

· 临床研究与实践 ·

儿童 Rapunzel 综合征诊治分析



全文二维码

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【摘要】目的 探讨儿童 Rapunzel 综合征的临床特点及诊治对策。**方法** 回顾性分析 2019 年 1 月至 2021 年 5 月丽水市中心医院收治的 5 例 Rapunzel 综合征患儿临床表现、治疗过程和转归资料。以检索词“Rapunzel/长发公主、毛发结石”检索万方、维普、中国知网数据库,以“Rapunzel syndrome,trichobezoar”检索 PubMed、Embase 数据库相关文献,剔除文献综述、重复报道病例、成人病例及无法获得详细资料的病例后,对相关文献进行复习和总结。**结果** 本院 5 例均为女性,4 例长发,1 例短发且有秃发。5 例均以呕吐、腹痛、腹部包块等胃肠道梗阻症状就诊。5 例均于术前经胃镜和(或)CT 等检查证实存在消化道异物,结合有食毛或拔毛史,经手术确诊为 Rapunzel 综合征。1 例伴电解质紊乱、多发小肠穿孔;1 例伴营养不良、体重下降;1 例并发肠缺血坏死,2 例无明显并发症及合并症。5 例均通过大切口剖腹手术取出毛发团块,平均手术时间 2 h,平均住院时间 10 d,均恢复良好顺利出院。出院后于本院精神卫生科随诊,随访时间 3 个月至 3 年,平均随访时间 1.7 年,随访期间无一例复发。共收集 Rapunzel 综合征相关文献 28 篇(52 例),男女比例为 3:49,平均年龄 11.73 岁,有拔毛史 35 例(35/52,67.3%);均行手术治疗,以开腹手术为主(39/52,75%),以胃肠切开取异物术为主要术式;51 例预后良好,1 例术前合并胃穿孔患儿术后因重度营养不良、持续消化道出血死亡。**结论** 儿童 Rapunzel 综合征往往引起营养不良等非特异性表现,可导致肠梗阻、隐匿性肠穿孔等并发症。CT 和胃镜可早期诊断,手术是该病的主要治疗手段,长期精神科随诊及心理治疗可预防复发。

【关键词】 肠梗阻; 胃肠结石; 胃肠吻合术; 肠穿孔; 外科手术; 儿童

基金项目:浙江省医药卫生科技计划项目(2021ZH070、2022ZH019)

DOI:10.3760/cma.j.cn101785-202106053-018

Rapunzel syndrome in children:a report of 5 cases with a review of the literature

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【Abstract】Objective Rapunzel syndrome is rare in children and delayed diagnosis and treatment may lead to various complications. The purpose of this paper was to explore the clinical characteristics and diagnostic and treatment countermeasures of Rapunzel syndrome in children. **Methods** From January 2019 to May 2021, retrospective review was conducted for clinical manifestations, treatment courses and outcomes of five hospitalized cases of children with Rapunzel syndrome. The search terms of “Rapunzel/Princess Rapunzel,trichobezoar” were employed for searching the databases of Wanfang, Wipu, CNKI and “Rapunzel syndrome,trichobezoar”. The relevant literatures of PubMed and Embase databases were summarized after excluding literature reviews, repeated cases, adult cases and cases for which detailed information was non-available. **Results** All five children were female, four with long hair and one with short hair and alopecia. Clinical manifestations included gastrointestinal obstruction such as vomiting, abdominal pain and abdominal masses, etc. All five cases were confirmed to have gastrointestinal foreign bodies by gastroscopy and/or CT pre-operation and the diagnosis of Rapunzel’s syndrome was finally confirmed by a history of hair eating or plucking. Complications were electrolyte disturbance and multiple small bowel perforations ($n=1$), malnutrition and weight loss ($n=1$) and intestinal ischemic necrosis ($n=1$). The remainders had no significant complications or comorbidities. Hair masses were removed by small incisional dissection. The average operative duration was 2h and the average hospital stay 10 days. All of them recovered well and were discharged successfully. After discharge, consultations with mental health de-

partment continued. None of them had recurrence during the mean follow-up period of 1.7 years (3 months to 3 years). Through a literature retrieval, a total of 28 literature reports in Chinese and English were collected, with 52 cases with a male to female ratio of 3 : 49 and 94.2% of females. The mean age was 11.73 years and there were 28 cases with a history of hair extraction (28/52, 53.8%). Fifty-two children were operated and laparotomy (39/52, 75%) predominated with gastroenterostomy for foreign body removal. The outcome was excellent in 51 of them, except for one child with preoperative gastric perforation who died postoperatively due to severe malnutrition and persistent gastrointestinal bleeding. **Conclusion** Rapunzel syndrome in children often causes non-specific manifestations such as malnutrition and can lead to complications such as intestinal obstruction and occult bowel perforation, and delayed treatment may be life-threatening. CT and gastroscopy may provide an early diagnosis and surgery remains a major treatment. Long-term psychiatric follow-ups and psychotherapy prevents a recurrence.

