· 病例报告 ·

成人阴茎囊性淋巴管瘤 1 例

杨庆凯[1],卢 剑[1],陆 敏[2],洪 锴[1△]
(北京大学第三医院 1. 泌尿外科, 2. 病理科,北京 100191)

[摘 要] 淋巴管瘤是由淋巴系统发育异常所致的先天性脉管畸形,发病率约 1/4 000,多见于儿童,头颈部为好发部位,成人阴茎部位发病极为罕见,临床易误诊漏诊。本文报告 1 例成人阴茎囊性淋巴管瘤病例,患者 42 岁,因阴茎根部肿物 30 余年、近半年明显增大入院。查体于阴茎根部偏左侧触及约 7 cm×4 cm×5 cm 囊性肿物,透光试验阳性,双侧睾丸、附睾及精索未见异常。超声提示阴茎和阴囊交界区囊性无回声灶,边界清、形态不规则;磁共振成像显示阴茎根部多房囊性病变,范围约 5.6 cm×3.8 cm×6.3 cm,影像学均支持囊性淋巴管瘤诊断。患者在硬膜外麻醉下行囊性肿物完整切除术,术中见肿物呈多房囊性,内含灰褐色液体;术后病理示囊壁内衬内皮细胞,免疫组织化学染色提示 CD31 阳性,确诊为囊性淋巴管瘤。基因检测提示体细胞 *PIK3CA* 基因 E545(GAG > GCG)杂合突变,与淋巴管瘤主要致病机制相符。淋巴管瘤主要与体细胞 *PIK3CA* 基因突变相关,突变可激活下游信号通路,促进淋巴内皮细胞异常增殖,进而形成囊性病变。临床表现以缓慢增大的无痛性囊性肿物为主,确诊依赖影像学与病理检查,需与腹股沟斜疝、鞘膜积液、精索静脉曲张等疾病鉴别。治疗以手术完整切除为首选,可有效降低复发;无法手术者可选择硬化治疗或西罗莫司、阿培利司等靶向药物。本例患者术后恢复良好,随访 1 个月无复发。成人阴茎囊性淋巴管瘤虽罕见,但具备典型临床、影像及病理特征,结合基因检测可明确诊断;早期完整切除预后良好,临床应加强对罕见部位淋巴管瘤的识别与规范处理。

[关键词] 阴茎;囊性淋巴管瘤;淋巴管畸形
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Cystic lymphangioma of the penis in an adult: A case report

YANG Qingkai[1], LU Jian[1], LU Min[2], HONG Kai[1△]
(1. Department of Urology, 2. Department of Pathology, Peking University Third Hospital, Beijing 100191, China)

SUMMARY Lymphangioma is a congenital vascular malformation arising from aberrant lymphatic development, with a reported incidence of approximately 1 in 4 000. The condition predominates in the pediatric population, with the head and neck representing the most frequently affected anatomical regions. By contrast, scrotal lymphangioma in adults is exceedingly rare and remains highly susceptible to misdiagnosis or diagnostic omission in clinical settings. Here, we report a case of cystic lymphangioma of the penis in a 42-year-old male who presented with a mass at the penile root persisting for over 30 years, with marked enlargement over the preceding six months. Physical examination revealed a cystic mass measuring approximately 7 cm×4 cm×5 cm on the left aspect of the penile root, with a positive transillumination sign. Bilateral testes, epididymides, and spermatic cords were unremarkable. Ultrasonography demonstrated an irregular cystic anechoic lesion at the penoscrotal junction with well-defined margins. Magnetic resonance imaging (MRI) revealed a multilocular cystic lesion at the scrotal root measuring approximately 5.6 cm×3.8 cm×6.3 cm. Both imaging modalities were consistent with cystic lymphangioma. The patient underwent complete surgical resection under epidural anesthesia. Intraoperatively, the mass was confirmed to be multilocular and contained taupe-colored fluid. Histopathological analysis revealed endothelial cell lining of the cyst wall, and immunohistochemical staining was positive for CD31, establishing the definitive diagnosis of cystic lymphangioma. Genomic profiling identified a somatic heterozygous missense mutation in the *PIK3CA* gene at codon E545 (GAG > GCG), consistent with the established pathogenic mechanism underlying lymphangioma. Mechanistically, somatic *PIK3CA* mutations activate downstream signaling cascades, driving aberrant proliferation of lymphatic endothelial cells and culminating in cystic lesion formation. Clinically, lymphangioma typically manifests as a painless, slowly enlarging cystic mass. Definitive diagnosis relies on integrated imaging and histopathological evaluation, with differential diagnoses encompassing indirect inguinal hernia, hydrocele, and varicocele. Complete surgical resection remains the first-line therapeutic strategy and is associated with a substantially reduced recurrence rate. For patients deemed ineligible for surgery, sclerotherapy or molecularly targeted agents, including sirolimus and alpelisib, represent viable alternatives. The patient achieved an uneventful post-

