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Management of newborns with anorectal malformation

Obravnava novorojenčkov z anorektalno malformacijo

Polona Studen Pauletić,[1,2] Domen Plut,[2,3] Diana Gvardijančič[1]

Abstract

This review article presents the management protocol of new-borns with anorectal malformation in Slovenia. The clinical presentation, diagnostics and surgical management are described. Optimal functional result is possible when the condition is recognised early and managed according to the described protocol. We have searched the literature for guidelines on anorectal malformation treatment, and present some data of a single centre (University Medical Centre Ljubljana, Slovenia) retrospective analysis of anorectal malformation management. Our achievements are the result of a well-coordinated multidisciplinary approach and adherence to the treatment protocol.

Izvleček

Pregledni članek predstavi protokol obravnave novorojenčkov z anorektalno malformacijo od kliničnega pregleda, diagnostičnih metod do kirurškega zdravljenja, ki novorojenčku omogoča odvajanje blata. Zgodnja prepoznava in ustrezno zdravljenje omogočata optimalne možnosti za dober funkcionalni izid. Pregledali smo sodobno literaturo o obravnavi anorektalnih malformacij in zbrali okviren pregled naših izkušenj v Univerzitetnem kliničnem centru Ljubljana. Naši rezultati so posledica uspešnega sodelovanja multidisciplinarnega tima in spoštovanja protokola obravnave.

1 Introduction

Anorectal malformations (ARM) are congenital developmental abnormalities that involve the development of the lower gastrointestinal tract, urinary tract, and genitals. The spectrum of developmental anomalies is diverse, ranging from less to more complex. The incidence of ARM is about 1 in 5,000 newborns (1).

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Ključne besede: anorektalna malformacija; radiološka diagnostika; kirurško zdravljenje; posteriorna sagitalna anorektoplastika

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The causes of these developmental abnormalities have not yet been fully clarified, but both genetic and environmental factors play a role in aetiopathogenesis. The development of the hindgut is controlled by a number of genes and signalling pathways. Genetic susceptibility combined with the influence of environmental factors can lead to developmental errors. More and more answers are being offered by research on animal embryos with either a genetic or chemically induced developmental disorder (2,3).

In ARM, prenatal diagnosis is often unreliable. In most cases, parents do not know that the child will have a congenital developmental abnormality.

2 Classification

The long-established Wingspread classification (1984) classifies ARMs into high, medium, and low (4). Later, the Krickenbeck classification (2005) became more established, in which ARMs are divided according to the exact position of the fistula, which affects the method of diagnosis and treatment, as well as the prediction of the outcome of bowel function after surgical treatment (Table 1) (5).

3 Treatment

3.1 Clinical examination

The diagnosis is usually confirmed by a thorough clinical examination at birth. The newborn has no anus and cannot pass meconium normally.

A clinical examination should focus on examining the abdomen, genitals, urinary tract and anus, as well as the lower back and sacrum with an assessment of gluteal muscle development. More normally shaped buttocks mean better developed muscles and a less complex anomaly. We also need to examine the navel carefully. If there is no umbilical artery (umbilical cord with two vessels), there may be no kidney (renal agenesis). The abdomen must be carefully palpated for possible pathological masses (enlarged kidney due to hydronephrosis, hydrocolpos, ectopic kidney, etc.) (5). The examination of each newborn should also determine the following: the presence of the anal opening, its size, patency and position in the perineum. If a normally large opening is present within the sphincter muscle complex but is not passable, we speak of the anal membrane, which is one of the less complex ARMs. In girls, we must also pay attention to the presence of vaginal and urethral openings. A complex form of ARM in girls is the cloaca. It is a developmental abnormality with a joint gastrointestinal tract, urinary tract and the vagina, which is often accompanied by hydrocolpos.

In ARM, when the end of the rectum is closed blindly or not in proper position, in approximately 95% of cases a thin connection between the rectum and another structure or the exit to the skin develops in the form of a fistula. The fistula can connect the rectum with different structures and is located at different heights. The simplest to treat is a low-lying fistula leading to the perineum (rectoperineal fistula, RPF), through which meconium secretion can be observed in the first hours after birth. The perineal fistula should not be mistaken for a

Table 1: Krickenbeck classification of anorectal malformations. Taken from Holschneider et al., Journal of Paediatric Surgery 2005 (5).

Common	Rare / regional variants
Rectoperineal fistula, RPF	Pouch colon
Rectourethral fistula (urethra), RUF	Rectal atresia/stenosis
• Rectobulbar	Rectovaginal fistula
• Rectoprostatic	H fistula
Rectovesical fistula (bladder neck), RVezF	Others
Rectovestibular fistula, RVF	
Cloaca	
• Common channel < 3 cm	
• Common channel > 3 cm	
No fistula	

normal anal opening. The fistula is usually smaller and is located at least partially outside the sphincter muscle complex. Sometimes in boys, a longer subcutaneous canal with meconium extending towards the scrotum is present.