【Key words】 Intestinal Obstruction; Bezoars; Gastroenterostomy; Intestinal Perforation; Surgical Procedures, Operative; Child

Fund program: Zhejiang Provincial Medical and Health Science and Technology Program (2021ZH070, 2022ZH019)

DOI:10.3760/cma.j.cn101785-202106053-018

Rapunzel 综合征又称长发公主综合征 (Rapunzel syndrome, RS), 此综合征根据格林兄弟同名童话中的主角长发公主 Rapunzel 命名。1968 年 Vaughan 等^[1]首次报道了以 RS 命名的这一罕见而严重的疾病症状, 患者长期吞食毛发, 以致大量毛发在胃肠道聚集形成较坚固的异物团块, 进而在胃肠蠕动作用下形成一鼠尾样异物, 自幽门延伸到空肠、回肠甚至结肠。国内外关于 RS 的文献多为个案报道。本文报道丽水市中心医院 5 例患儿的诊治经过, 并进行相关文献复习, 为此类疾病的临床诊疗提供参考。

病例资料

一、本院患儿临床资料

丽水市中心医院 2019 年 1 月至 2021 年 5 月共收治 5 例 RS 导致急性胃肠梗阻患儿, 年龄 5~12 岁, 均为女性。4 例长发, 1 例短发且有秃发现象。病程(从出现症状到就诊时间)4~7 d, 均以呕吐、腹痛、腹部包块等胃肠道梗阻症状就诊, 1 例存在电解质紊乱, 1 例营养不良、体重下降, 另外 3 例全身情况尚可。5 例均有食毛或拔毛史, 术前均经胃镜和(或)CT 检查证实存在消化道异物。

5 例均于入院 24 h 内于全身麻醉下行急诊手术, 1 例尝试腹腔镜手术失败而中转剖腹探查术, 4 例直接行剖腹探查术。取右中腹横行小切口入腹, 探查至肠管扩张狭窄移行处寻及异物, 切开胃或肠管取出异物, 予肠管减压, 探查全胃肠道有无隐匿性胃肠穿孔、肠缺血坏死、消化道畸形。1 例术中切开小肠后取出鼠尾状毛发粪石团块, 小肠肠壁淤血严重, 浆肌层多

处坏死灶, 行“小肠切开异物取出术 + 肠部分切除肠吻合术”。3 例鼠尾状毛发团块位于胃内, 尾端延伸入十二指肠(图 1), 其中 2 例术前胃镜检查发现毛发团块, 曾尝试胃镜下取出失败, 直接行胃切开取异物术; 1 例位于胃内且鼠尾端延伸入十二指肠、小肠内, 距十二指悬韧带 20~50 cm 处空肠段发现 8 处隐匿性穿孔, 各穿孔灶之间仅相隔 3.0~5.0 cm, 为避免遗漏穿孔灶而行小肠部分切除术, 最终行“胃切开异物取出术 + 小肠部分切除肠吻合术”(图 2)。平均手术时间 2 h, 平均住院时间 10 d, 5 例均经手术治疗痊愈出院。出院后精神卫生科随诊, 随访时间 3 个月至 3 年, 平均随访 1.7 年, 随访期间无一例复发。5 例患儿临床表现、手术情况及治疗结果见表 1。

二、文献复习

共搜索到 RS 中英文文献报道 28 篇, 病例数 52 例(表 2)。



注 A: CT 平扫横断位, 图像显示胃腔、十二肠腔内巨大混杂含气密度影(白色五角星), 团块内强化不明显, 团块尾部见“鼠尾征”延伸入小肠(白色箭头); B: 冠状位 CT 平扫, 见十二肠腔内巨大混杂含气密度影, 团块内强化不明显, 团块尾部可见“鼠尾征”延伸入小肠(白色箭头)

图 1 1 例 Rapunzel 综合征患儿的术前 CT 图像
Fig. 1 Preoperative CT image of a child with Rapunzel syndrome

