

# 手术年龄对先天性巨结肠术后功能影响的相关研究进展

张韵涵, 郭振华\*

国家儿童健康与疾病临床医学研究中心, 重庆医科大学附属儿童医院普外新生儿外科, 重庆

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## 摘要

先天性巨结肠(Hirschsprung's Disease, HSCR)的手术年龄尚存在争议, 手术年龄对于患儿术后肛门功能及生活质量是否存在影响, 已引起医学界广泛关注。当前对于HSCR的治疗主要是手术治疗, 在Swenson, Duhamel, Rehbein和Soave等经典术式基础上, 衍生出单纯经肛门拖出术(Transanal Endorectal Pull-Through, TERPT)及腹腔镜手术(Laparoscopic-Assisted Pull-Through, LAPT), 较传统术式大大减少创伤, 缩短肠功能恢复时间外, 也使得手术治疗趋向于低龄化。当前争论的焦点在于患儿的手术年龄及术式的选择上。虽然目前并无充足的证据证明存在使患儿获得最优术后功能的最佳手术年龄及最佳手术方式, 但是TERPT及LAPT均被证实是在新生儿期也可安全进行的手术操作, 并且对于全身情况较好的HSCR患儿, 尽早手术治疗, 术后可以获得较为满意的肛门功能及生活质量, 但为进一步明确手术干预时机, 仍需更多的循证医学证据。

## 关键词

先天性巨结肠, 手术, 术后功能

# Advancements in the Effect of Surgical Age on Postoperative Function in Hirschsprung's Disease

Yunhan Zhang, Zhenhua Guo\*

National Clinical Research Center for Child Health and Disorders, Department of General & Neonatal Surgery, Children's Hospital of Chongqing Medical University, Chongqing

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\*通讯作者。

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## Abstract

The optimal timing for surgical intervention in Hirschsprung's Disease (HSCR) remains a topic of debate within the medical community. The potential impact of varying ages of surgery on postoperative anal function and quality of life in pediatric patients has garnered significant interest among researchers and practitioners. The current primary treatment for HSCR is surgical intervention, which includes traditional methods such as Swenson, Duhamel, Rehbein and Soave, as well as advanced techniques like Transanal Endorectal Pull-Through (TERPT) and Laparoscopic-Assisted Pull-Through (LAPT). Compared to traditional surgery, these two surgical methods significantly reduce trauma and shorten the recovery time of intestinal function, while also shifting the trend of surgical treatment towards a younger demographic. The current debate is centered on the optimal age of children for surgery and the most effective surgical methods. Currently, there is insufficient evidence to determine the ideal surgical age and method that would result in the best postoperative function in children. Nevertheless, studies have demonstrated that both TERPT and LAPT are safe surgical procedures during the neonatal period. For pediatric patients with favorable conditions, early surgical intervention can lead to favorable outcomes in terms of anal function and quality of life post-operation. But additional evidence-based research is required to enhance the understanding of the optimal timing for surgical intervention.

## Keywords

Hirschsprung's Disease, Surgery, Postoperative Function

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## 1. 引言

先天性巨结肠(Hirschsprung's Disease, HSCR)是一种肠神经系统发育障碍疾病,即肠管的肌间神经丛和黏膜下神经丛中缺乏神经节细胞,导致功能性肠梗阻。发病率为 1/5000 [1]。当前对于确诊的 HSCR 患儿,均应采取手术治疗,并且根据病变肠管范围及患儿全身情况,采取不同的手术方案[2]。由此导致的手术年龄低龄化,以及手术对患儿生长发育及肛门功能带来的影响,已引起医学界广泛重视[3] [4]。新生儿肠道免疫功能尚不成熟,手术操作可能损伤内括约肌等精细结构,因此对于手术年龄的选择,患儿术后功能的评估,以及特殊类型 HSCR 的术式选择及功能等,仍然存在较多争议。本文就以上问题,结合国内外相关研究进行综述如下。

## 2. 先天性巨结肠手术治疗方式的选择

对于确诊的 HSCR 患儿,均需进行手术治疗[2] [5]。手术的原则是切除无神经节细胞的肠管,防止进一步发生的功能性肠梗阻[6]。目前手术方案可分为一期手术和分期手术,其选择应根据患儿自身情况(如基础状况,病变肠管范围等)和家庭情况进行综合评估。但明确适应症至今并未有统一标准[2]。

### 2.1. 一期手术

自 20 世纪 40 年代起,小儿外科医师对术式进行不断的改良与尝试,但 Swenson, Duhamel 和 Soave 等术式仍被视为治疗 HSCR 的经典术式。在 20 世纪 90 年代早期,Georgeson [7]发表了第 1 例经肛门拖

