PATIENT JOURNEY

Rectosigmoid Hirschsprung's Disease (HSCR)

AUGUST 2023

European Reference Network | ERNICA

INTRODUCTION

What is HSCR?

Hirschsprung's Disease (HSCR) is a rare medical condition, characterised by a lack of nerve cells in the bowel, starting from the anus upwards. The lack of nerve cells (ganglion cells) makes bowel movement (peristalsis) difficult and leads to serious chronic constipation.

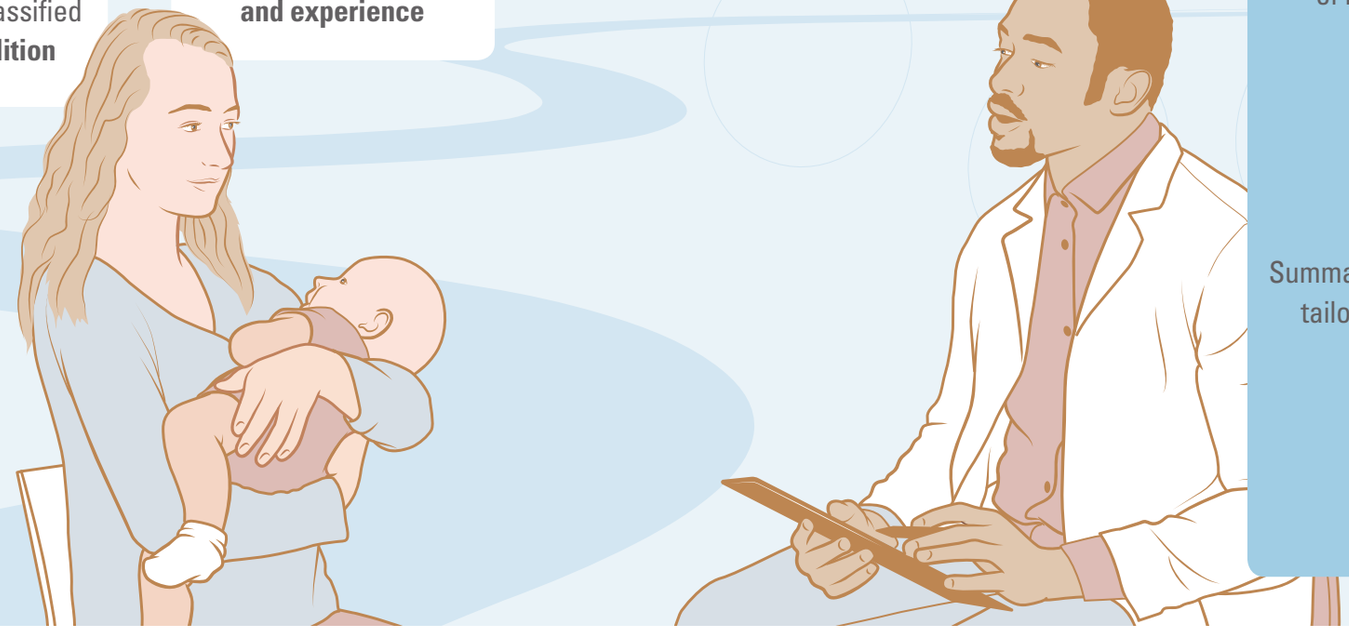
HSCR is a **congenital disorder**, meaning a child is born with it

The condition affects only one in **5000** babies. It is therefore classified as a **rare condition**

Diagnosis and surgery of HSCR are **challenging** and require **expertise and experience**

Treatment should **only** be performed by **specialists**

Every case is different



European Reference Network for rare Inherited Congenital Anomalies (ERNICA)

ERNICA expert centres for HSCR

Guideline for the management of Rectosigmoid HSCR (2020)