表 1 本院 5 例 Rapunzel 综合征患儿的临床表现、手术情况及治疗结果

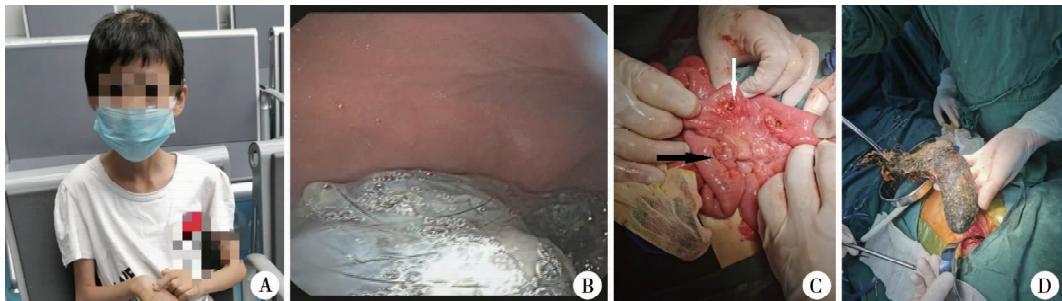
Table 1 Clinical presentations, surgical approaches and outcomes of 5 children with Rapunzel syndrome

病例编号	性别	年龄(岁)	头发类型	有无嗜毛症	病程(d)	症状	手术方式	并发症	住院时间(d)	预后
1	女	12	长发	有	4	急性肠梗阻表现	小肠切开取异物术 + 小肠部分切除术	肠缺血、坏死	11	存活
2	女	8	短发	有	5	腹胀、腹部包块	胃切开取异物术	营养不良、体重下降	9	存活
3	女	5	长发	有	6	腹痛	胃切开取异物术 + 小肠部分切除术	电解质紊乱、多发小肠穿孔	12	存活
4	女	10	长发	有	7	饱腹感、呕吐	胃切开取异物术	无	9	存活
5	女	11	长发	有	6	急性腹痛、呕吐	胃切开取异物术	无	9	存活

表 2 2005—2021 年文献报道的儿童 Rapunzel 综合征病例资料

Table 2 Rapunzel syndrome in children reported in the literature from 2005 to 2021

发表年份	例数	性别比例 (男:女)	年龄 (岁)	病史	拔毛史	处理	并发症/合并症	预后
2005— 2009 ^[2-3]	2	0:2	10~17	慢性腹痛、食欲下降	1 例	胃肠切开取异物术	无	均存活
2010— 2019 ^[4-25]	30	0:30	5~17	急慢性腹痛、呕吐、腹部包块、肠梗阻、胃痛、饱腹感等	19 例	20 例胃肠切开取异物术, 1 例腹腔镜胃切开术, 9 例内镜下异物取出术	阑尾炎、肠套叠、贫血、十二指肠梗阻、急性胰腺炎、肠穿孔、术后发生真菌性腹膜炎等	均存活
2020— 2021 ^[26-29]	20	3:17	8~17	急性腹痛、呕吐、全身水肿、晕厥发作等	15 例	胃肠切开取异物术	蛋白丢失性肠病、胃穿孔、胰腺炎、肠套叠等	死亡 1 例, 余存活



注 A: 患儿外观营养不良、秃发; B: 胃内可见毛发团块; C: 术中小肠图, 可见空肠多发隐匿性穿孔, 拨开浆肌层表面粘连可发现穿孔灶(白色箭头), 肠壁浆肌层表面假性瘢痕粘连(黑色箭头), 拨开粘连可见隐匿性穿孔灶; D: 术中于胃肠内取出毛发团块

图 2 1 例 Rapunzel 综合征患儿外貌、胃镜图像及术中所见

Fig. 2 External appearance, gastroscopic images and intraoperative views of a child with Rapunzel syndrome

讨 论

RS 是一种罕见病, 是指由于吞入毛发在胃内形成毛团, 在胃肠蠕动作用下形成一鼠尾样异物, 从幽门延伸到空肠、回肠甚至结肠的毛发结石^[9]。由于腔内结石体积较大, 可发生梗阻或穿孔的急性表现。毛发结石好发于拔毛癖人群, 又称冲动控制障碍 (trichotillomania, TTM), 被认为是一种反复拔除毛发的强迫行为, 常导致大片头皮光秃。TTM 患病率为 0.5%~2.0%, 以青少年时期(10~13 岁)发病居多,

年轻女性常见^[30]。超过 20% 的 TTM 患者拔出头发后会选择将其吃掉, 从而导致 RS。相关研究表明, TTM 不仅是心理疾病, 还可能与基因、环境因素有关^[31-32]。本组病例中我们根据胃肠道梗阻表现, 结合患儿长发或脱发等情况, 合理怀疑患儿存在 TTM。

Mirza 等^[28]认为 RS 的临床表现取决于毛发结石在胃肠道的大小、位置和停留时间, 认为 RS 可表现为三种模式: ①急性表现伴并发症; ②隐匿性表现, 病灶稳定; ③在儿童中偶然发现病变被误诊为其他慢性疾病, 如腹部结核/恶性肿瘤。