出术治疗先天性巨结肠的微创手术方式, 随后的研究证明其在住院时间, 喂养时间, 疼痛程度均低于传统开放手术, 得到了广泛应用。腹腔镜辅助手术随后也被报道用于 Duhamel 和 Swenson 手术[8] [9]。截止目前, 并没有前瞻性研究明确证明一种术式优于其他手术方式, 不同术式均在世界各地广泛开展, 并且在长期功能随访研究中均获得相似的结果[10]。

近年来, 随着微创技术的发展, 单纯经肛门手术(Transanal Endorectal Pull-Through, TERPT)和腹腔镜辅助手术(Laparoscopic-Assisted Pull-Through, LAPT)由于避免了开腹路径导致的相关并发症, 手术安全性大幅度提高, 从而得到了广泛的开展。虽然这两种手术方式多被定义为两种手术方式, 但在操作方面有相似之处, 两者均涉及会阴区解剖, 都需要切除无神经节细胞的直肠及齿状线以上区域的吻合[11]。主要区别在对于直肠的解剖路径不同。目前尚未有直接比较腹腔镜手术和经肛门手术的疗效的长期随访研究。在长期随访中, 相对传统开放手术, 接受 TERPT 手术治疗的患儿虽然对于肠梗阻, 肠粘连等并发症有着更低的发生率, 但是术中过度牵拉肛门, 更易损伤内外括约肌, 由此导致术后失禁, 污粪的发生率更高[12]。由于操作的局限性, TERPT 术式更适用于短段型及部分常见型 HSCR。目前已有多项研究证明, 腹腔镜辅助下先天性巨结肠一期根治手术可以降低手术创伤, 缩短患儿禁食时间, 并且相对开腹手术, 术后便秘和小肠结肠炎(Hirschsprung's Disease Associated Enterocolitis, HAEC)的发生率较低[13] [14]。但相较经肛门手术, 腹腔镜手术对于小儿外科医生的操作提出了更高的要求。

此外, 对于特殊类型的 HSCR, 尤其是新生儿期的患儿一期手术治疗时机尚存在争议。有研究表明, 新生儿期虽然因肠管黏膜水肿程度较轻, 切开操作相对简易, 但术后住院时间长, 肛门操作损伤内外括约肌风向较高[15]; 同时新生儿期 HSCR 准确诊断的难度较高, 术前钡灌肠假阴性发生率较高, 直肠黏膜活检敏感度较年长儿降低[16]。因此部分小儿外科医师倾向于患儿年龄和体重增长后再行手术治疗, 这也尽可能避免因新生儿未成熟的肠道免疫系统导致 HAEC 发作可能。然而目前随着多项研究证明无论是 LAPT 还是 TERPT 均可在新生儿期安全开展, 且术后可获得基本满意的疗效[17] [18]。越来越多的小儿外科医师选择对于非全结肠型, 基础状况良好的 HSCR 患儿行一期根治手术治疗。

## 2.2. 分期手术

近年来, 大多数小儿外科医师主张 HSCR 的治疗首选一期根治手术并尽量避免造瘘手术。但是如果患儿术前即合并肠穿孔, 严重营养不良, 严重 HAEC 等并发症时, 应选择分期手术治疗[2]。截止目前, 已有多项回顾性研究验证一期手术具有麻醉次数少, 恢复快, 避免长期肠造瘘相关并发症等优势[18], 但对于比较一期手术和分期手术治疗效果, 仍需多中心的随机对照研究去进一步验证。

此外, 对于全结肠型巨结肠(Total Colonic Aganglionosis, TCA)的首选术式还未达成共识, 分期手术治疗是目前应用最为广泛的治疗方式。虽然小儿外科医师们对 TCA 的治疗做出多次尝试, 但无论是回肠直肠吻合(如 Soave 术式), 还是将正常肠管与无神经节肠管侧端吻合(如 Duhamel 术), 并未发现哪种术式表现出突出的优势[19] [20]。通过比较手术时间, 术后恢复时间以及 HAEC 等并发症发病率等方面, 有学者认为接受分期手术的患儿可减少 HAEC 的反复发作, 并可改善术后污粪及肛周溃疡的症状[21] [22]。也有回顾性研究表明: 对于全结肠型患儿行一期手术安全有效[23]。因此, 当前对于 TCA 的手术方式的选择并没有统一的意见, 需要在综合评估患儿病情的基础上, 根据术者的经验和技能决定。

## 3. 先天性巨结肠术后功能相关评估

能否正常排便是衡量 HSCR 患儿术后生活质量的重要指标。上世纪 90 年代初, 国内外学者为开展术后排便功能相关研究, 制定了不同的排便功能评估量表, 如 Rintala 量表(唯一可在健康儿童与畸形儿童间进行排便功能比较的量表) [24], 在肛门直肠疾病中广为应用的 Kelly 评分, 改良 Wingspread 评分量表

[25]及 Heikkinen 量表[26]。这些量表都涉及了排便频率, 控便能力, 污粪等症状的评估, 但并未有研究证明在评估排便功能效率方面表现优异的特定评分量表。