△ Corresponding author's e-mail, kenhong99@hotmail.com
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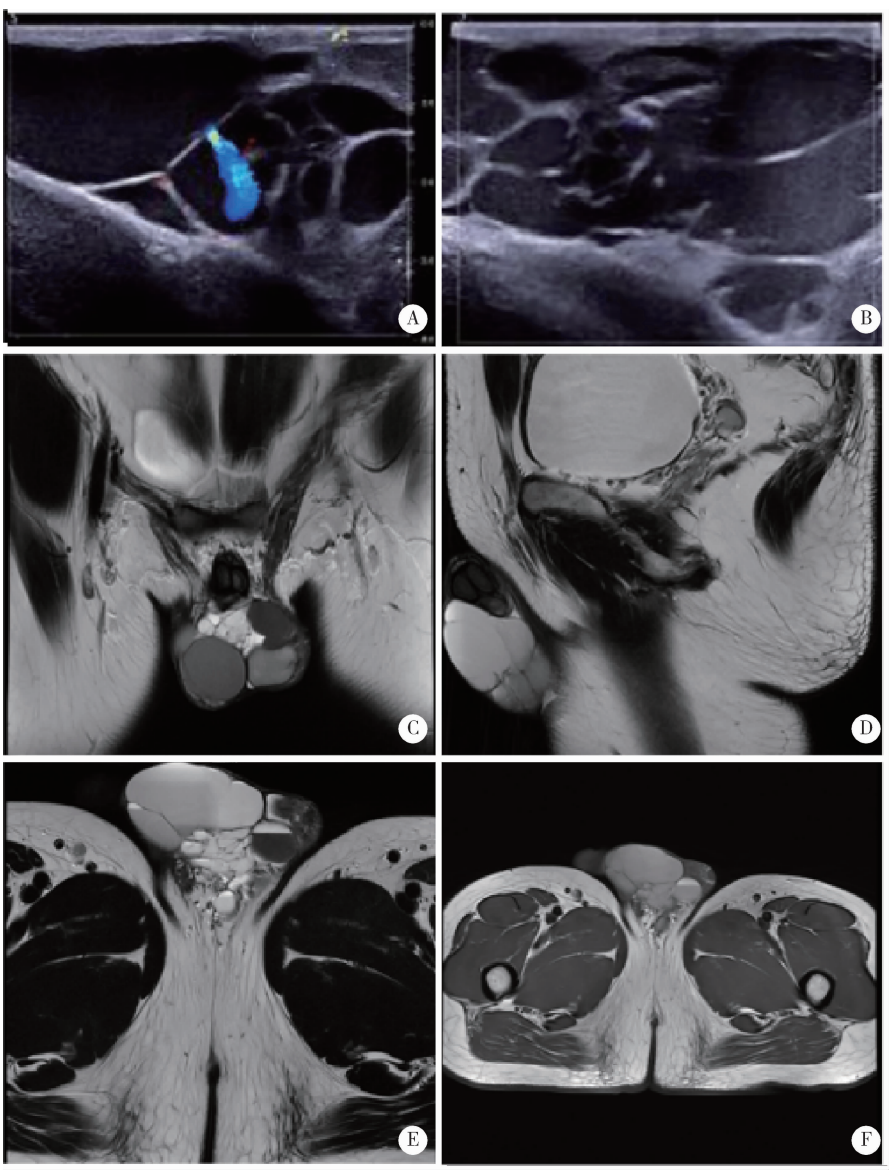
operative recovery with no evidence of recurrence at one-month follow-up. Although adult scrotal cystic lymphangioma is a rare clinical entity, it exhibits characteristic clinical, radiological, and pathological features that, in conjunction with molecular genetic analysis, enable accurate diagnosis. Early complete resection confers a favorable prognosis. Heightened clinical awareness and standardized management of lymphangioma at atypical anatomical sites are warranted.

