PATIENT JOURNEY

Intestinal failure in Short Bowel Syndrome (SBS)

FEBRUARY 2025

European Reference Network | ERNICA

1 INTRODUCTION
What is SBS?

Short bowel syndrome (SBS) is one of the causes of intestinal failure (IF). SBS results from extensive surgical resection of bowel required to treat a congenital intestinal defect and/or intestinal necrosis. This leads to an intestine that does not function sufficiently.

The most prevalent underlying conditions leading to chronic intestinal failure in children are;

1 - Short bowel syndrome.

2 - Intestinal motility disorders (e.g. Paediatric Intestinal Pseudo Obstruction, PIPO).

3 - Congenital enteropathies (e.g. microvillus inclusion disease).

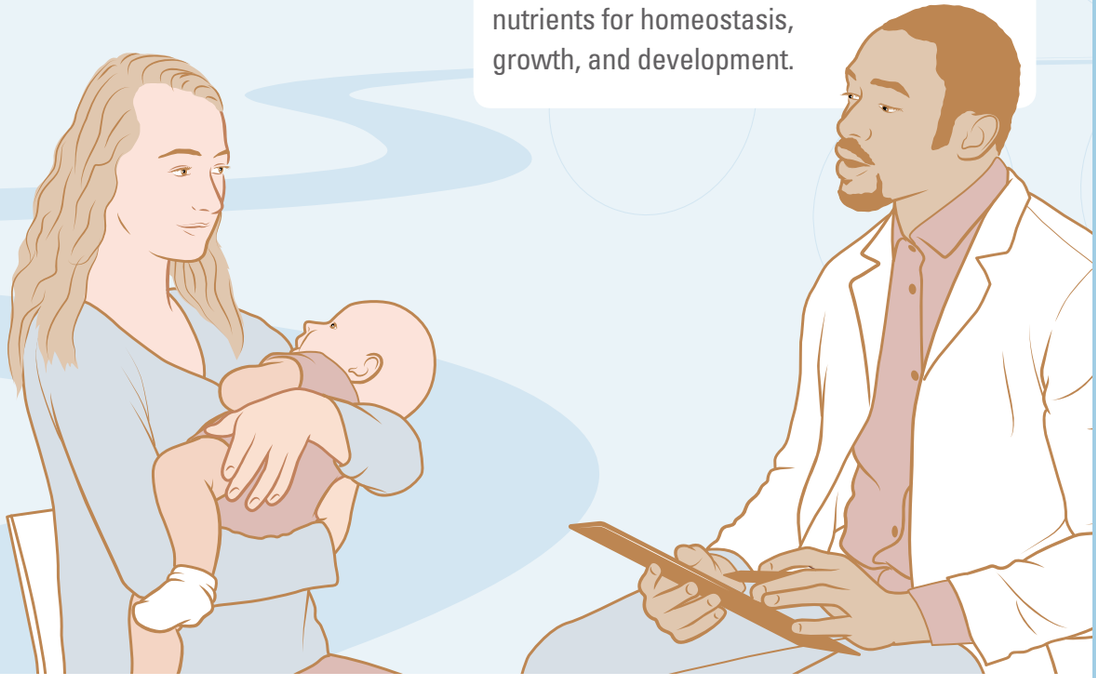
This patient journey provides general information to help you understand the disease SBS. However, please keep in mind that **each patient case is unique**.

About this patient journey

This patient journey for **Short Bowel Syndrome** (SBS) emphasises the high complexity of managing this challenging and multifaceted group of this disease. This journey shows the importance of a tailored, multidisciplinary approach to care, the need for ongoing research and the critical role of patient and family support throughout a patient's life.

A rare condition

Intestinal failure (IF) in children is a rare condition characterised by the gut's inability to digest and absorb required nutrients for homeostasis, growth, and development.



