异性烯醇化酶、癌胚抗原、糖类抗原199、甲胎蛋白(alpha-fetoprotein, AFP)和异常凝血酶原均正常;肝功能及肾功能均正常。

影像学检查:肝磁共振提示肝右叶巨大肿块灶,肝右叶见一大小为12.7 cm×12.5 cm×15.2 cm的混杂信号肿块, T₁加权成像(T₁-weighted imaging, T₁WI)呈等-稍低信号, T₂加权成像(T₂-weighted imaging, T₂WI)呈等-高信号。弥散加权成像(diffusion weighted imaging, DWI)示中央可见片状等-稍高信号区,相应表观弥散系数(apparent diffusion coefficient, ADC)为等-低值,增强模式下动脉期病变明显不均匀强化,其内见大量细小紊乱肿瘤血管影,门静脉期、延迟期较前廓清(较肝实质强化程度减低),中央见不均匀强化及片状无增强区(图1),考虑纤维板层型肝细胞癌。肝常规超声结果(图2A)显示,肝右叶见一大小为15.2 cm×11.3 cm的强弱不等回声肿块,形态不规则,边界不清,分布不均匀,肿块内可见多个小片状强回声区,肝中静脉受压移位。彩色多普勒血流成像(color Doppler flow imaging, CDFI)提示肿块周边可见条状血流信号。超声造影(图2B~2H)示肿块于动脉期呈快速、向心性、不均匀高增强,可见条索状血管影;门静脉期呈等增强;延迟期呈低增强,中央可见片状3期无增强区,提示纤维板层型肝细胞癌可能性大。

行“右半肝切除术+胆囊切除术”,术后病理提示: (肝右叶)单发肿块(大小为14.3 cm×13.5 cm×10.0 cm),瘤细胞伴异型,核分裂象>1个/mm²,并有病理性核分裂;可见灶性坏死,小灶可见瘤组织浸润周围肝实质;见可疑脉管内瘤栓,未见明确神经侵犯(图3)。免疫组织化学:人黑色素瘤黑色素45(human melanoma black 45, HMB45)(+), MelanA(+), S100

(-), 平滑肌肌动蛋白(smooth muscle actin, SMA)(+), AFP(-), 细胞角蛋白(cytokeratin, CK)19(胆管+), 肝细胞抗原(hepatocyte)(-), 磷脂酰肌醇蛋白聚糖-3(glypican-3)(-), CK8/18(-), 肌细胞生成素(myogenin)(-), MYOD1(-), 表皮生长因子受体(epidermal growth factor receptor, EGFR)(弱+),

CD34(血管+), Ki-67(2%+)。特殊染色结果: Masson染色(纤维+), 过碘酸雪夫染色(periodic acid-Schiff stain, PAS)(-)。综合考虑为肝右叶巨大PEComa。

随访6个月, 患者无明显症状, 未见复发迹象。本研究已获得患者知情同意。

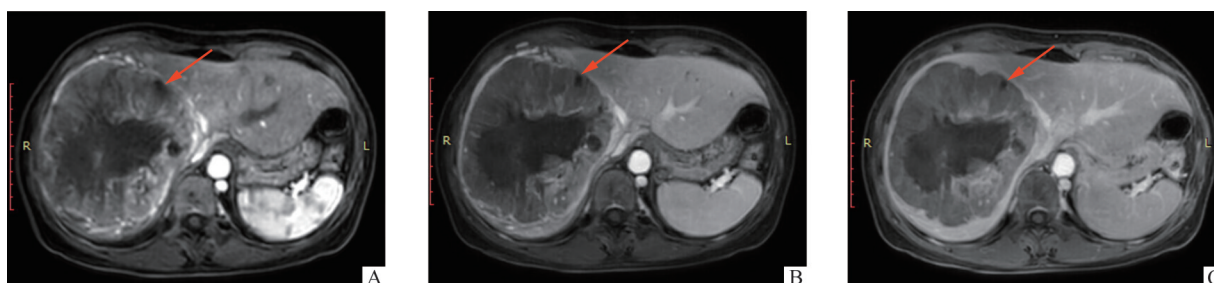


图1 磁共振成像

Figure 1 Magnetic resonance imaging

A: During the arterial phase, the mass in the right lobe of the liver exhibits marked heterogeneous enhancement, with a large number of fine and disordered tumor vascular shadows visible within. B: In the portal venous phase, there is relatively more distinct washout compared to the arterial phase (with reduced contrast enhancement compared to liver parenchyma), the central region shows heterogeneous enhancement and patchy areas without enhancement. C: In the delayed phase, it shows further clearance compared to before. Red arrow indicates the tumor.