多项研究表明 HSCR 术后生活质量显著下降的患儿, 多存在严重排便失禁, 污粪等问题[12] [27] [28]。但 Neuvonen 等人的研究表明在采用不同生活质量评分的条件下, 接受手术的 HSCR 患儿术后远期生活质量评分和未接受胃肠手术的对照组评分具有相似可比性。因此在对 HSCR 患儿进行生活质量评估时, 除了关注排便功能, 也需重视其心理, 社会功能的评估。但是当前不同研究对于生活质量的定义缺乏统一标准, 且人口多样性导致社会心理评估的不同结果, 目前尚未有评估 HSCR 患儿生活质量标准化方案

根据专家共识[2]推荐, HSCR 患儿术后应在 1 个月, 6 个月, 1 年, 2 年常规随访, 定期评估排便及控便能力, 如有术后并发症(如失禁, 污粪等), 可以通过饮食结构调整, 心理疏导, 排便习惯训练等干预治疗帮助重建排便功能, 提高患儿生活质量[29], 大多数术后排便异常均可通过上述保守治疗措施得到改善, 但根据文献统计仍有约 1%~10%的患儿需再次手术治疗改善排便功能[30]。

#### 4. 不同手术年龄对 HSCR 术后功能的影响

随着对该疾病的认识的提高, 80%~90%的患儿在新生儿期即可明确诊断[31]。与此同时随着麻醉和微创手术的发展, 多项研究均证明了在新生儿期开展 TERPT 手术的安全性[32] [33]。但是, 新生儿期 HSCR 患儿术后的肠道功能一直是备受关注的焦点问题, 多项研究表明新生儿可能由于肠道免疫功能不完善, 肛门内外括约肌发育不全等因素, 所以在新生儿期接受一期手术治疗的患儿术后易存在便失禁, 长期污粪, HAEC 反复发作等问题, 严重影响患儿的生活质量[34] [35]。同时也有学者认为患儿尽早手术, 可避免因粪便长期积存导致肠管扩张肥厚, 缩短了手术时间及术后恢复时间, 术后可获得较为满意的肠功能[36]。

一项基于 780 名短段型先天性巨结肠患儿术后功能的荟萃分析显示: 在较小的婴儿中实施 TERPT 手术与术后并发症(如吻合口狭窄, 污粪等)的发生率增加相关( $P < 0.01$ ) [37]。此外, 有回顾性研究表明手术年龄在 2 月龄以下是患儿术后出现排便问题的危险因素[38]。这一方面可能是新生儿和小婴儿的肛门括约肌, 盆腔神经更容易因牵拉操作损伤; 另一方面是在低月龄儿中肠管神经节细胞的病理诊断更具挑战性, 针对这一点, 尚未有文献报道明确的有效解决方案。尽管上述研究表明患儿尽早手术术后倾向于表现较差的肠功能, 但也存在推迟手术可能导致因粪便积存致肠管肥厚, 手术难度增加, 术后并发症发生率增加的可能性。

目前, 手术年龄对 HSCR 患儿术后远期功能的影响尚无明确定论。部分学者对接受手术治疗的患儿随访至成人, 约半数的患者有满意的功能结果[26]; 在一项针对 HSCR 患儿术后功能的前瞻性研究中, Conway 等人评估患儿术后远期功能结果, 证明患儿肠功能随生长发育而改善, 尽管约半数的患儿长期评估结果差强人意[39]。此外, 有回顾性研究表明, 在新生儿期接受手术的患儿, 约 70%在随访 7 年以上获得较为满意的肠功能[40]。但上述研究缺乏对全结肠型患儿的评估数据, 并且不同年龄接受手术的患儿随访时间缺乏统一的标准, 因此对于明确手术年龄对于远期功能的影响, 不仅需连续规律的全面功能评估结果, 还需在评估肠道功能基础上, 综合评估心理健康及生活质量[41]。

#### 5. 总结与展望

综上所述, 目前 HSCR 的最佳手术年龄并没有明确的定论, 并且对于患儿术后功能及生活质量缺乏长期的规律随访评估, 这对患儿未来生长发育及社会功能尤为重要。目前临床对于明确诊断的 HSCR 患儿大多采取一期经肛门手术或腹腔镜辅助手术治疗。但是对于特殊类型的 HSCR 患儿(如新生儿期 HSCR, TCA, 合并严重 HAEC 等并发症), 需综合评估患儿病情, 根据医师的经验及能力选择制定合适的个体化



治疗方案。TERPT 及 LAPT 均是安全有效的外科治疗方案, 应在临床积极推广和应用。术后患儿均需进行长期规律的功能评估, 对于存在失禁, 长期污粪的患儿, 应及时进行肛门功能康复锻炼, 可有效改善生活质量。但是基于术后功能明确先天性巨结肠最佳手术年龄, 仍需多中心的前瞻性研究来验证。

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