ERNICA Intestinal failure diagnosis

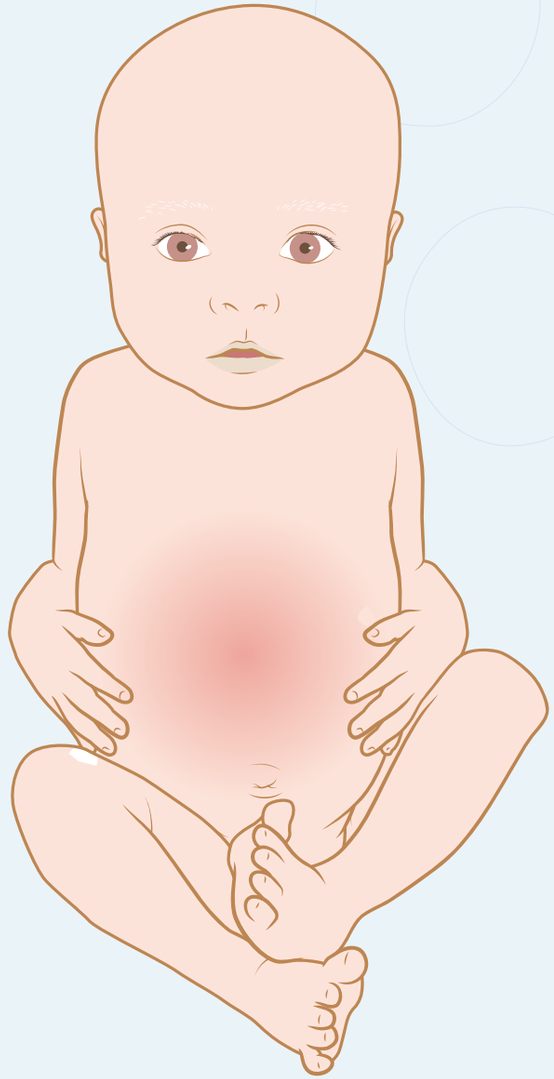
Educational animation for parents and families on Intestinal failure

European Reference Network for rare Inherited Congenital Anomalies (ERNICA)

PATIENT JOURNEY

2 First symptoms

In the case of short bowel syndrome (SBS), symptoms arise after loss and/or surgical removal of a substantial portion (greater than 50%) of the small intestine, with or without the ileocecal valve or colon. These symptoms include diarrhea, malabsorption and nutrient deficiencies.



Necessary action

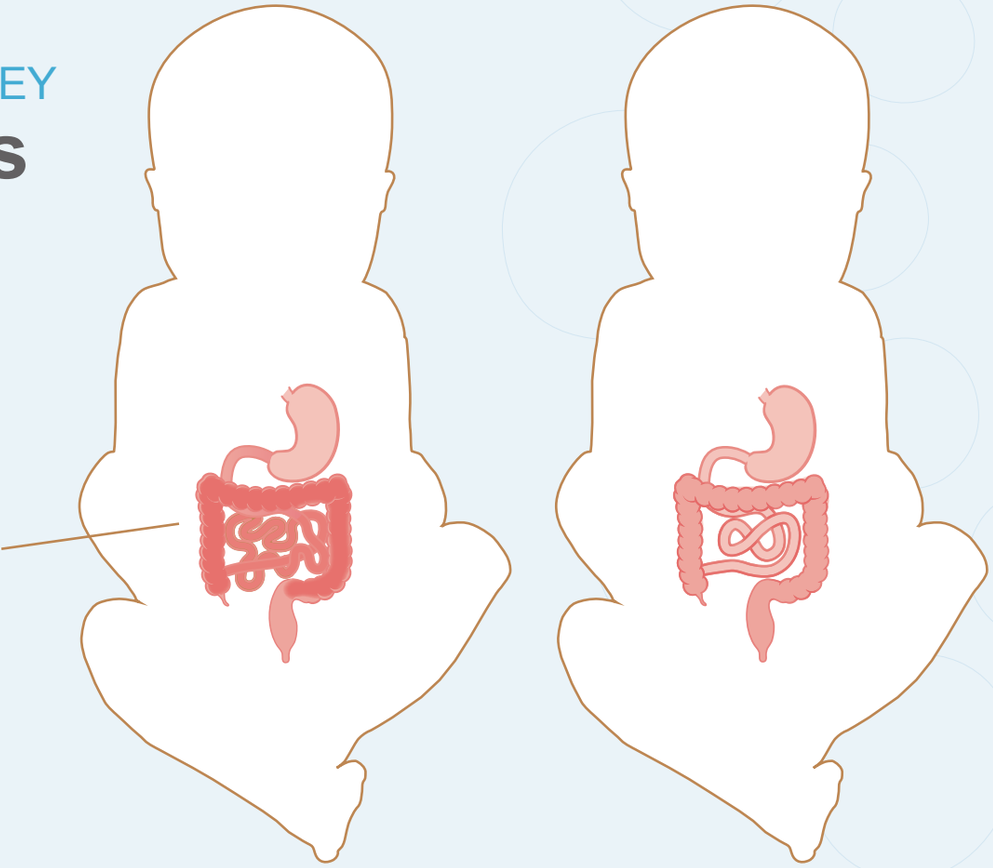
- In the case of suspected SBS, the child should be taken to an intestinal failure expertise centre for medical examination.
- Depending on the specific situation, surgery for stoma creation (an opening of the bowel to allow the passage of feces and air), gastrostomy, or intestinal biopsies may be required. Additionally, endoscopy with biopsies may be necessary to assess the upper and lower gastrointestinal tract.
- Genetic testing on indication
- In some cases, ultrasound scans during pregnancy can detect something that indicates higher risk - (for example, the presense of another condition such as gastroschisis). In such cases, it's important to guide the parents, provide them with specific information and/or tests and monitor the pregnancy carefully.
  In these cases it is also good to contact an expertise centre and make a plan for the birth and possible complications.