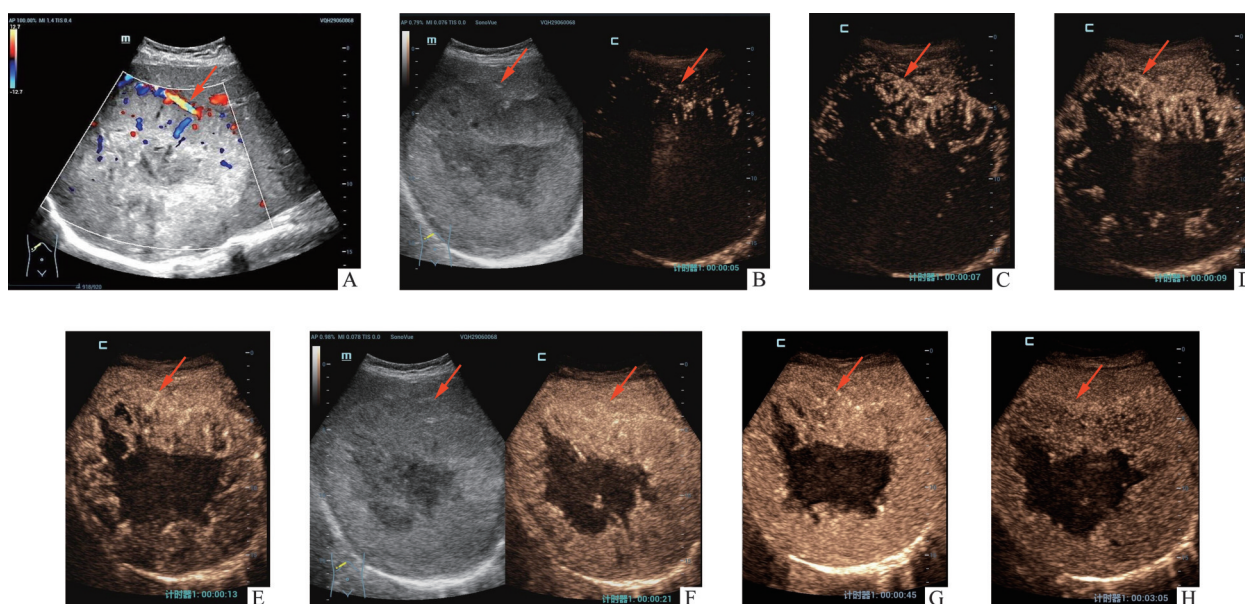


图2 超声影像

Figure 2 Ultrasonographic images

A: A large mass with heterogeneous echogenicity was detected in the right lobe of the liver. Multiple small patchy hyperechoic areas were observed within the mass. CDFI revealed relatively abundant blood flow signals. B-E: Ultrasound contrast imaging shows the mass demonstrating rapid, heterogeneous, centripetal hyperenhancement during the arterial phase [5 s (B), 7 s (C), 9 s (D), 13 s (E)], with cord-like vascular shadows visible and an irregular, non-enhancing area in the central portion. F: Exhibits heterogeneous hyperenhancement in the late arterial phase. G: Isointense enhancement in the portal venous phase. H: Demonstrates hypointense enhancement in the delayed phase. Red arrow indicates the tumor. CDFI: Color Doppler flow imaging.

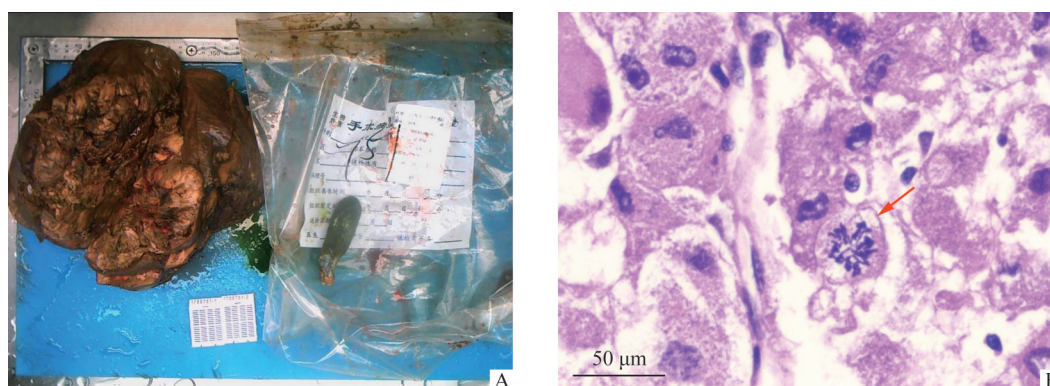


图3 肿瘤大体标本及HE染色结果

Figure 3 Gross specimen of the tumor and the results of HE staining examination

A: Gross specimen of the resected mass from the right lobe of the liver. B: Pathological mitotic figure (red arrow; HE staining).