Most fistulas are located higher. They connect the rectum with the urinary tract (urethra – rectourethral fistula (RUF), bladder – rectovesical fistula, RVezF) or reproductive system (vaginal entrance – rectovestibular, RVF) in girls. A form of ARM without a fistula is also possible. It is more common in newborns with trisomy of chromosome 21. In newborns without the anal opening and without a visible fistula, we wait at least 24 hours before surgery so that the intestine fills with air and gives us a more reliable assessment of the height that the last part of the colon is located at, which significantly influences the decision on surgical treatment.

During the remaining clinical examination, we must actively look for any associated developmental abnormalities. These are present in approximately half of patients with ARM (6). The incidence of individual associated developmental abnormalities may vary. In general, however, the most common developmental abnormalities are urinary and genital (40–50%), heart (30–35%), spinal cord (25–30%), and gastrointestinal tract (5–10%). Three or more developmental abnormalities at the same time, as part of the VACTERL/VATER association (V – vertebrae/spine and sacrum, A – intestinal atresia (rectum, other, e.g. duodenum), C – congenital anomalies of the heart, TE – oesophageal atresia with tracheoesophageal fistula, R – renal abnormalities, L – limb abnormalities) occur in 4–9% (7). A newborn's wellbeing is most at risk of obstruction of the oesophagus with a fistula into the respiratory tract (tracheo-oesophageal fistula), which is clinically manifested by respiratory distress and swallowing disorders as well as salivation of the newborn, and is diagnostically confirmed by the insertion of a nasogastric tube, which on a chest X-ray is twisted into a loop in the upper part of the chest. The newborn is also at risk of potential congenital malformations of the heart, which can be identified by clinical examination and confirmed by echocardiogram. Radiological investigations are crucial to assess the presence of any other associated developmental abnormalities (8).

3.2 Diagnostics

Laboratory tests in newborns with ARM are not specific. A basic metabolic panel is required as well as determining the blood type and blood clotting factors. Basic screening tests are also required. Urine analysis makes sense in boys with ARM within the first 24 hours if there is no visible fistula, as the presence of meconium in the urine is a direct evidence of the presence of a urinary tract connection.

3.2.1 Primary radiological examination

Radiological diagnostics, namely X-ray or ultrasound (US) examinations in clinically unclear cases, help to decide on the appropriate treatment with assessing the height of the closed segment.

It is important that the X-ray imaging is performed in a child with ARM within 18–24 hours after birth so that the gas in the blind-ending rectum can move completely to the lowest part of the closed intestine, as an earlier assessment could therefore be erroneous (9). X-ray imaging is performed with the child lying in prone position. The child must be in this position for at least three minutes before imaging so that the gas in the intestine can be distributed to the lowest part. Before imaging, the area on the skin where the anal opening should be present is marked with a radiopaque marking. X-ray imaging is performed with a horizontal beam. In the X-ray evaluation, the distance between the lowest part of the large intestine and the mark on the perineum is measured. A distance greater than 2 cm indicates a high ARM, and a distance less than 2 cm indicates a low ARM (Figure 1). The dose of ionizing radiation that the child receives during this imaging is very small in Slovenia (DAP = 0.1–0.2 mGy·cm^2, an estimate of around 0.005–0.001 mSv), as the estimated annual dose due to the natural background is around 2.4 mSv (10).

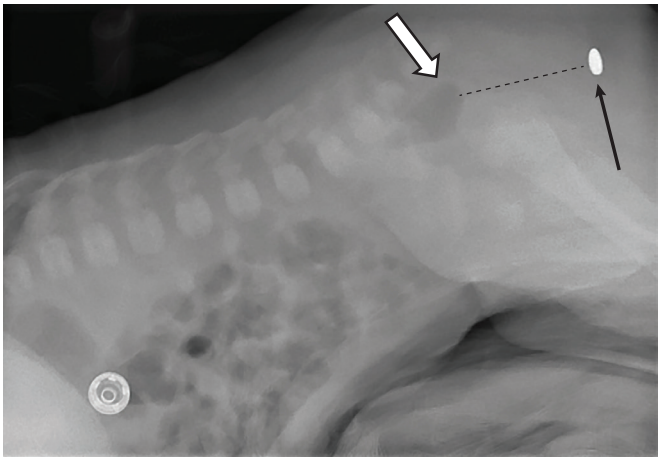

Figure 1: X-ray imaging of the abdomen with a horizontal beam of rays in a newborn with anorectal malformation. The black arrow indicates a radiopaque marking on the skin where the anal opening should be present. The hollow arrow indicates the gas-filled blind end of rectum.