Ideally

- Parents are well informed by doctors about the suspected disorder and next steps.
- Parents are informed that SBS is a rare condition, which needs specialist knowledge and treatment.

Animation for parents and families: what is paediatric intestinal failure

3 PATIENT JOURNEY
Diagnosis



How to get a diagnosis:

To obtain a diagnosis of intestinal failure, a combination of clinical evaluation, laboratory tests including blood tests, urine and feces samples, imaging studies, and, if indicated, biopsies and genetic testing are needed.

SBS: clinical diagnosis follows after loss and/or surgical removal of a significant portion of the intestine and is dependent of the extent of surgery. It is confirmed by the presence of clinical symptoms.

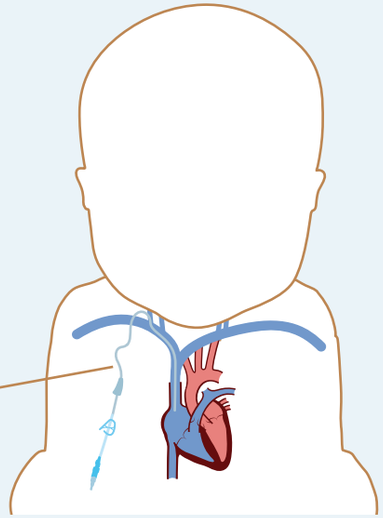
Necessary action

- Confirmation of SBS diagnosis and start of multidisciplinary specialized care as early as possible.
- Parents should learn about how to look after their child's particular needs.
- Support from a specialised nurse/stoma therapist, if required.

Ideally

- Early diagnosis of SBS
- The measures mentioned under Necessary action will increase the quality of care provided to the child.
- Psychological, financial and social support.
- Parents should be referred to an **IF/Home parenteral nutrition (HPN)** patient organisation (if this is available), for more information and advice and contact with fellow patients. Contact with other parents and patients can help them immensely. As SBS is a rare condition, patients and their parents can sometimes feel isolated or may feel isolated. Early contact can help them identify and support their needs.

4 PATIENT JOURNEY Surgery

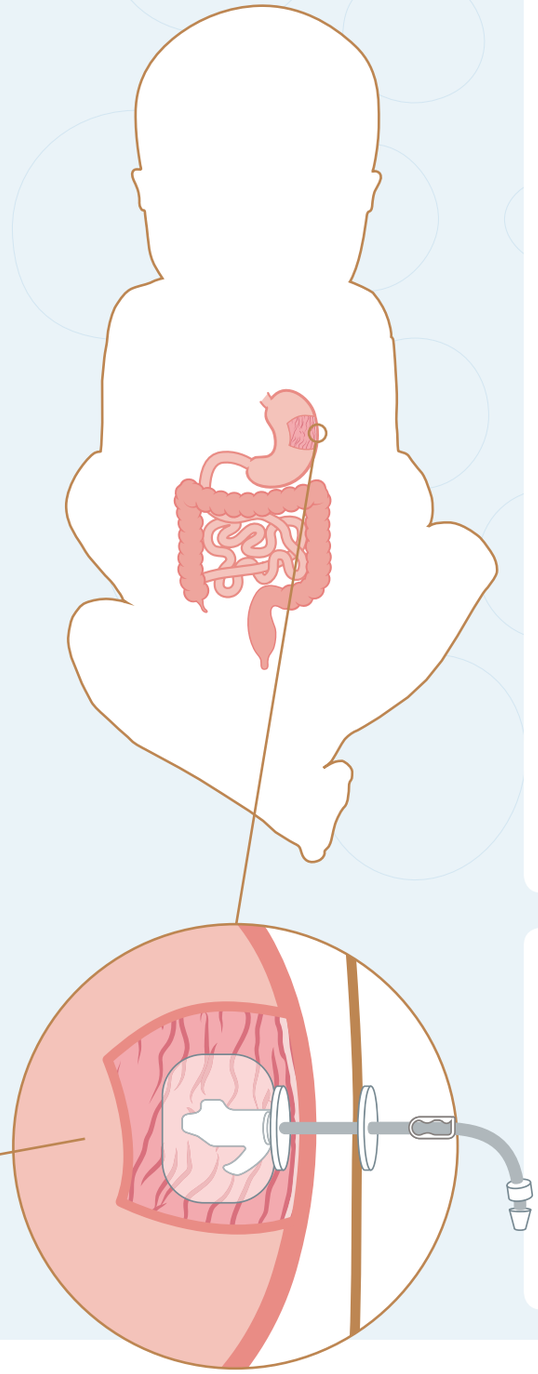


The central venous line passes through the superior vena cava into the heart chamber. Parenteral nutrition is administered into the bloodstream via this route.