KEY WORDS Penis; Cystic lymphangioma; Lymphatic malformation

1　病例资料

1.1　患者一般情况

患者男性,42 岁,因发现阴茎肿物 30 余年于 2023 年 12 月 7 日入院。患者 30 余年前发现阴茎根部肿物,大小约 2 cm × 2 cm,无其他不适,未诊治。近半年来肿物增长明显加快,查体阴茎根部偏左侧可及 1 个约 7 cm × 4 cm × 5 cm 的囊性肿物,挤压无明显缩小,透光试验阳性,双侧睾丸、附睾及精索触诊无明显异常,与肿物无明显关联。患者已育 2 子,均体健,暂无生育诉求。检验及检查:患者病程极其漫长,且肿物位置及性质均不提示睾丸来源,因此,未行睾丸肿瘤标志物检验,仅完善术前常规检验。超声及磁共振成像提示阴茎根部囊性肿物(图 1),考虑为淋巴管囊肿,拟行囊性肿物切除术。



A, B, ultrasound revealed an irregular cystic anechoic area at the penoscrotal junction with clear margins; C – F, MRI revealed a multilocular cystic lesion at the scrotal root with clear boundaries. MRI, magnetic resonance imaging.

图 1　囊性淋巴管瘤超声及磁共振成像

Figure 1　Ultrasonography and MRI of cystic lymphangioma

1.2 入院诊断

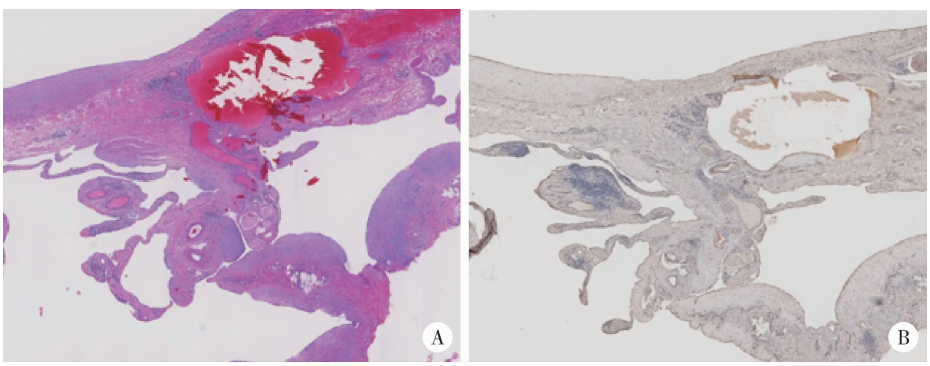
临床诊断:(1)阴茎肿物;(2)淋巴管囊肿可能。

1.3 治疗经过

患者于2023年12月11日在硬膜外麻醉下行阴茎囊性肿物切除术。术中可见肿物呈囊性不规则分叶状,向阴囊深面延伸,最大直径约7 cm,将肿物完整切除后于阴囊底留置引流管,彻底止血后逐层关闭阴囊切口并将完整组织送病理学检查,病变组织肉眼可见肿物切面呈多房囊性,壁厚0.1~0.2 cm,内外壁光滑,内含灰褐色液体,符合囊性淋巴管瘤特征。免疫组织化学染色(immunohistochemistry, IHC)提示CD31阳性(图2),进一步证实为囊性淋巴管瘤。术后取部分病变组织进行基因检测,结果提示*PIK3CA*基因E545(GAG>GCG)杂合突变(图3)。患者术后1个月门诊复诊,伤口恢复良好。