Estimation of the height of the closed segment can also be performed by an ultrasound examination (US), with a high-frequency ultrasound probe. The assessment is most easily performed with a transperineal approach (through the perineum), as it can directly measure the distance between the site on the skin where the anal opening should be present, where the ultrasound probe is placed, and the lowest part of the rectum, which in ultrasound is seen as a bag filled with meconium or gas (11).

As part of the primary radiological management of a newborn with ARM, we must also perform investigations to assess possible associated developmental abnormalities of other organ systems. X-rays (chest, spine and pelvis), ultrasound (heart, abdomen and spinal canal) and, if necessary, magnetic resonance imaging (MRI) are performed, especially in children with severe skeletal or nervous system disorders (12). All primary radiological diagnosis is usually performed within the first three days (13).

All the information obtained helps the surgeon to decide on the appropriate surgical treatment of ARM, i.e. on the direct perineal surgical repair (one-stage treatment) or colostomy and postponed definitive surgical care (multi-stage treatment).

3.2.2 Secondary radiological examination

In children who have undergone colostomy (multi-stage treatment), the type of ARM should be anatomically defined before the definitive surgical repair. This requires contrast-enhanced X-ray examinations: micturating cystourethrogram (MCUG) and distal colostogram (Figure 2). The two examinations are performed at the same time, so that the lowest part of the gastrointestinal tract, urinary tract and possible fistula filled with a contrast agent are present on the final roentgenogram.

At the examination, we first perform an MCUG. A catheter (usually the thinnest gastric tube or even an umbilical catheter) is inserted into the bladder lumen through the urethra, through which the bladder is filled with a contrast agent (CA). The catheter is left inserted in the bladder. This is followed by performing distal colostogram. A urinary catheter is inserted into the distal stoma (mucous fistula). The catheter balloon is filled and the catheter is withdrawn so that the balloon

