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Congenital Anomalies of the Anus and Rectum*

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There is confusion in the minds of many clinicians about the exact anatomical features of the individual lesions of anorectal anomalies and therefore about the way in which they should be treated. This arises partly because some, like the covered anus, are visible in the perineum, whereas in others part of the anomaly is visible, for example, in a low vaginal fistula, but the more important part, the blind end of the rectum, lies deep in the pelvis separated from the skin by the levator ani sheet of muscle. The indiscriminate use of the term 'imperforate anus' adds to the confusion because it implies that only the anus is abnormal and that the rest of the anal canal and rectum are unaffected. This term suggests that by incising the skin over the anus and cutting deep enough into the midline of the perineum the bowel can be opened and the obstruction relieved. As will be seen later, this term can properly be applied only to the so-called 'covered anus', and the other types should be classified anatomically according to their nature.

Confusion also arises because of the misleading classification of these anomalies into 'high' and 'low' types, the 'low' types including all those in which there is a visible external fistula, and the 'high' those in which the lesion is believed to lie above the levator ani which forms the muscular sheet of the pelvic floor. In fact only covered anus and anorectal stenosis are 'low' lesions in the sense that the abnormality lies below the levator ani. All the others are 'high' lesions in the sense that the rectum ends blindly above the levator ani muscular dia-

phragm, though it may communicate with the exterior by a fistula which passes between the limbs of the puborectalis sling of the levator ani to the urethra, the posterior fornix or lower end of the vagina, or to the perineum. The passage of meconium through such a fistula may be another source of confusion, since even when the perineum is carefully inspected the abnormal source of meconium may not always be noticed. Finally, in the rare atresia of the middle third of the rectum, the normal anus, anal canal, and lower rectum may give the impression that the whole anorectal region is normal and that the cause of the obstruction lies much higher in the bowel.

Congenital anomalies of the anus and rectum were classified by Ladd and Gross (1934) into 4 main groups (Types I to IV). The advantage of their classification is that it is based on the normal and pathological anatomy and is simple. I use a modification of it (Table I). In this there are 4 main types of lesion: anorectal stenosis; covered anus; anorectal atresia involving the lower third of the rectum with or without an associated fistula; and rectal atresia or stenosis affecting the middle third of the rectum.

Anorectal Stenosis

Anorectal stenosis is usually limited to a narrow fibrous ring at any point in the anal canal but most often near the anorectal junction; the anal canal and rectum are otherwise normal (Fig. 1a, b, c). This fibrosis in the wall of the anal canal may rarely be much more extensive and involve a large part or the whole of the anal canal. When the stenosis is simply a narrow ring this can usually be dilated and an anal canal of normal calibre obtained, though dilatation may have to be repeated. If the anorectal stenosis is very extensive it may be necessary to excise the fibrous tissue and mobilize the rectum, so that it can be brought down and sutured to the lower part of the anal canal or the perineal skin. Anorectal stenosis does not prevent the passage of some meconium and the anus looks normal. It is only when perhaps days, weeks, or months after birth the baby is noticed to be straining hard to pass a stool that digital examination of the anal canal reveals a severe degree of stenosis or else inspection of the stool reveals

*In the Personal Practice series of articles an author is invited to give his own views on some current practical problem.
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TABLE I
Classification of Anorectal Anomalies with Number of Patients, Sex, and Deaths in Each Type

<table>
<thead>
<tr>
<th>Lesion</th>
<th>Male</th>
<th>Female</th>
<th>Total Cases</th>
<th>Deaths</th>
</tr>
</thead>
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<tr>
<td></td>
<td>No.</td>
<td>No.</td>
<td>No.</td>
<td>%</td>
</tr>
<tr>
<td>Anorectal stenosis</td>
<td>8</td>
<td>6</td>
<td>14</td>
<td>8</td>
</tr>
<tr>
<td>Anorectal atresia</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Lower third</td>
<td>61</td>
<td>64</td>
<td>125</td>
<td>70</td>
</tr>
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<td>Perineal fistula</td>
<td>4</td>
<td>8</td>
<td>12</td>
<td>7</td>
</tr>
<tr>
<td>‘Low’ vaginal fistula</td>
<td>-</td>
<td>46</td>
<td>46</td>
<td>26</td>
</tr>
<tr>
<td>‘High’ vaginal fistula</td>
<td>-</td>
<td>6</td>
<td>6</td>
<td>3</td>
</tr>
<tr>
<td>Rectourethral fistula</td>
<td>33</td>
<td>-</td>
<td>33</td>
<td>18</td>
</tr>
<tr>
<td>No fistula</td>
<td>24</td>
<td>-</td>
<td>28</td>
<td>16</td>
</tr>
<tr>
<td>Rectal atresia</td>
<td>1</td>
<td>1</td>
<td>2</td>
<td>1</td>
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</tbody>
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Totals                        | 100  | 78     | 178 | 36  | 20.2%

(a) Tracing of median section of pelvis of normal stillborn male, and (b) female, to show normal anatomy. The levator ani has been shown with a dotted line and the puborectalis sling with a solid line. (c) Anorectal stenosis, otherwise normal anatomy. (d) Covered anus, otherwise normal anatomy.

that it is being squeezed out with much effort as a thin streak like a pencil or toothpaste. By this time the obstruction will have caused secondary hypertrophy and dilatation of the rectum which will take months or years to subside and will lead to difficulty in management, even when the stenosis has been adequately dilated. It is usually possible, however, to treat anorectal stenosis by dilatation and obtain a satisfactory result with complete continence.

