



Hirschsprung's disease and Down syndrome: From the reappraisal of risk factors to the impact of surgery.

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Abstract

INTRODUCTION:

The association of Hirschsprung disease (HSCR) and Down Syndrome (DS) is not uncommon (HSCR+DS). This paper aims at reporting the results of a 24-year series focusing on surgical approach, complications and long term outcome.

MATERIALS AND METHODS:

The notes of all patients admitted with a diagnosis of HSCR+DS have been retrospectively reviewed. Surgical details, intraoperative complications, long term issues and functional outcome have been recorded. The results have been compared to those of patients without DS and were assessed based on surgical approach.

RESULTS:

A total of 23 HSCR+DS out of a series of 385 HSCR (6%) have been included. Preoperative enterocolitis (HAEC) was reported by 32%. Associated anomalies were detected in more than half of the patients. In particular, Congenital Heart Defects (CHDs) were reported by 57%. Postoperative complications (mostly symptomatic anal sphincter achalasia) were experienced by 55%. Constipation was experienced by 30%; severe continence issues, by 53%. One patient suffering from severe CHDs died. With regard to complications, only symptomatic anal achalasia requiring intrasphincteric BoTox injection was significantly more frequent in HSCR+DS (30% vs 10%, $p = 0.0071$). Similarly, continence proved to be significantly worse in HSCR+DS.

DISCUSSION:

With the exception of symptomatic anal achalasia, HSCR+DS patients proved not to have a higher likelihood of complications compared to HSCR alone. On the other hand, functional results in the long term are worse. As a consequence, long term follow up and personalized rehabilitation programs are warranted for this delicate subset of HSCR patients.

LEVEL OF EVIDENCE:

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KEYWORDS:

Continence; Down; Enterocolitis; Hirschsprung; Minimally invasive surgery