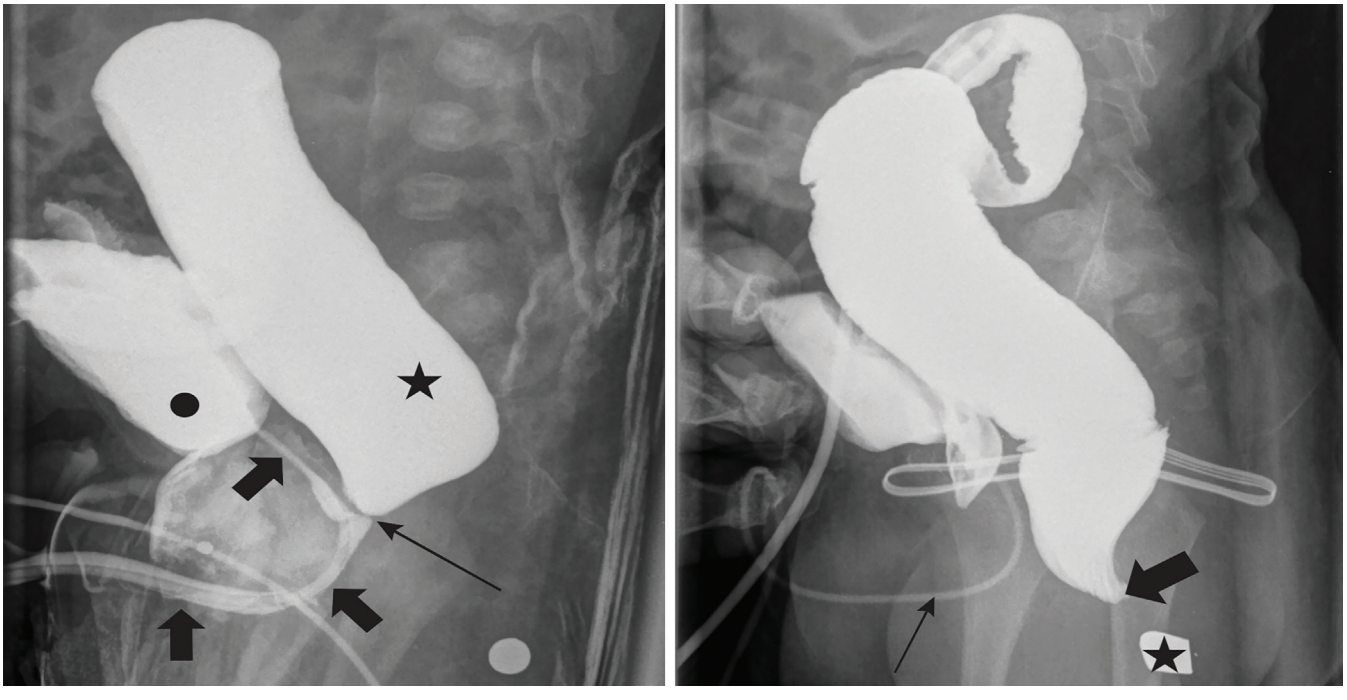

Figure 2: Micturating cystourethrogram in a toddler with anorectal malformation. **Left** – MCUG and distal colostogram in a 2-month-old boy with ARM with a short rectourethral fistula. The bladder is marked with a dot, the rectum with an asterisk. The wide arrows indicate the urethra. The narrow arrow indicates a short rectourethral fistula leading to the bulbar part of the urethra. **Right** – MCUG and distal colostogram in a 3-month-old boy with ARM with no fistula. The wide arrow indicates a blind end of rectum ending low with a short distance to the skin. The urinary catheter showing the course of the urinary tract is marked with a thin arrow. The examination shows no sign of fistula.
Legend: MCUG – micturating cystourethrogram; ARM – anorectal malformation.

seals the stoma opening well from the inside. Adequate sealing is crucial for the display of the fistula, as it allows high pressure to be achieved inside the lowest part of the gastrointestinal tract and the flow of the CA even into very thin openings. The surgeon then injects a water-soluble contrast agent through the catheter into the lumen of the lowest part of the large intestine until the intestine is completely filled, showing a possible fistula. When the CA is injected, the surgeon also gets a sense of the pressure needed to fill the intestine and fistula. The entire procedure is performed under X-ray supervision and with constant communication between the radiologist and the surgeon. At this imaging, the dose of ionizing radiation is slightly higher (DAP of 10–30 mGy·cm^2, which is estimated at about 0.5–1 mSv).

ARM type assessment by displaying the fistula is also possible with MRI, which requires general anaesthesia in young children. In addition to the precise definition of the ARM type, MRI also allows to some extent to show the development of the pelvic floor muscles, which is considered a predictor of faecal continence after treatment (14). In our country, MRI is not included in the regular protocol for the management of all children with ARM. The assessment of the development of the pelvic floor and sphincter muscles is indicated by the shape of the buttocks, and electrostimulation performed by surgeons during the operation is helpful.

In 2019, a case report of 6 patients with ARM was published, in whom the contrast-enhanced ultrasound method was used to define the type of ARM and display the fistula. The method has proven successful, but further research will be needed in a larger number of children to confirm this method. Because the contrast-enhanced ultrasound method does not use ionizing radiation and is completely harmless to children, this method may be able to replace conventional X-ray methods in children with ARM in the future (15).

3.3 Surgical treatment

The decision on the method of surgical treatment is influenced by the complexity of the abnormality and