2 讨论

肝 PEComa 好发于中青年女性, 临床症状不典型, 影像学表现无特异性^[2]。2023 年刘彬等^[3]对 19 篇文献的 452 例肝 PEComa 进行统计分析, 结果显示肿瘤最大径为 0.5~26.0 cm, 中位最大径在 4.0~9.3 cm, 其中 55.17% 的肿瘤最大径 < 5.0 cm; 术前影像学诊断正确率仅为 16.92%, 32.6% 误诊为肝细胞癌。PEComa 特征性表达黑色素细胞标志物(HMB45、MelanA)和平滑肌标志物(SMA)^[4]。目前 PEComa 的明确危险因素尚不清楚, 但部分病例与结节性硬化症(tuberous sclerosis complex, TSC)相关, 该遗传病导致的 *TSC1/TSC2* 基因突变及 mTOR 通路激活是其最重要的已知遗传易感因素。其治疗策略和预后高度个体化, 以根治性手术为主, 对于不可手术的患者, mTOR 抑制剂、介入栓塞等可作为治疗选择, 所有患者均需进行长期、密切的随访^[5,6]。

肝 PEComa 的影像学表现与肿块的成分有关。肝 PEComa 常为圆形或类圆形肿块, 边界清晰, CT 平扫呈低密度, 但也可因其内所含成分不同而表现为混杂密度^[7]; MRI 表现为 T_1 WI 稍低信号, T_2 WI 稍高信号肿块, 或者为混杂信号。CT 及 MRI 强化呈现多样性, 通常表现为动脉期明显增强, 门静脉期和延迟期廓清^[8]。根据部分文献报道, 超声表现亦缺乏特异性, 病灶既可呈低回声^[9], 也可呈高回声^[10], 较大者(平均直径 > 7 cm)可呈高回声或不均质混合回声^[4,11], CDFI 对血供显示敏感, 绝大多数病灶内部或周边可探及丰富的血流信号及粗大滋养血管。超声造影动脉期多呈高增强, 门静脉期及延迟期可快速减退呈低增强, 也可呈等或高增强^[9-11]。但由于报道的病例数较少, 还需要大样本验证。

回顾本病例, 分析肿块影像学特点: 在常规超

声中呈混合回声, 提示成分复杂; 超声提示肿块周边可见条状血流信号, 2 种影像学增强模式下均为动脉期高增强且有大量血管影, 说明肿块血供丰富; MRI 中病灶在 T_1 WI 呈等-稍低信号, T_2 WI 呈等-高信号, 提示病变细胞密度较高或富含蛋白质成分, 可能表现为水分子弥散受限。但要判断水分子是否为真性弥散受限, 需结合 DWI 和相应 ADC 值^[12], 这是判断细胞密度和恶性程度的重要工具。细胞越密集, 水分子弥散越受限, DWI 信号越高。本病例中 DWI 示中央可见片状等-稍高信号区, 相应 ADC 为等-低值, 说明是真性弥散受限, 这是恶性肿瘤的常见特征。本病例病灶的影像学表现与文献报道中的有相似之处: 本病例中巨块型病灶表现为混合性肿块, 且为富血供的特征, Ji 等^[4]和 Gao 等^[5]报道的部分病例也有类似表现; 肿块表现为“高-等-低”的超声造影增强模式与 Gao 等^[5]报道的部分病例一致; MRI 增强的表现与 Krawczyk 等^[8]报道的病例动脉期明显不均匀强化, 门静脉期、延迟期廓清吻合。郝传玺等^[13]及石建功等^[7]均指出肿块内含脂肪成分及增强后肿块内或边缘迂曲增粗的血管影是 PEComa 的典型征象, 本病例中病灶增强时见大量条索状血管影符合 PEComa 的典型表现。

然而, 这些特点并非 PEComa 特有, PEComa 常误诊为肝细胞癌、肝腺瘤、血管瘤^[3,14-15]。肝 PEComa 的影像学误诊源于其组织学异质性。这一情况深刻揭示了 PEComa 能够模拟多种肝富血供肿瘤的影像表现的本质。