First steps

Most children with SBS have a central venous catheter (CVC). There are different types of catheters, but for PN treatment it is important that the CVC is a tunnelled catheter. This type of catheter is placed under the skin and into a central vein. This type of catheter is often used for long-term purposes.

For SBS, the first surgery is the resection leading to SBS. But to treat SBS, the first surgery is to close the stoma when possible. Next steps may be autologous intestinal reconstructive surgeries in selected patients (treatment of strictures, tapering and lengthening procedures). In cases where the remaining small intestine is very short, the use of Malecot tubes may be preferred over more traditional stoma solutions. This approach helps to preserve as much of the small intestine as possible.



This is a gastrostomy, an opening (stoma) through the abdominal wall and into the stomach. It is used for removing the contents of the stomach, typically through a gastrostomy tube. It is commonly performed to relieve gastric distension or bloating, remove excess gas, fluids, or food, and prevent vomiting.

Necessary action

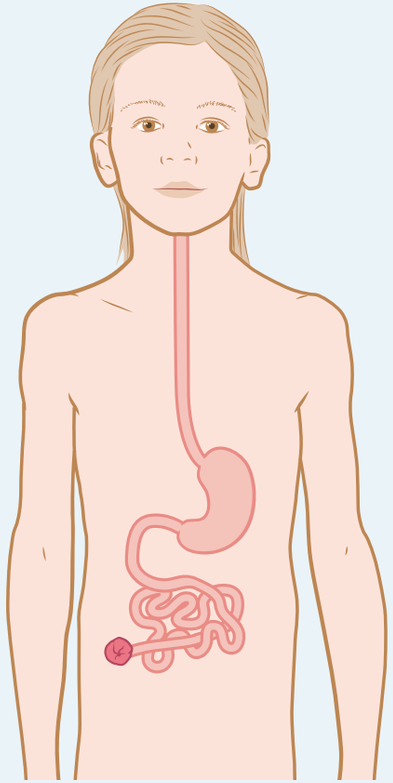
- Start training on how to manage Parenteral Nutrition (PN), including taking care of the central venous catheter. If needed, provide training on how to care for stomas (gastro-, ileo-, jejuno- and or colostoma).
- For children with a temporary stoma, a second surgery should be planned. In the meantime, children should always be optimally fed by enteral and parenteral nutrition.
- Before any surgery, parents should be well informed about the procedure, its potential risks and benefits, as well as possible post-surgery complications and their management.

Guidelines on paediatric parenteral nutrition: home parenteral nutrition

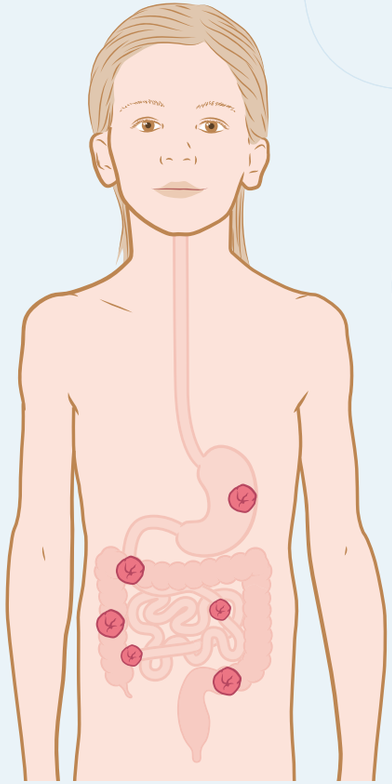
Ideally

- Information about problems that might arise after surgery and how these can be managed.
- Follow-up care at the clinic and provision of contact details in case of emergencies.
- Parents should have access to their child's medical records and operation reports if requested.