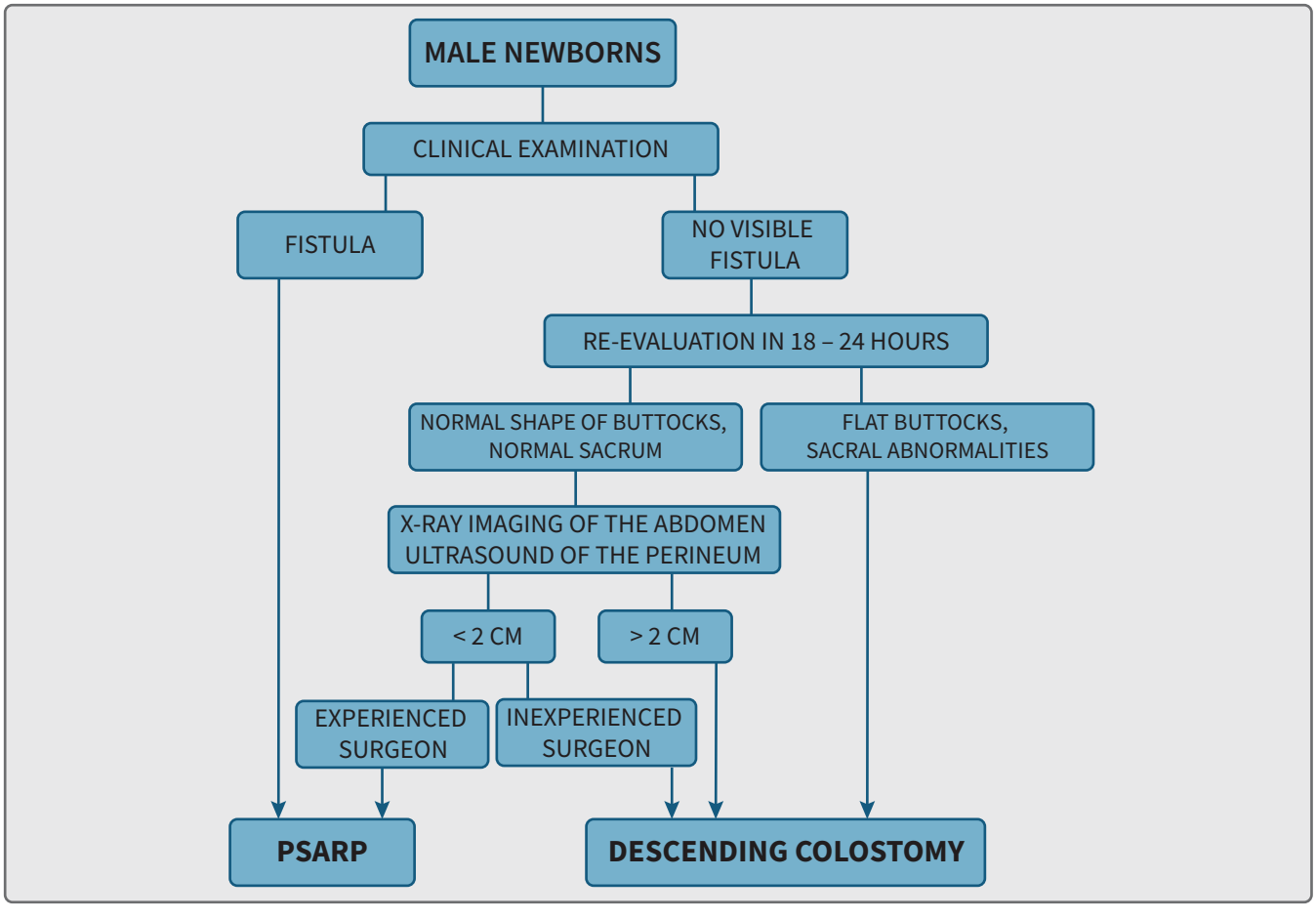

Figure 3: Schematic representation of the management of male newborns with anorectal malformation.
Legend: PSARP – posterior sagittal anorectoplasty; UZ – ultrasound examination; RTG – X-ray imaging.