Summary for clinicians and a version tailored to patients and families

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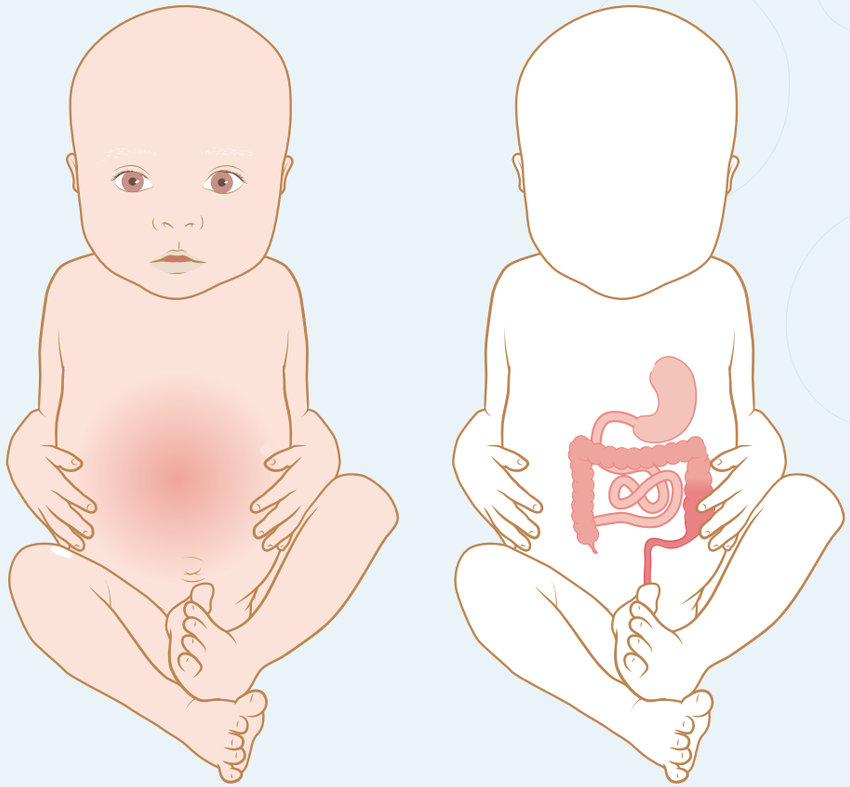
First symptoms

At birth the baby looks healthy. The symptoms indicating HSCR can arise soon after birth or gradually, for example, when the baby starts eating solid food. The symptoms vary from case to case.

The following may be an indication of HSCR:

- Delayed or no production of meconium (first faeces)
- Refusal to drink milk
- Failure to thrive, low weight
- Vomiting and dehydration
- Bloated tummy
- Constipation
- Anal stimulation (for example, with a rectal thermometer) leads to an explosive evacuation of faeces and gas
- Enterocolitis (inflammation of the intestine)



Necessary action

- If unwell, the baby should be brought for medical evaluation
- Colonic irrigation (washing out the bowel, enema) helps to relieve the intestine and to produce faeces
- If colonic irrigation does not work sufficiently, surgery may be needed to create a temporary stoma (opening in the body for faeces to pass through)