RS 的诊断需结合拔毛癖和(或)食毛癖病史，并通过影像学检查得到证实。腹部 X 线可提示肠梗阻表现，B 超可提示肠腔扩张、肠腔内团块。CT 扫描是一种较好的检查方法，不仅可以识别异源结石，还可以确定其扩展范围。典型 RS 的 CT 图像可见毛发团块一侧呈长短不一的鼠尾状延伸，称为“鼠尾征”，胃内毛发团大多明显可见尾部呈鼠尾状伸入十二指肠甚至小肠，孤立小肠内肿块可能“鼠尾征”不十分明显^[2]。胃石(尤其含气胃石)以及孤立胃石排入小肠，需与粪块鉴别，部分胃石边缘能显示出较完整的环形高密度影，此环在粪块中较罕见；此外表现为含气蜂窝状影的较大粪块常位于大肠内，若小肠内较大的含气蜂窝状影团块影需考虑胃石下排可能^[33]。部分胃石内部可形成“年轮征”，可能与毛发与食糜等物长期混合沉积、硬化形成巨大结石有关。临幊上若出现急性肠梗阻症状，CT 显示胃内和(或)小肠内出现“鼠尾征”“年轮征”、含气蜂窝状影团块影表现，需合理怀疑存在 RS，及时询问是否有嗜毛史或拔毛史。

胃镜检查对胃黏膜毛发结石具有诊断价值，但其治疗价值有限。胃内毛发结石往往体积较大，手术及麻醉时间长，单纯经胃镜难以完整取出结石。Nita 等^[26]报道内镜下取出毛发结石的成功率为 30.7%，同时报道了 1 例因内镜下采用烧灼法切除毛发而意外发生小肠和胃多发穿孔病例，考虑与术中烧灼合成毛发产生潜在危险气体以及毛发的物理磨损有关。黄一敏等^[25]报道了胃镜辅助经腹壁小切口胃切开术治疗胃内毛发结石，丁媛媛等^[13]报道内镜下分次取毛发异物的成功案例，表明内镜治疗仅对胃内毛发结石可行，但内镜下取毛发结石易遗漏肠管隐匿性穿孔。另外控制 RS 复发也是治疗的关键环节，有专家建议术后 6 个月、12 个月和 24 个月对其他患有 TTM 症的患者进行内镜随访^[12]。

外科剖腹切开取石仍是治疗 RS 引起肠梗阻的主要手段。本研究 1 例患儿手术中发现多发小肠隐匿性穿孔，我们分析该胃肠道隐匿穿孔的形成是由于肠蠕动使毛发食物混合团块反复向前移动导致反复切割肠管，该处肠管与周围组织粘连堵塞穿孔灶，穿孔灶表面浆肌层假性粘连，因而并未表现出急性肠穿孔表现。若术中发现肠管水肿胀气，即便术前无肠穿孔征象，亦需仔细探查十二指肠、小肠等处，以免遗漏隐蔽穿孔。

术后长期精神卫生科随访是预防 RS 复发的重要手段。降低 TTM 复发率的治疗包括药物和非药

物治疗相结合，并要求多学科团队以及家人和朋友参与。选择性 5-羟色胺再摄取抑制剂和氯米帕明被大部分专家认为是 TTM 的一线药物。其他用于治疗 TTM 的药物有拉莫三嗪、奥氮平、n-乙酰半胱氨酸、肌醇和纳曲酮。习惯逆转训练在减轻 TTM 症状方面有明显效果且有强力证据支持^[34]。

综上所述，RS 罕见，临幊上出现急性肠梗阻表现结合 CT 典型“鼠尾征”、“年轮征”等表现，同时伴有脱发、营养不良等情况，应该高度怀疑存在 RS，可进一步完善上消化道内镜检查，早期诊断和及时手术干预有助于改善预后，精神病学评估和处理是防止复发的关键。

利益冲突 所有作者声明不存在利益冲突

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(收稿日期: 2021-06-19)

本文引用格式:林小燕,张恒,卓金伟,等.儿童 Rapunzel 综合征诊治分析[J].临床小儿外科杂志,2023,22(1):92–96. DOI:10.3760/cma.j.cn101785-202106053-018.

Citing this article as: Lin XY, Zhang H, Zhuo JW, et al. Rapunzel syndrome in children: a report of 5 cases with a review of the literature[J]. J Clin Ped Sur, 2023, 22(1): 92–96. DOI: 10.3760/cma.j.cn101785-202106053-018.