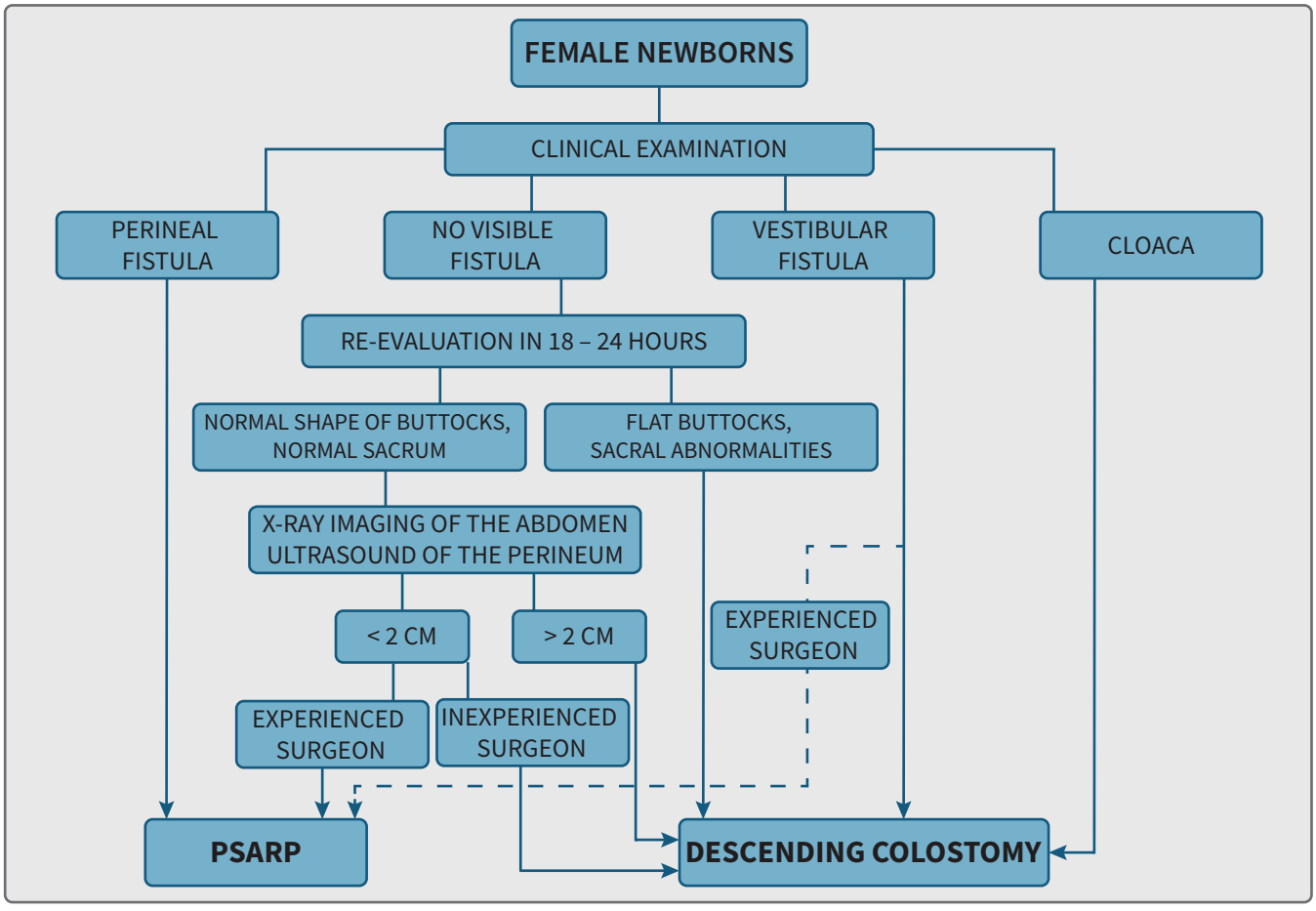

Figure 4: Schematic representation of the management of female newborns with anorectal malformation.
Legend: PSARP – posterior sagittal anorectoplasty; UZ – ultrasound examination; RTG – X-ray imaging.

any accompanying developmental abnormalities. The surgery is planned within 24–48 hours after birth. After a clinical examination and primary radiological examinations, a decision must be made as to whether definitive one-stage surgical treatment with posterior sagittal approach (posterior sagittal anorectoplasty, PSARP) is feasible in the newborn or whether multi-stage (three-stage) treatment with colostomy will be required as the first stage of surgical treatment. Schematic approach to treatment in boys and girls is shown in Figures 3 and 4. As a rule, all newborns with a visible fistula in the perineum and without associated developmental abnormalities that would jeopardize the procedure are treated in one stage. This avoids the need for additional procedures and complications that may be associated with the presence of a colostomy in the newborn. If the fistula is not visible after 18–24 hours and the primary radiological examination indicates that the blind-ending rectum is located higher (the distance between the rectum and the mark on the perineum is greater than 2 cm), we decide on a multi-stage treatment.

In girls, the most common form of ARM is a fistula into the opening of the vagina (rectovestibular fistula). One-stage treatment in these cases has been shown to be effective and safe, but only if performed early enough (within the first 48 hours) when the risk of perineal wound infection is lower (16,17). At the same time, insufficient experience of surgeons is the most common reason for the need to re-do surgical treatment in specialized centres after failed primary attempts (18).

The safest treatment of such anomalies is multi-stage, which is chosen in most cases.

3.3.1 Colostomy

The first stage of surgical treatment in more complex forms (or in life-threatening accompanying developmental abnormalities) is a colostomy, which allows the newborn to pass stool, which must be ensured within the first 48 hours after birth. The ideal colostomy is in the left lower quadrant of the abdominal wall, namely in the middle of the triangle formed by the lower edge

of the ribs, the navel, and the edge of the intestine. It is formed at the beginning of the sigmoid colon, a part of the large intestine, where the intestine is completely interrupted. The last part is formed into a mucous fistula.

Later, a colostogram can help accurately identify the type of abnormality and plan the operation accordingly (see the section on diagnostics or radiologic examinations).

3.3.2 Posterior sagittal anorectoplasty

Since 1980, the gold standard of surgical treatment has been posterior sagittal anorectoplasty (PSARP), introduced by Peña and de Vries (19). With the posterior sagittal approach, the sphincter muscle complex is well defined. The muscles are also well shown by electrostimulation, the rectum is isolated, the fistula is separated from the urinary tract/genitals by careful preparation, and a new anal opening is formed within the previously mentioned sphincter muscle complex.

At low ARMs, i.e. the less complex ones, the procedure is performed as the only procedure within 36–48 hours after birth.

As a continuation of treatment of higher ARMs, i.e. in more complex cases, PSARP is usually performed at 2–6 months of age. When the fistula and the blind-ending rectum are located very high, additional preparation of the intestine is sometimes required, which can be performed by laparotomy or laparoscopically. The new anal opening is formed to a size guided by the development of the sphincter muscles, which in most cases, is too narrow for normal defaecation. Therefore, after their surgical wound in the perineum has healed, all patients undergo the process of dilating the new anal opening.

