European Reference Network
for rare or low prevalence complex diseases

Network Inherited and Congenital Anomalies (ERNICA)

# ERNICA GUIDELINES FOR THE MANAGEMENT OF RECTOSIGMOID HIRSCHSPRUNG'S DISEASE (HSCR)

## A SUMMARY OF THE RECOMMENDATIONS FOR HEALTHCARE PROFESSIONALS

# INTRODUCTION

The ERNICA guidelines for the management of rectosigmoid Hirschsprung's disease[1] were published online in the *Orphanet Journal of Rare Diseases* in 2020 (in English) and present ERNICA recommendations for the diagnosis and management of rectosigmoid Hirschsprung's Disease until adulthood. Recommendations are based on a comprehensive literature review and expert consensus from multidisciplinary healthcare professionals and patient organisation representatives. AGREE II and GRADE approaches to guideline development were adopted and levels of evidence and agreement are noted in the guideline. Most recommendations made are based on expert opinion, as there was limited high-quality clinical evidence available to consider.

This document seeks to provide a summary of the recommendations made in the ERNICA guideline. This summary is targeted towards healthcare professionals and designed to complement the fuller guideline document. This summary is available in all European languages.

[1] Kyrklund K, Sloots C, de Blaauw I, Bjørnland K, Rolle U, Cavalieri D, Francalanci P, Fusaro F, Lemli A, Schwarzer N, Fascetti-Leon F, Thapar N, Johansen LS, Berrebi D, Hugot JP, Crétolle C, Brooks AS, Hofstra RM, Wester T, Pakarinen MP. ERNICA guidelines for the management of rectosigmoid Hirschsprung's disease. *Orphanet Journal of Rare Diseases.* 2020; 15(1), 164.

# KEY RECOMMENDATIONS

## DIAGNOSIS

- ✓ The diagnosis of Hirschsprung's Disease (HSCR) should be based on representative rectal histology, and should be confirmed before pull-through surgery.
- ✓ Rectal biopsy is indicated if the clinical history and physical signs are suggestive of HSCR.
- ✓ Biopsies should be evaluated by an experienced consultant histopathologist, seeking external consultation if necessary.

## TREATMENT OF HIRSCHSPRUNG'S DISEASE

- ✓ Patients with HSCR should undergo pull-through surgery in centres with at least two pediatric colorectal surgeons and pathological, radiological and anesthetic expertise, including pediatric and neonatal intensive care and specialized nursing 24/7.
- ✓ Centres performing pull-through surgery for HSCR should have the capabilities to manage the entire care pathway.
- ✓ Centres that operate on HSCR patients should demonstrate active involvement in quality control and improvement.
- ✓ Parents and patients should receive information about the availability of patient support organisations as soon as possible.

## SURGICAL MANAGEMENT

- ✓ Patients should receive saline rectal irrigations 1–3 times per day to decompress the bowel until the definitive pull-through operation.
- ✓ A stoma is indicated if rectal irrigations do not sufficiently decompress the bowel, or there are complications such as enterocolitis unresponsive to non-operative treatment, or bowel perforation.
- ✓ When possible, a pre-operative contrast enema is recommended to guide on the likely level of aganglionosis.
- ✓ At pull-through surgery, one dose of broad-spectrum intravenous antibiotics should be given preoperatively.
- ✓ Centres should perform the type of pull-through in which they have the most experience, including management of post-operative complications and follow-up.

- ✓ The pull-through operation should be performed when the patient is stable and growing well, and the bowel has been sufficiently decompressed.
- ✓ The anal canal should be preserved during pull-through surgery.
- ✓ The colon should be transected at least 5 to 10 cm proximal to the first normal biopsy minimize the risk of a transition zone pull-through.

## POST-OPERATIVE CARE AND FOLLOW UP

- ✓ Patients should receive specialist pediatric and nursing care during the early post-operative period, and anaesthetic consultation should be available on request.
- ✓ Enteral feeding can be started gradually when the patient has recovered from anesthesia and is clinically stable.
- ✓ The urinary catheter should be removed as soon as normal micturition is expected after pelvic floor surgery.
- ✓ The coloanal anastomosis should be calibrated around 2–3 weeks after pull-through surgery.
- ✓ Children with HSCR should receive regular follow-up to adulthood within the context of an interdisciplinary care team, led by a pediatric surgeon.
- ✓ Access to care and specialist consultation should be available.
- ✓ The introduction to adult medical disciplines should be prepared well before transition.

## HIRSCHSPRUNG'S DISEASE ASSOCIATED ENTEROCOLITIS (HAEC)

- ✓ HAEC should be clinically suspected in the presence of diarrhea with explosive, foul-smelling stool, and/or > 4 points from the Pastor et al (2009) HAEC score items[2]
- ✓ In suspected HAEC, there should be a low threshold for hospital admission.
- ✓ Following admission to hospital, patients with HAEC should be treated with intravenous fluid resuscitation, intravenous broad-spectrum antibiotics and rectal washouts.
- ✓ Intersphincteric botulinum toxin injections are recommended for patients with recurrent or persistent symptoms of outlet obstruction and/or HAEC.
- ✓ Prophylactic antibiotics may be considered for patients with frequently recurring or persistent HAEC.

[2] Pastor AC, Osman F, Teitelbaum DH, et al. Development of a standardized definition for Hirschsprung-associated enterocolitis: a Delphi analysis. *J Pediatr Surg*. 2009; 44:251–6.

- ✓ At present, there is insufficient evidence to support recommending the routine use of probiotics for the prevention of HAEC.

- ✓ In children with recurrent HAEC, consultation with a gastroenterologist and endoscopy should be considered.

## MANAGEMENT OF FECAL INCONTINENCE AND OBSTRUCTIVE DEFECATION

- ✓ Children with normal intellectual development who are not continent of stool by 4 years of age should be considered for further evaluation.
- ✓ The management of fecal incontinence should aim for age-appropriate continence in children with normal intellectual development.
- ✓ Patients with an intact anal canal and appropriate pull-through but fecal incontinence should receive medical management as the first-line treatment.
- ✓ Patients with fecal incontinence and damaged anal canal should receive bowel management.
- ✓ Children with persistent obstructive symptoms following pull-through surgery should undergo further evaluation and treatment

## GENETIC ASSESSMENT

- ✓ In non-syndromic HSCR, genetic testing of RET should be considered.
- ✓ In syndromic HSCR, patients should be offered referral for genetic consultation and screening for the specific gene associated with the syndromic phenotype.


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Co-funded by
the Health Programme
of the European Union