由于巨块型肝癌直径 > 10 cm, 增强模式下表现为“快进快退”^[16], 所以本例罕见肝 PEComa 在外院 CT 误诊为巨块型肝癌伴液化坏死。但肝癌患者多有慢性肝病史^[17]及 AFP、异常凝血酶原等肿瘤标志物的升高, 本病例患者因腹部症状就诊, 既往病史无特

殊,肝相关肿瘤标志物阴性,可相鉴别。本院超声及MRI均误诊为纤维板层型肝细胞癌,从常规超声表现看,该病例的混合回声、特别是其内见片状高回声区且周边环状血流与纤维板层型肝癌常表现为混合回声、内部存在典型的中央瘢痕或钙化且病灶血流丰富的特征高度相似,初步影像层面难以快速区分;而在超声造影层面,本病例造影中肿块呈“高-等-低”的增强模式,内部可见条索状、放射状排列的血管增强影,超声造影及MRI增强均可见中央3期无增强区,这与纤维板层型肝细胞癌动脉期高增强,门静脉期及延迟期较前廓清,且中央瘢痕无强化的增强模式几乎一致,进一步模糊了二者的影像边界^[18];该病例的巨块型表现与纤维板层型肝细胞癌病灶体积较大的特点相契合。基于这些影像学表现和病史,该病易误诊为纤维板层型肝细胞癌。

该病例罕见之处主要体现在本例肿块最大径达15.2 cm,在平扫CT、MRI及常规超声上均表现为不均质肿块,提示肿瘤内部可能存在液化坏死、增生的纤维组织或钙化等,但本病例MRI未提示有脂肪成分,这也表明该例属于肝乏脂肪型PEComa这一亚型^[19]。此亚型的影像学表现不易与巨块型肝癌及纤维板层型肝细胞癌鉴别。常规超声可见中央高回声区,考虑为纤维板层型肝细胞癌的中央瘢痕,同时,此病例内部出现条索状、放射状血管增强影及中央3期无增强区,这种血管分布及中央的特征与纤维板层肝细胞癌的影像学表现更为相似,也是导致误诊的关键因素。

PEComa最终确诊还需要病理诊断,本病例呈现HMB45(+),MelanA(+),SMA(+),的关键免疫表型,符合PEComa的典型免疫表达。且S100、肝细胞标志物hepatocyte、glypican-3和CK8/18阴性,可基本排除黑色素瘤和典型肝细胞癌^[20]。CK19仅胆管阳性进一步排除胆管细胞癌。而CD34阳性证实了该肿瘤富血供的特点^[4]。Masson染色显示间质胶原纤维沉积,与PEComa间质特征相符。Ki-67(2%+)与肿瘤的生物行为有关,但不能排除恶性可能,Yamamoto等^[21]报道的结肠恶性PEComa的Ki-67为2.9%。结合核分裂象>1个/mm²及病理性核分裂象,根据PEComa良恶性标准^[3],考虑本病例为恶性PEComa。病理的形态学及免疫组织化学支持肝PEComa的诊断,这是本病例诊断最关键的依据。

在面对上腹部不适、无肝病病史的中青年女性检查发现肝占位病变时,应从多维度分析。影像学应多模态联合。CT评估畸形血管与增强模式;MRI多序列提供脂肪、扩散、强化模式、肝胆期信号等信息;超声及超声造影实时观察病灶的增强动力学。

面对增强后有杂乱血管影或未检测到脂肪的肝脏占位,均应主动将PEComa等罕见病纳入鉴别,临床表现、既往病史、实验室检查及影像学表现仅能提供线索,最终确诊必须依靠病理学及免疫组织化学检测HMB45、MelanA、SMA,并判断PEComa的良恶性,Ki-67为较低阳性表达也不能完全排除病灶恶性的可能,还需根据病灶大小、核分裂象等分析。

综上所述,肝PEComa罕见,其临床表现、实验室指标及影像学特征均缺乏特异性,容易误诊。当影像学(如超声、CT、MRI)发现肝脏占位性病变,若患者临床特征不典型、无肝病病史且血清AFP正常,尤其对于中青年女性,应高度怀疑PEComa。由于其具有潜在恶变的风险,应尽早寻求病理学确诊并选择合适的治疗方式进行干预,使患者获得良好的预后。

作者贡献声明: 晏斯婕、张立嫒 研究构思,论文撰写与修改;杨翼腴、谢家媛 病例数据采集;张波 研究构思,论文修改与指导,对文章的知识性内容作批评性审阅。所有作者阅读并同意最终的文本。

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