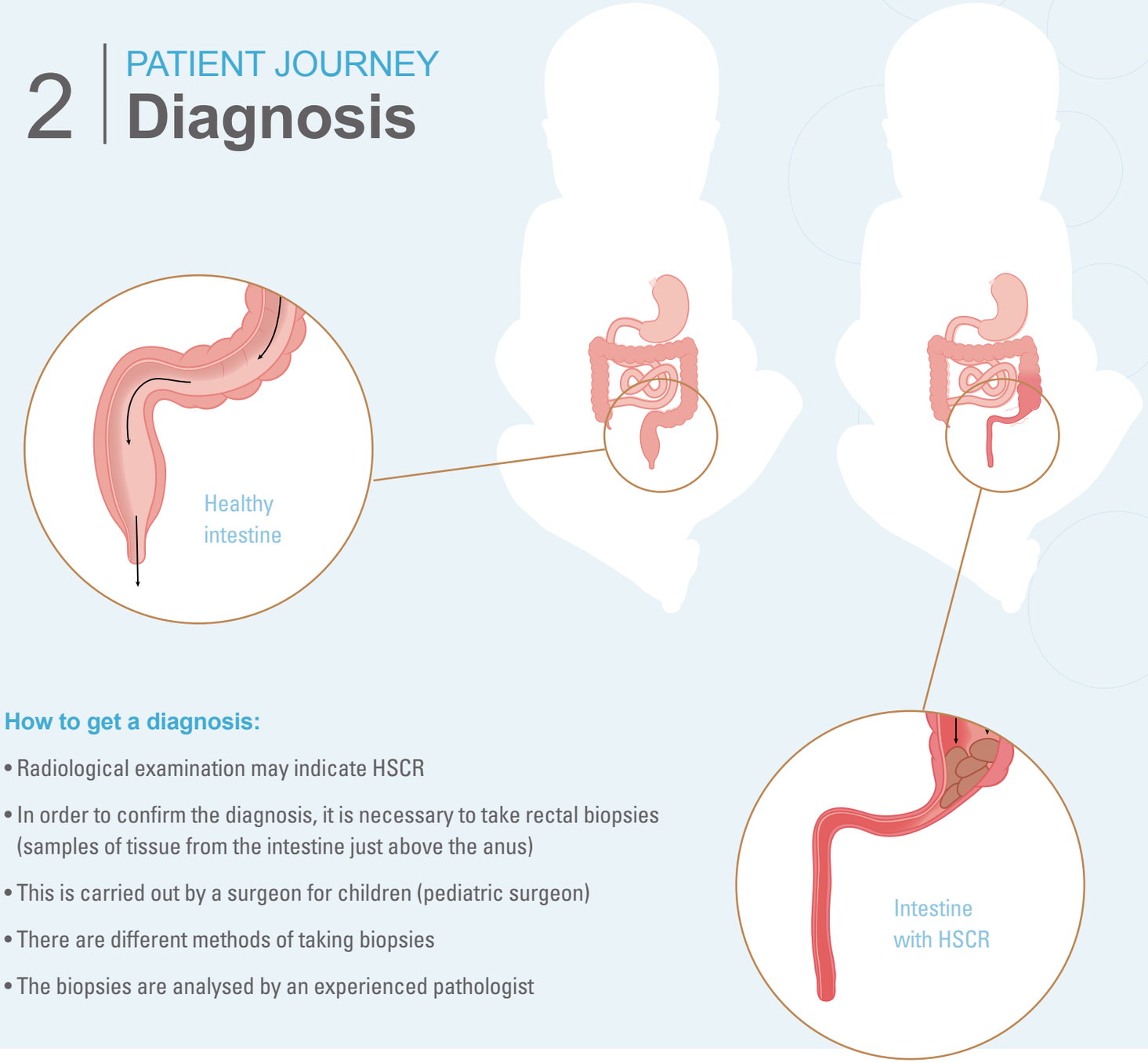
Ideally

- Parents are well informed by the doctors about the suspected disorder and next steps
- They are informed that HSCR is a rare condition, which needs specialist knowledge and treatment
- They are referred to a patient organisation for more information and advice
- Psychological support should be available as well as information on possible financial and social support

Animation for parents and families: What is Hirschsprung's Disease?

Patient organisations involved in ERNICA:

2 PATIENT JOURNEY Diagnosis



How to get a diagnosis:

- Radiological examination may indicate HSCR
- In order to confirm the diagnosis, it is necessary to take rectal biopsies (samples of tissue from the intestine just above the anus)
- This is carried out by a surgeon for children (pediatric surgeon)
- There are different methods of taking biopsies
- The biopsies are analysed by an experienced pathologist

Necessary action

- Confirmation of the HSCR diagnosis through biopsies
- Parents should learn how to look after their child's particular needs until corrective surgery (of affected part of the intestine) is performed
- Medication and/or enemas, or looking after the temporary stoma might be required
- Breastfeeding is recommended if possible

Ideally

- Early diagnosis of HSCR
- The measures mentioned under "Necessary action" reduce the risk of constipation /enterocolitis (inflammation of the intestine) and enable the baby to thrive

If required, the following should be available for parents:

- Support from a specialised nurse/stoma therapist
- Psychological, financial and social support

Animation for parents and families: How is Hirschsprung's Disease diagnosed?

