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First symptom ...

... treatment...

...long term follow up...

1. First Symptom

Congenital malformation of the bowel/anus such as absence of anal opening, incorrect position of anus or anal stenosis. Failure to pass meconium
Signs of bowel obstruction including abdominal distension and vomiting.
In females: presence of one single orifice (cloaca) in which the vagina, the rectum and the urethra join together thus forming one single common channel.

2. Diagnosis

Prior to discharge all infants are examined appropriately. Any infant (particular if breast fed) presenting with any of the following should be referred for further assessment:
Delay in the passage of meconium (>48 hrs); constipation within a few days/weeks of life; passing ribbon stools; abdominal distension and vomiting.

3. Treatment & 4. Surgery

4. Long-term Follow Up

Problems with bowel management, urological issues with problems fertility/infertility, sexual problems, gynaecological ones and psychological issues. All these issues are part of the adult world. Only now that we are more exposed to the adult population and we are starting studying them we know that the malformation is not something that it is possible to correct in early life and then everything is over. Community support and follow-up is vital following surgery, as bowel problem may develop later problems. All children should be put on a bowel management programme and the parent/carer given advice and information.

Note: immediate surgical intervention opening a colostomy on the abdomen of the patient in order to give the possibility to the child to pass meconium. Evaluation of the associated defects, urological evaluation such as: sonography, MRI, x rays, cystoscopy, urodynamic etc. Holistic care to maintain health status.

Ideally: Infant seen in specialized centre expert in anorectal malformation where under the care of clinicians can give all the treatments needed.

Note: Timely diagnosis will be made on the presenting clinical signs and symptoms which may trigger further investigation including abdominal x rays and rectal biopsy. If the infant has not passed meconium prior to an early discharge following birth then the family should be informed to contact their health care profession if no meconium is passed within 48 hours after birth.

Ideally: Family are given access to written information regarding the infants problem and if possible put in touch with other families, patients' organization who have had a child with a similar. Parents after the opening of the colostomy may have a paper in which is outlined each stage of the malformation with the relative timing and exams to be carried out and the next meetings at the hospital. Parents get a brochure/booklet explaining what is the malformation and its implications.

Treatment: The family may well need emotional support as well as practical help. Particularly if the infant is transferred to specialist centre away from home.

Ideally: The family is offered accommodation if the infant has been transferred a long distance from home so they can stay close to the infant. The family know who is the case manager of their child (meaning who is the doctor in charge of their child), they are also offered psychological consultation.

Surgery: As surgery is sometimes completed in stages then the family should be given clear information regarding the child's treatment plan timeline with opportunities to ask questions.

Ideally: The family is in an expert centre for this kind of procedures and they are reassured that the treatment being offered is the optimum available to meet their child's needs and they are aware of expected outcomes. Multidisciplinary team is needed in this stage to evaluate the patient's need and the family should be able to talk to the different clinicians to know what they are considering for their child.

Ideally: The family are aware that the surgical intervention to correct the abnormality is only the start of the process and their child may well require ongoing bowel monitoring and management as necessary. The family should have access to written information and a management plan. The child is under the care of a clinician with specific expertise in the management of bowel problems in children. The family should be offered assistance by a specialized nurse who is able to help the family at home to understand how to cope with the child and the catheterization or the colonic irrigations as well as how to deal with a colostomy or with rush after the colostomy closure.