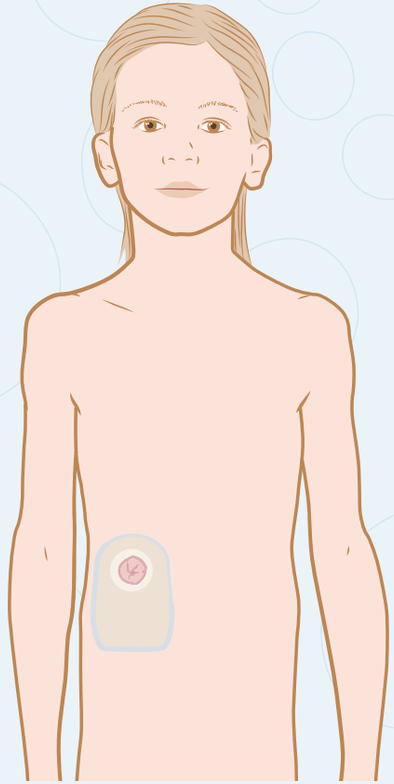
4 PATIENT JOURNEY
Surgery



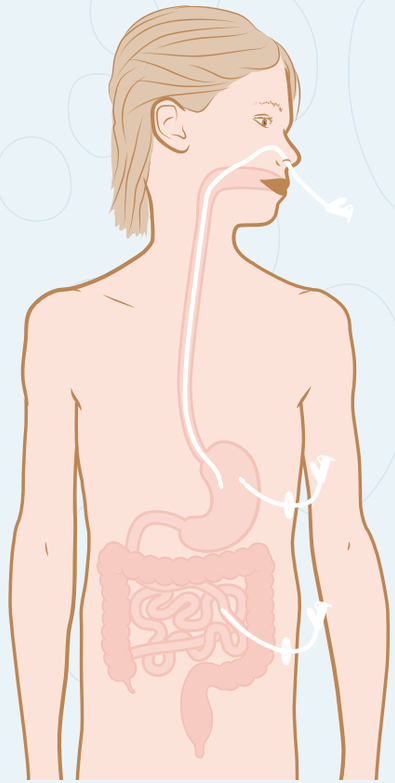
This is a stoma. There are several types of stomas in the abdomen. A stoma is a piece of intestine that has been brought to the surface through an opening in the abdomen and attached to the skin.



Gastrostomy can be placed in the stomach to administer nutrition or to drain gastric contents. The stoma can also be placed in the small or large intestine. Some stoma allow both: stool goes further to bowel and to the stoma bag instead.



Stool can be collected in a bag and replaced when necessary.