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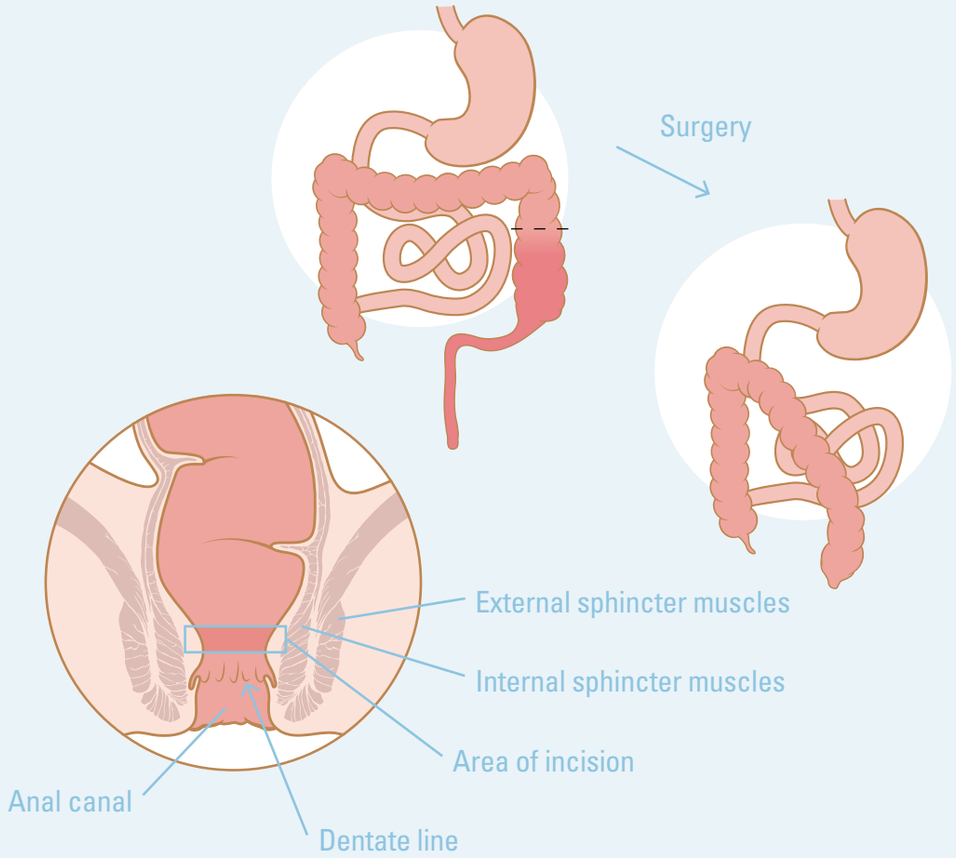
Surgery

Surgical removal of the affected part of the intestine is usually performed 2-3 months after diagnosis, when the child is stable and growing well and there are no signs of enterocolitis (inflammation of the intestine).



Necessary action

- Correct removal (resection) of the part of the intestine which is lacking nerve cells
- Preservation of the anal canal in order to maintain continence (ability to control bowel movements)

In preparation for the surgery:
A test called a "contrast enema" should be done. During this test, a special liquid is passed into a baby's bowel through the anus. Then an X-ray is performed. The liquid can be seen on the X-ray and may show which part of the bowel is affected

Further biopsies may be necessary if:
- It is unclear, how long the part is where nerve cells are missing (length of aganglionosis)
- A long segment without nerve cells is suspected

During the operation:
- Biopsies (samples of tissue) are taken to confirm the presence of nerve cells
- If the child has a temporary stoma, this might be removed at the same time, or alternatively in a separate operation