2 分析与讨论

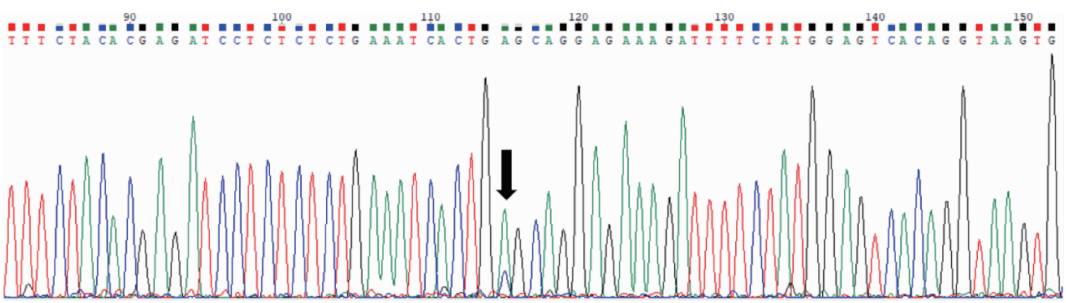
淋巴管瘤是由淋巴系统发育异常所致的先天性脉管畸形,发病率约1/4 000[1],多见于儿童,头颈部为好发部位,成人阴茎部位发病极为罕见,临床易误诊漏诊。在胚胎时期,淋巴内皮细胞自胚胎静脉中发育迁移而来,形成原始淋巴结构,脉管系统通过响应邻近间充质组织表达的血管内皮生长因子C,以离心出芽方式进一步扩张[2-3]。淋巴丛成熟过程中受到血管生成素2、转录因子FOXC2和GATA2等多种信号通路调控[4-7],上述分子或通路异常会导致淋巴发育成熟受阻,进而引起淋巴管结构功能异常[8]。根据国际脉管异常研究学会(International Society for the Study of Vascular Anomalies, ISSVA)分类,可以将淋巴管瘤分为普通型淋巴管瘤、泛发性淋巴管异常如卡波西样淋巴管瘤病(Kaposiform lymphangiomatosis, KLA)、戈谢病伴淋巴管瘤、通道型淋巴管瘤、"获得性"进行性淋巴管异常(淋巴管瘤)、原发性淋巴管水肿,其中原发性淋巴管水肿又可以分为若干亚型:如Nonne-Milroy综合征(致病基因*FLT4/VEGFR3*);原发性遗传型淋巴水肿(致病基因*VEGF-C/GJC2/Connexin 47*)-双行睫(致病基因*FOXC2*);少毛症-淋巴水肿-毛细血管扩张症(致病基因*SOX18*);原发性淋巴水肿伴骨髓发育不良(致病基因*GATA2*);Hennekam淋巴管扩张-淋巴水肿综合征(致病基因*CCBE1*);小头畸形伴或不伴脉络膜视网膜病变,淋巴水肿,智力障碍综合征(致病基因*KIF11*);淋巴水肿-后鼻孔闭锁(致病基因*PTPN14*)[9]。



A, cystic spaces lined by endothelial cells in dermis (HE ×100); B, CD31 immunostaining highlights the endothelial lining of the cysts (IHC ×100). HE, immunostaining; IHC, immunohistochemistry.

图2 囊性淋巴管瘤的组织学特征

Figure 2 Histological features of cystic lymphangioma.



Dual green and blue peaks at position 115 indicated a heterozygous *PIK3CA* exon 545 mutation(GAG>GCG) in somatic cells of the patient.

图3 囊性淋巴管瘤基因测序结果

Figure 3 Genetic sequencing results of cystic lymphangioma

2.1　病因及临床表现

目前认为体细胞突变为淋巴管瘤的主要原因。在 80% 的常见淋巴管瘤中,编码磷脂酰肌醇-4,5-二磷酸 3-激酶(Phosphatidylinositol-4,5-bisphosphate 3-kinase catalytic subunit α, PIK3CA)的 *PIK3CA* 基因突变是主要驱动因素[10-11],且大多数突变聚集在螺旋结构域 E542、E545 和激酶结构域 H1047 上,这些突变可能导致其下游蛋白激酶 B-哺乳动物雷帕霉素靶蛋白信号通路激活,进一步促进淋巴内皮细胞增殖[8]。本例患者存在体细胞 E545 (GAG > GCG) 杂合突变,与文献[8]一致。

淋巴管瘤可以发生在全身任何部位,主要是淋巴系统所在区域最为常见。大囊型淋巴管瘤由一个或多个体积≥2 cm^3 的囊腔构成,囊肿内充满嗜酸性粒细胞和富含蛋白质的液体,呈光滑半透明的肿块[12];微囊型则是一个或多个体积 <2 cm^3 的囊腔构成,主要影响皮肤和黏膜表面,表现为小的、半透明的出血性囊泡[13],混合型两者都有。淋巴管瘤的临床表现主要取决于病变大小、性质、所在位置及深度,有的表现为分布在皮肤上的小泡,有的表现为囊性肿物。本研究中患者表现为进行性增大的阴茎根部囊性肿物,透光试验阳性,大小与体位无关,符合囊性淋巴管瘤的特征。

2.2　诊断

影像学检查是诊断囊性淋巴管瘤的主要手段。超声是首选的辅助检查,有助于判断淋巴管瘤的大小、性质、部位、深度及与周围血管的关系,可以为进一步的治疗提供依据。MRI 可在超声的基础上进一步明确瘤体特征与周围组织的关系。也可进行诊断性穿刺,抽出液体若为淡黄色清亮液体提示为淋巴管瘤,结合病理若为血性液体,提示淋巴管瘤伴出血。本文患者超声提示阴茎阴囊交界囊性无回声区,边界清晰形态不规则,MRI 提示阴茎根部多房囊性病变,边界清晰,范围约 5.6 cm × 3.8 cm × 6.3 cm,影像学均符合囊性淋巴管瘤的特征。