3.3.3 Colostomy closure

The third stage of surgical treatment is the closure of stoma to establish the continuity of the colon to the anus. The procedure is not performed at a certain age, but when, with regular dilatation of the anal opening with the help of parents, we achieve a sufficient size of the opening that will allow unobstructed defaecation. This is usually within 2–4 months of the PSARP, and optimally before the age of 12 months.

4 Our experience

At the Department of Paediatric Surgery of the University Medical Centre Ljubljana, newborns with congenital ARMs have been managed according to the presented protocol since 1997. We reviewed the documentation for the treated cases in the period 2005–2019. One patient was excluded from the study due to death unrelated to ARM treatment.

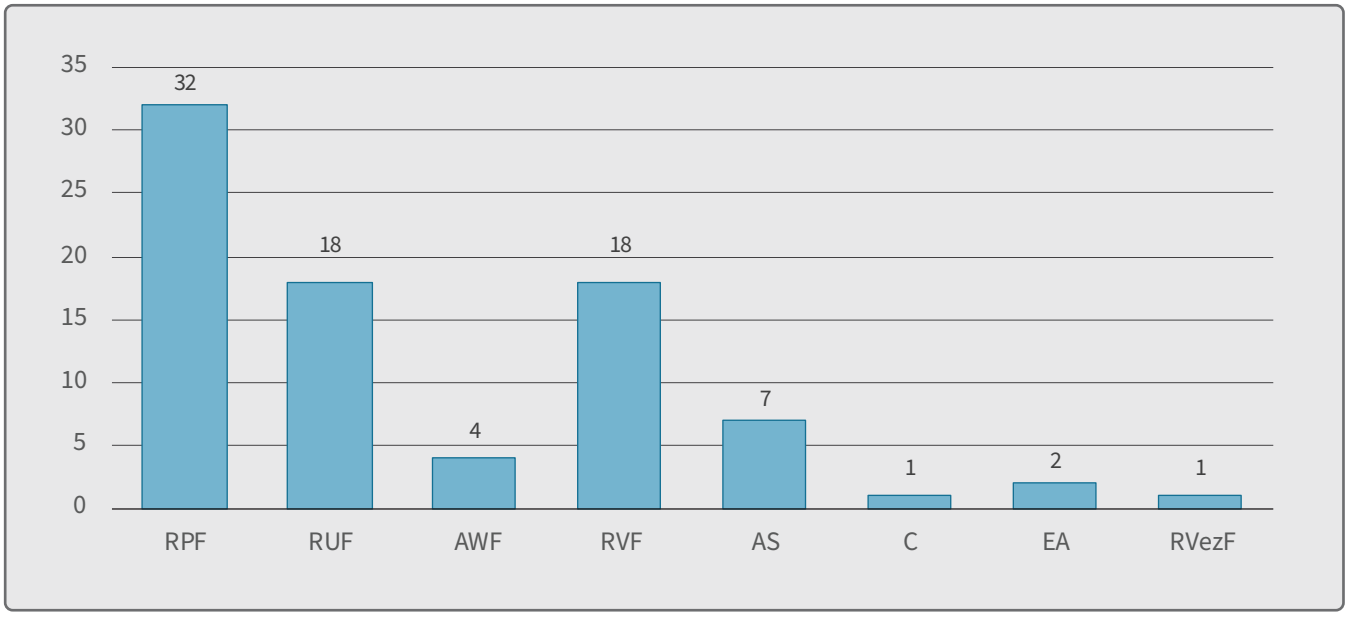

Figure 5: Newborns with anorectal malformation treated at the University Medical Centre Ljubljana, 2005–2019 - layout according to the type of ARM.
Legend: RUF – rectourethral fistula; AWF – atresia without fistula; RPF – rectoperineal fistula; RVF – rectovestibular fistula; AS – anal stenosis; C – cloaca; EA – ectopic anus; RVezF – rectovesical fistula.