Ideally

Parents are well informed about:
- How the operation is done and what the potential risks are
- Problems that might arise after surgery, and how these can be managed
- Follow-up care at the clinic and contact details for emergencies
- After the operation, parents get a copy of the operation and pathology report

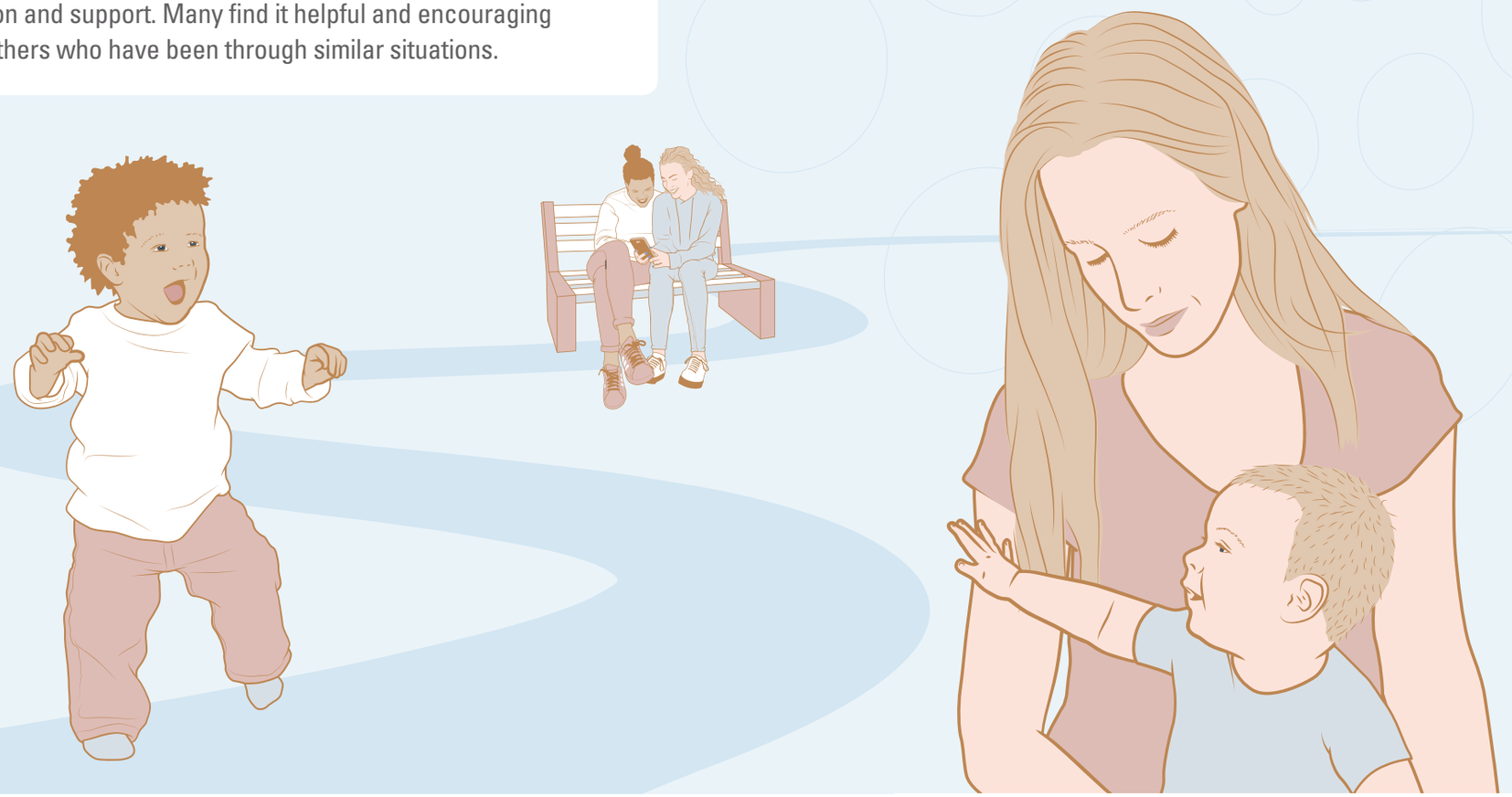
Animation for parents and families: Surgical treatment for Hirschsprung's Disease

Follow-up care

Some children are free of symptoms after corrective surgery. Some continue to have symptoms, which need to be managed. As HSCR is a rare and complex condition, short and long-term follow-up care should be available to all patients.