2.3　鉴别诊断

患者病史极为漫长,且肿物生长缓慢,考虑为慢性良性疾病,因形态不规则呈囊状,透光试验阳性,且肿物位于阴囊阴茎交界处,需要与以下疾病相鉴别:(1)腹股沟斜疝:可见腹股沟可复性包块,包块可进入阴囊,平卧时可还纳,嵌顿时无法还纳,透光试验阴性;(2)鞘膜积液:透光试验阳性,触之有波动感,但是无囊性感,超声提示液性暗区,但是无分隔或囊性结构;(3)精索静脉曲张:平卧时包块缩小或消失,站立位时蚯蚓状团块明显,触之为静脉丛,多见于青年男性,超声显示精索内静脉扩张(内径≥2 mm),进行瓦尔萨尔瓦(Valsalva)动作时血流信号增强。

2.4　治疗

一般情况下,淋巴管瘤的生长较为缓慢,不危及生命,但是患者的病情也会根据病变生长部位的不同出现不同情况,倘若出现压迫、感染等症状也会危及生命,病变生长在面部、颈部也会影响容貌,因此,针对不同部位、不同类型的淋巴管瘤往往需要医务人员选择不同的治疗方式。

药物治疗:目前使用最广泛的药物为西罗莫司,该药物为哺乳动物雷帕霉素受体抑制剂,动物实验表明,西罗莫司可以有效抑制 *PIK3CA* 驱动的淋巴管瘤及复杂性淋巴管异常小鼠模型的淋巴管异常增殖[14-15],既往大量研究表明,西罗莫司可以有效缓解多种类型的淋巴管瘤[1]。外用型西罗莫司在毛细淋巴管瘤及浅表微囊性淋巴管瘤中具有较好疗效。曲美替尼可与西罗莫司发挥协同作用[8]。阿培利司是特异性磷脂酰肌醇 3-激酶抑制剂,被美国食品药品监督管理局批准用作 *PIK3CA* 突变介导的乳腺癌的治疗,研究表明该药物针对 CLOVES(congenital lipomatous overgrowth, vascular malformations, epidermal nevi, skeletal/scoliosis and spinal anomalies)综合征患者,可使病灶体积减小 37%[8]。

硬化治疗:适用于大囊型和混合型淋巴管瘤,创伤小,与手术相比操作简单,瘢痕小,同时复发率也较低,目前常用的硬化剂有博来霉素、平阳霉素、溶血性链球菌制剂 OK-432、无水乙醇等[1]。一篇针对儿童囊性淋巴管瘤硬化治疗的荟萃分析指出,临床上常用的硬化剂为溶血性链球菌制剂 OK-432,其次还有博来霉素、玉米蛋白乙醇溶液(alcoholic solution of zein, ASZ)及多西环素。经皮硬化治疗的患者中,48% 反应率极佳。针对不同病变类型的统计中,大囊型患者治疗效果(66%)优于微囊性患者(23%),上述常用药物针对大囊型病变的成功率相近,均为 80% 以上,而针对微囊型 ASZ 成功率为 77%。同时,上述药物均具有一定的副作用,如发热、局部炎症、疼痛等[16]。另一篇仅聚焦于溶血性链球菌制剂 OK-432 硬化治疗的研究提示,70% 患者经过硬化治疗后有良好或极好的效果,既往接受过治疗、病变位置较差(例如纵隔),以及注射次数多,是预后不良的主要因素[17]。

手术治疗:通过手术切除淋巴管囊肿需要考虑病变部位周围重要血管神经的浸润情况,手术指征如下:病灶较小可完全切除,且对外观和功能影响

小;有症状的微囊性淋巴管瘤;硬化治疗无法消除,且仍有症状的大囊型及混合型淋巴管瘤;有危及生命的并发症;对外观影响较大[1]。本文中患者的淋巴管囊肿位于阴茎阴囊交界处,且瘤体相对较大,严重影响外观及功能,因此,选择手术切除。

淋巴管瘤的病因主要为体细胞 *PIK3CA* 突变,其下游通路可促进淋巴内皮细胞增殖,临床表现因囊型、部位而异。诊断以影像学检查为主,需与腹股沟斜疝等疾病鉴别。治疗需个体化,包括药物治疗、硬化治疗及手术切除,其中手术切除适用于瘤体较大、影响外观功能等情况。

利益冲突 所有作者均声明不存在利益冲突。

作者贡献声明 杨庆凯、卢剑:提出研究思路,设计研究方案,收集、分析、整理数据,撰写论文;陆敏:收集、分析、整理数据;洪锴:提出研究思路,总体把关及审定论文。所有作者均参与论文修改,并对最终文稿进行审读和确认。

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