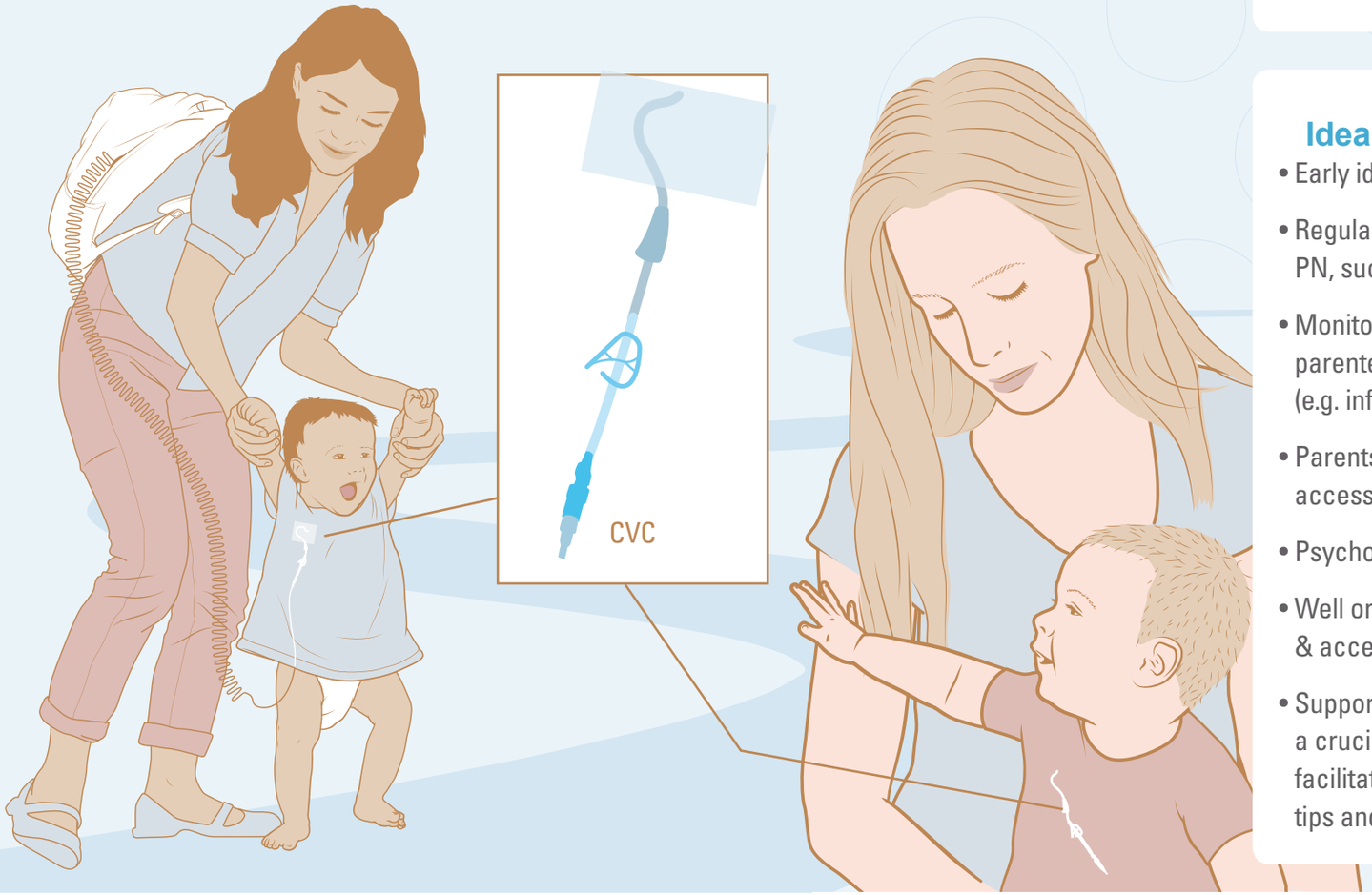


There are several types of tubes for feeding: nasogastric tube, gastric tube and jejunal tube.

PATIENT JOURNEY

5 Follow up care

Before a patient with SBS can go home, the focus should to be on stabilising their condition, correcting any fluid and electrolyte imbalances, and providing nutritional support, often through parenteral nutrition (PN).
If possible, oral and/or enteral nutrition should always be started.



Necessary action

- Regular medical check-ups are needed to identify and treat potential problems early.
- Specialised nurses help to deal with problems such as the growth of skin e.g. around where the CVC is inserted.
- Help with nutrition for eating or growing problems.
- Multidisciplinary treatment including the availability of psychological and physiotherapy support.

Ideally

- Early identification of potential problems.
- Regular check-ups by a medical team involved with PN, such as gastroenterologists, surgeons and dieticians.
- Monitoring for complications of SBS and its treatment, such as parenteral nutrition associated liver disease, central problems (e.g. infections, trombosis, and technical failure) and growth failure.
- Parents know who to contact in case of emergencies and have access to **24/7** support.
- Psychological, financial and social support.
- Well organized, trusted delivery of PN at home & access to medical supplies.
- Support to address quality of life. Patient organisations play a crucial role in supporting families by fostering connections, facilitating the sharing of experiences, and providing valuable tips and advice to help them navigate challenges together.

5 PATIENT JOURNEY
Follow up care

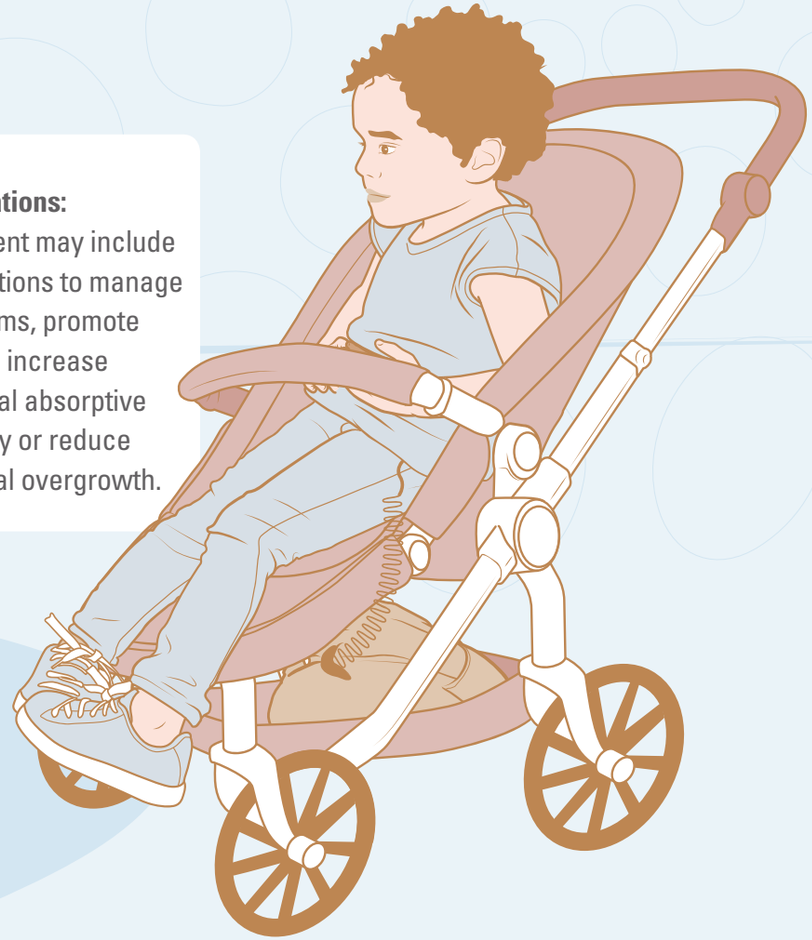
Long-term Management:

PN: Many patients with SBS are dependent on PN. The ultimate goal is to improve quality of life, and care should be managed at a specialist centre. In cases of rare diseases, such as SBS, it's important for doctors, patients, and parents to be informed about the relevant expertise centres in their country and beyond.

Surgical Interventions: Depending on the cause and complications, surgery may have a place in the treatment plan. Decisions should always be made in good collaboration with the IF team including dietitian and paediatric gastroenterologist.

SBS: Medications and procedures to increase the intestinal absorptive area or reduce or increase motility can be considered in expertise centers Surgery might be needed: for example, in case of severe problems caused by enlarged part(s) of the small intestine.

Medications: Treatment may include medications to manage symptoms, promote motility, increase intestinal absorptive capacity or reduce bacterial overgrowth.



Tips and tricks

Animation for parents and families: changing the central venous catheter dressing for children at home

Animation for parents and families: disconnecting the parenteral nutrition infusion for children at home

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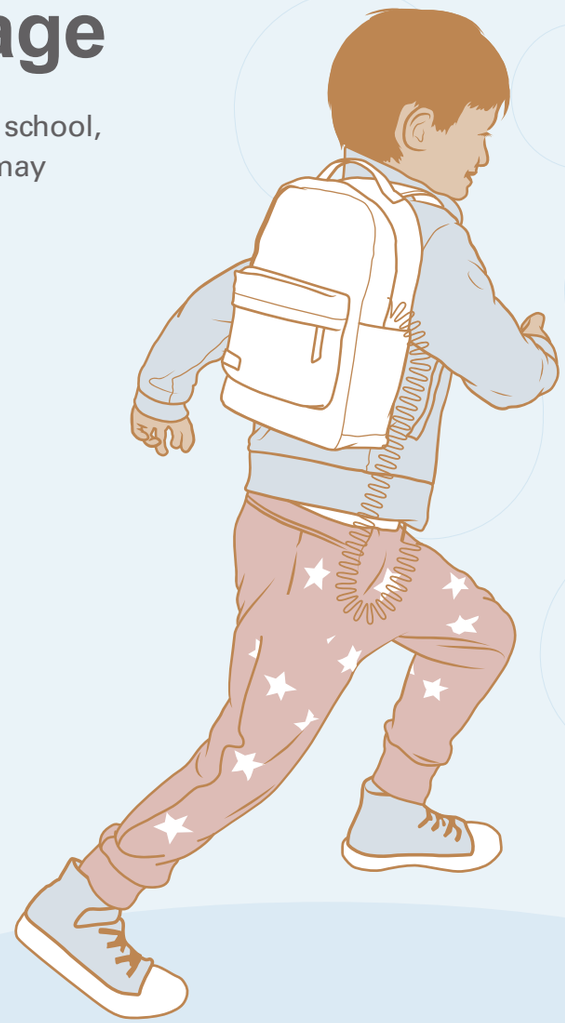
Primary school age

A lot changes in a child's life when they start school, especially if they depend on PN. Questions may arise including: Can they take part in typical everyday activities of children their age? What support might be needed at school?



Starting from primary school age, it's important to raise awareness of your child's condition and and support self-management. Steps for this may include:

- Help the child understand their condition and what it involves.
- Involve the child in their own care, such as assisting with stoma management and medication preparation.
- Allow the child to take on some responsibility during regular check-ups.

Starting this is crucial, as it supports both the child and their parents to make a smooth transition into puberty and adulthood.

Necessary action

- Research which school is the most appropriate for the child to go to; regular school or specialised school.
- Give training and instructions to teachers or caretakers and/or nominate someone to be a point of support.
- Provide other support that may be needed. Contact with other parents and patients might be of benefit.
- A child with SBS might have the need to go to the toilet many times during the day. Making sure there are possibilites so that the child feels safe (for example separate toilet and the awareness so that it's not questioned if child leaves in the middle of class).

Ideally

- The child can go to school as parents feel well supported.
- The child can take part in typical everyday activities in the same way as children their age.