You're not alone!

Patient organisations provide parents and patients with useful information and support. Many find it helpful and encouraging to meet others who have been through similar situations.



Patient organisations involved in ERNICA:

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4 After the operation and first years

Although not always the case, difficulties may arise after the operation. Individual solutions need to be found.

Possible short and/or long-term difficulties:

- Wound-healing difficulties
- Diarrhoea (frequent liquid faeces)
- Irritated, sore skin around the anus
- Difficulties becoming completely continent (clean/able to control bowel movements)
- Ongoing constipation and bloated tummy, possible causes can be:
  -Scarring and narrowing of the anal canal
  -Obstruction in the intestine
  -Bowel emptying difficulties
  -Tight sphincter muscles (in the anal canal)
- Enterocolitis (inflammation of the intestine)
- Problems with eating and growing

Necessary action

- Sufficient emptying of the colon is essential
- Regular medical check-ups are needed to identify and treat potential problems early
- Specialised nursing helps to deal with perineal skin problems (around the anus), as well as establishing good toilet routines
- Further improvement might be through:
  - Medication
  - Bowel management (colonic irrigation/enemas)
  - Nutrition
  - Multidisciplinary, psychological and physiotherapeutic treatment
  - In rare cases, further surgical procedures are needed

Animation for parents and families: Hirschsprung's associated enterocolitis in children

ERNICA guideline for the management of Rectosigmoid HSCR (2020)

Summary for clinicians and a version tailored to patients and families

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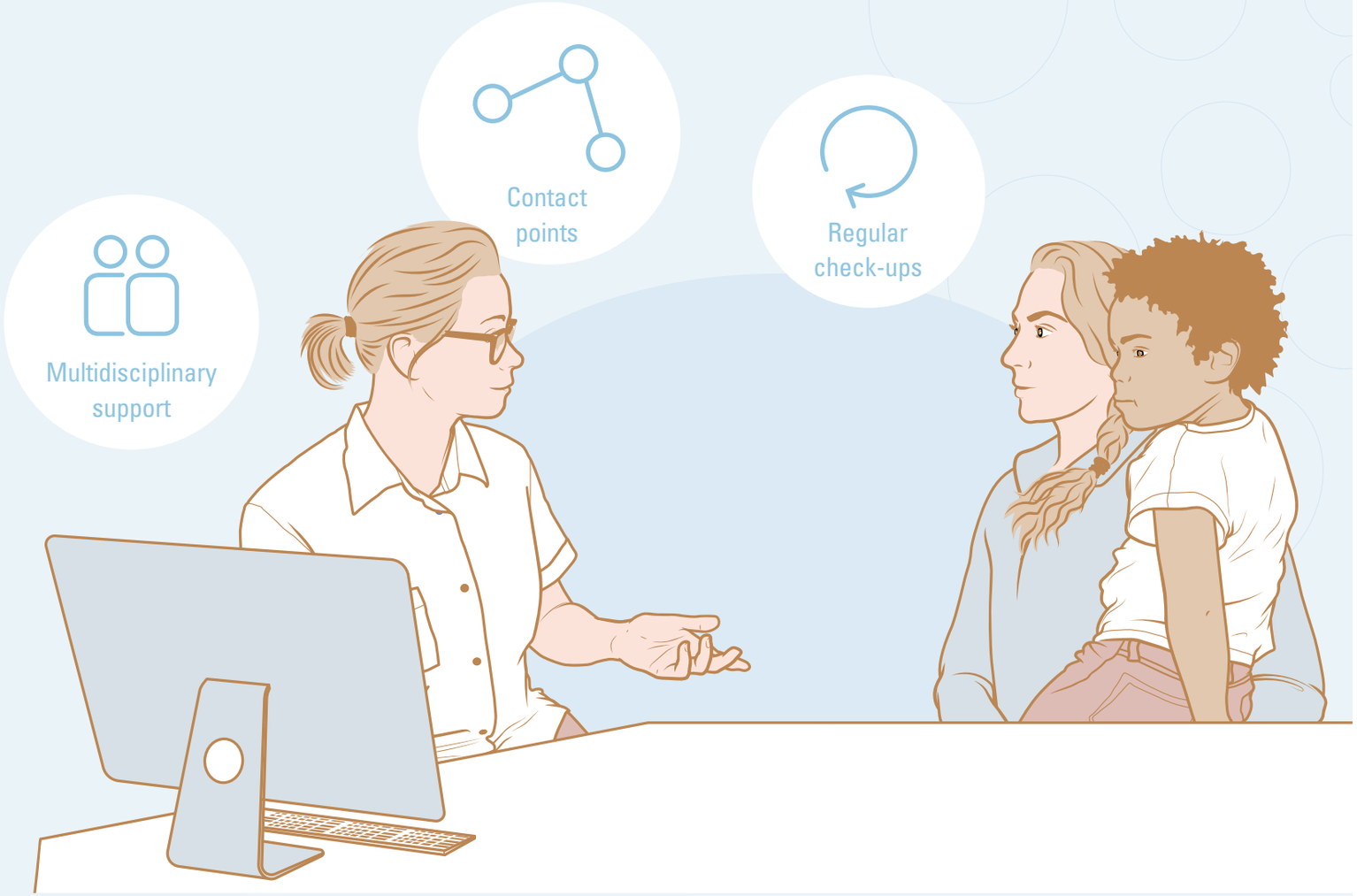
4 After the operation and first years