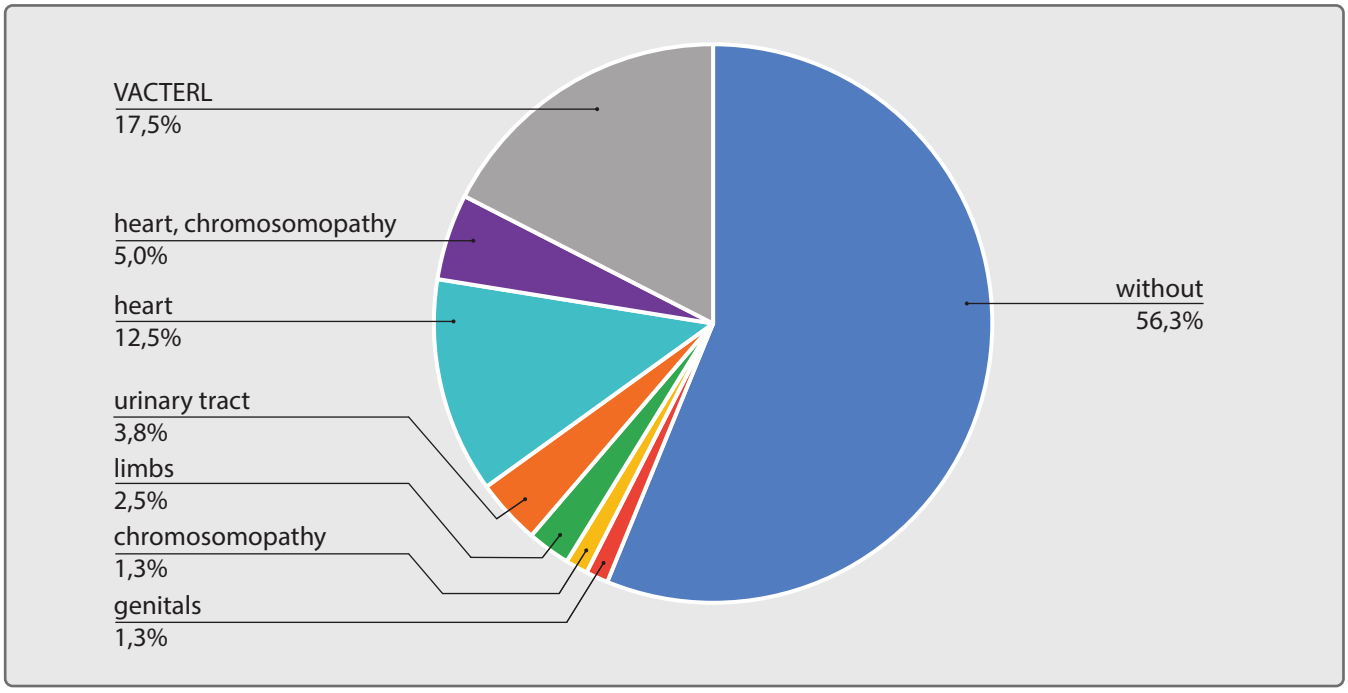

Figure 6: Associated developmental abnormalities in children with anorectal malformation treated at the University Medical Centre Ljubljana in the period 2005–2019.

During this period, we managed 83 newborns with ARM. There were slightly more boys (46/83) in whom ARM with perineal fistula (RPF) predominated, 32/83 (38.6%). In girls, in almost half of the cases (18/37) it was ARM with rectovestibular fistula (RVF) (Figure 5).

Associated developmental abnormalities were detected in 36/83 (43%) newborns with ARM. The most common association was VACTERL in 17.5%. The most common single associated developmental abnormality was congenital developmental abnormality of the heart (12.5%), followed by congenital malformations of the urinary tract and chromosomopathy (Figure 6).

Of the newborns with ARM, 3 were premature but without other associated developmental abnormalities.

In 43 newborns (52%), definitive PSARP was performed. All newborns with perineal fistula (RPF), two newborns with rectovestibular fistula (RVF), and all newborns with anal stenosis (AS) were included in this group. For more complex forms, we opted for a multistage treatment.

PSARP was performed as the second stage of surgical treatment after colostogram at a median age of 4.5 months, i.e. ranging from 30 to 286 days. Among our patients, only one boy has not yet been definitively treated because of a more complex form of the anomaly. During the observed period, surgical treatment was not completed due to complications in one girl with cloaca. For more complex and rarer forms of surgical treatment, we use the experience of experts in colorectal paediatric surgery from abroad. However, there is no established path for us to send such children to specialized centres for treatment abroad.

5 Conclusion

Treating newborns with anorectal malformations is a complex process that involves several stages. Early identification and an appropriate decision on surgical treatment contribute to optimal long-term results. The management protocol, which has been used at the University Medical Centre Ljubljana since 1997 and is constantly being upgraded with improvements, is the result of good multidisciplinary cooperation. In our opinion, and in comparison with the published literature, the outcome of treatment according to this protocol is good, except for very rare, more complex forms of malformations, of which we have little experience so far. Most importantly, of course, the outcome of management is such that it allows the patient to optimally control the stooling, micturiction and sexual function throughout life.

Conflict of interest

None declared.

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