Intestinal failure vacation tool

7 PATIENT JOURNEY Puberty

As a child moves into adulthood and goes through puberty, several key questions often come up:

- How can the teenager become independent / autonomous and manage their own body with a CVC and PN?
- How can the teenager deal with their condition in their personal/social life?
- Does their condition affect sexuality /relationships?

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 SBS specific 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 from around 15-16 years of age at the latest.
- The process includes discussing sexuality issues.

Ideally

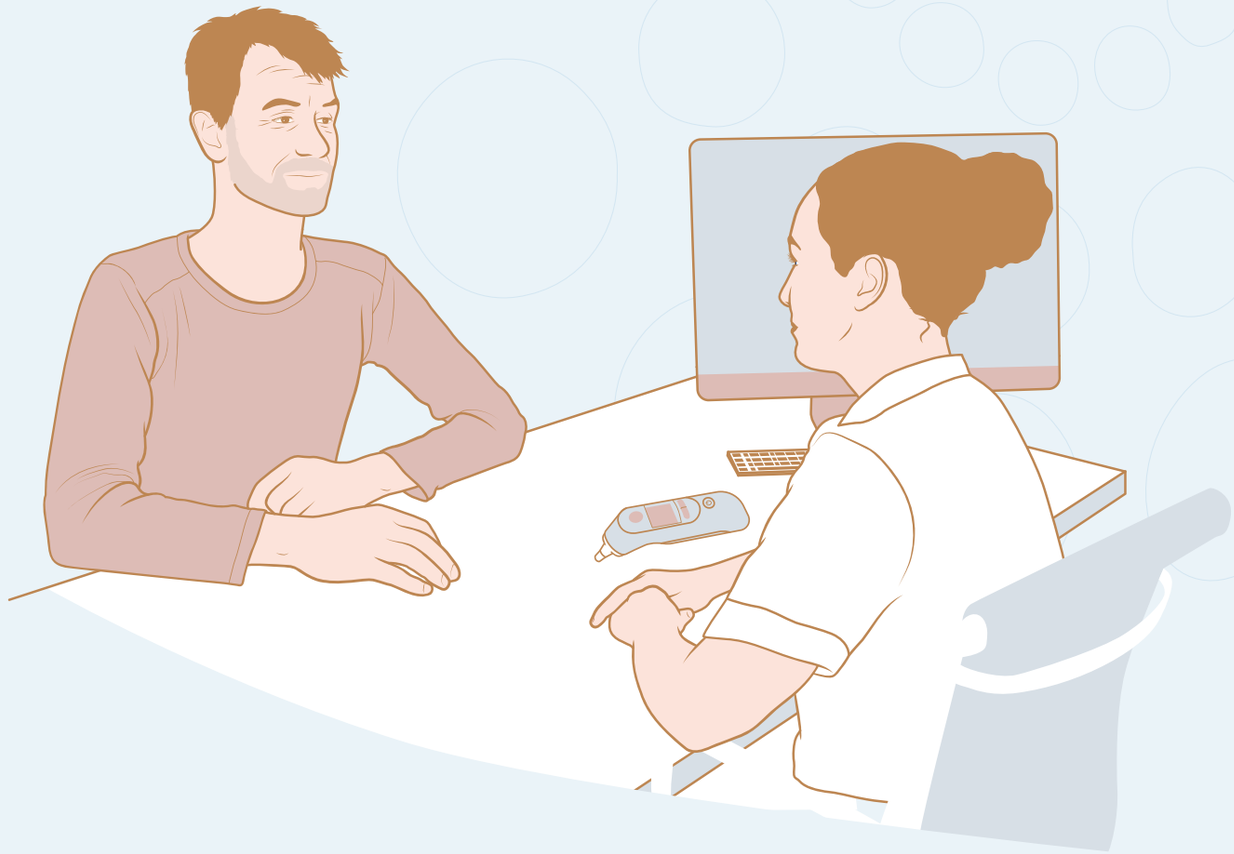
- The teenager can lead an independent life with as few restrictions as possible and knows how to manage their condition.
- They find their own way of dealing with their condition in their private and public life and knows where to get help, if needed. Contact with patients the same age may be of benefit.
- Parents support their teenager to find their own way independently into adulthood.

Animation for parents and families: Transition of care: **Parent-child interactions**

Intestinal failure vacation tool

8 PATIENT JOURNEY
Adulthood

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



Questions often asked by SBS patients:

- What can be expected throughout the course of life?
- How likely is it to pass on SBS genetically?
- Are there special issues or considerations in the event of pregnancy?
- What kind of complications could occur in the long-term?

Necessary action

- If necessary, medical check-ups by a doctor who has knowledge of SBS.
- Consultation of specialists, should 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 SBS-associated restrictions possible.

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