Ideally

- Early identification of potential problems
- Supervision and regular check-ups, ideally by the surgeon who operated on the child
- Parents know how to identify enterocolitis (inflammation of the bowel)
- They know who to contact and how to manage an emergency situation
- Although the child might take longer to be clean without nappies/diapers, they manage sufficiently to pass faeces and to be continent (clean/ able to control bowel movements)
- Psychological, social and financial support should be available for the family
- After surgery, genetic counselling can be offered to the family

Animation for parents and families: Hirschsprung's associated enterocolitis in children

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Primary School age

The following questions may help to assess your child's individual situation and whether further medical improvement is needed:

- What is the child's quality of life?
- Can they take part in typical everyday activities of children their age?
- Which measures could potentially improve the continence situation (ability to control bowel movements)?
- What support might be needed at school?

Necessary action

- Regular medical check-ups with the surgeon in order to identify potential difficulties
- According to the child's needs, treatment by a multidisciplinary team led by the surgeon (See step 4: After the operation - Necessary action)

Ideally

- Structured, regular check-ups, maybe less often than at the earlier stage
- The child can join in everyday activities of peers
- Parents know what possibilities there are for supporting their child at school

Children often benefit from getting to know other children with Hirschsprung's disease, for example through a patient organisation

Patient organisations involved in ERNICA

6 | PATIENT JOURNEY Puberty

The following questions may help to assess your teenager's individual situation and whether further support is needed:

- How can the teenager become independent/autonomous and manage their own body?
- How can the teenager deal with the condition in their social lives?
- Does the HSCR condition affect sexuality/ partnerships?

Necessary action

- Regular medical check-ups in order to identify potential problems
- If needed, interdisciplinary therapeutic support, including professional sexual advice

Important psychological factors are:

- Self-confidence
- Development of management skills for residual symptoms
- Social functioning

Transition process into adulthood and adult care

- Many small steps towards independence are needed from an early age on into adulthood
- The medical transition process should start at the latest from around 13 - 14 years of age
- The process includes discussing sexuality issues

Ideally

- The teenager can lead an independent life with as few restrictions as possible and know what is good for them
- They find their own way of dealing with their condition in their private and public lives and know where to get help, if needed
- Role of the parents: Parents support their teenager in finding their own way into adulthood - this includes letting go of the teenager and allowing them to find their own way

Videos on transition from pediatric to adult care

ERNICA sexual support website for patients, parents and healthcare professionals

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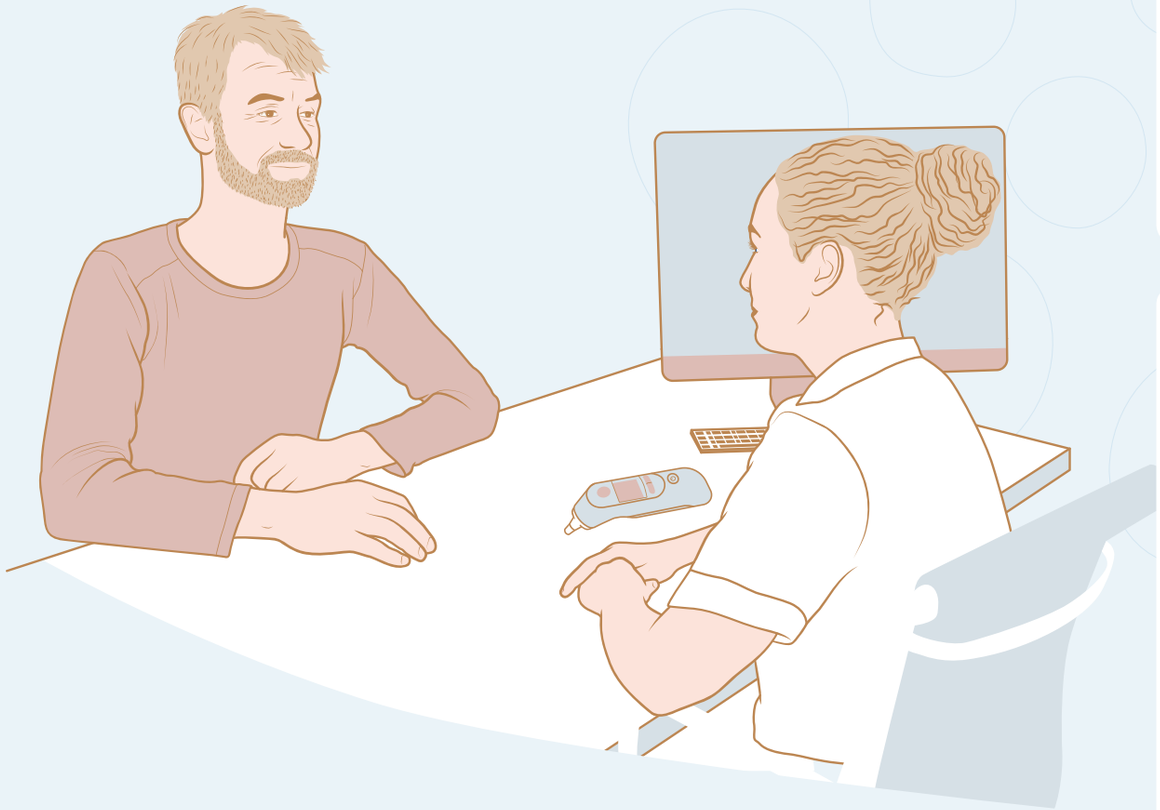
Adulthood

Adults need a doctor for adult treatment who has knowledge and experience of HSCR. Some questions related to the condition may only arise in later stages of life.



Questions often asked by HSCR patients:

- If there are residual symptoms: How is improvement possible?
- What can be expected in the course of life?
- How likely is passing on the condition genetically (clinical geneticist advice)?
- Are there special issues in the event of pregnancy?

Necessary action

- If necessary, medical check-ups by a doctor who has information about the surgical background of HSCR
- Consultation of specialists, should it be required

Ideally

- The adult knows which experts can be contacted if medical advice or treatment are needed
- The adult has a good quality of life with as few HSCR associated restrictions as possible

Videos on transition from pediatric to adult care

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