Covered Anus

In this type of lesion the rectum, pelvic floor, puborectalis sling, and anal canal are all present and normal. The only abnormality is a lid of skin which closes the anus (Fig. 1d) but through which there may be a small fistulous tract passing forwards on to the midline of the scrotum (Fig. 2), or a small opening at one side of the skin lid. In

Fig. 1.—(a) Tracing of median section of pelvis of normal stillborn male, and (b) female, to show normal anatomy. The levator ani has been shown with a dotted line and the puborectalis sling with a solid line. (c) Anorectal stenosis, otherwise normal anatomy. (d) Covered anus, otherwise normal anatomy.

Fig. 2.—Covered anus with sinus running forwards on to the scrotum containing meconium in proximal part and yellowish inspissated mucus in distal part.
other patients there may be a rolled ridge of skin across the centre of the skin lid with or without a fistula (Fig. 3a). This is the only anorectal anomaly which could properly be called 'imperforate anus' since the rectum and the anal canal are normal apart from the cover of skin at the lowest part of the anal canal. Unfortunately, when the skin cover has been excised, whether or not the mucous membrane of the anal canal and the skin of the perineum at the site of the anus are sewn together, the superficial and subcutaneous anal sphincters do not function normally. The child is continent but the anus is patent and there is usually a leakage of faecal stained mucus from the lining of the anal canal (Fig. 3b); the resultant staining of the underclothing may cause embarrassment when the child has to undress in public at school. After excision of the skin lid the orifice of the anus and the anal canal should be repeatedly dilated in order to maintain an adequate passage.

**Anorectal Atresia**

This involves the lower third of the rectum and the anal canal and the rectum ends blindly above the pelvic floor (Fig. 4). In addition, in some patients a fistula runs from the blind end of the rectum, through the limbs of the puborectalis sling of the levator ani to enter the upper end of the vagina (high rectovaginal fistula, Fig. 5), or the lower end of the vagina (low rectovaginal fistula, Fig. 6). In either sex the fistula may open in the perineum in front of the anal dimple which overlies the superficial and subcutaneous parts of the external anal sphincter (rectoperineal fistula, Fig. 7, 8). In the male a fistula may open into the prostatic or membranous urethra, or very rarely into the

**FIG. 3.—**(a) Covered anus with roll of skin and sinus on the right side of it through which meconium has escaped. (b) 21 months later to show patent anus (from Wilkinson, 1969).

**FIG. 4.—**Characteristic appearance of the perineum in a boy with anorectal atresia without a fistula. The site of the anal sphincter can be seen in the centre of the perineum and a little white discharge at the urethral orifice but no meconium.
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Fig. 5.—Tracing of median section of female pelvis as in Fig. 1(b). The paths of high and low rectovaginal fistulae and of a rectoperineal fistula are superimposed.

Fig. 6.—Low level rectovaginal fistula with anorectal atresia (from Wilkinson, 1969).

Fig. 7.—Rectoperineal fistula with anorectal atresia in neonate with imperforate hymen (from Wilkinson, 1969).

Fig. 8.—Rectoperineal fistula in girl aged 9 months to show patulous opening without sphincters separated by bridge of normal skin from vagina.
bladder (rectourethral or rectovesical fistula, Fig. 9 and 10). In both sexes, in the absence of a fistula, there is complete low intestinal obstruction which requires relief by some type of surgical operation.

The rectoperineal fistula, like the other fistulas, passes between the limbs of the puborectalis muscle and provided it can be dilated sufficiently does not require any other treatment; the child is continent because of the relation of the fistula to the puborectalis sling. The presence of any type of fistula in the female is apt to be misleading because meconium is passed and it is not always appreciated at the outset that the meconium is coming from an abnormal opening. Diagnosis depends mainly on a careful inspection of the perineum and on an insistence in seeing the orifice from which the meconium is being discharged.

It is very important to recognize that a fistula consists of a fibrous wall with an epithelial lining, and that there is no striated muscle in its wall and no external sphincter associated with its orifice. It is therefore anatomically wrong to call such a fistulous opening an 'ectopic anus' since anus implies that there is a sphincteric muscular ring round the orifice which can close it by contraction.

In the past it has been strongly recommended that the low fistula opening somewhere near the lower end of the vagina should be dealt with by a local operation. In the original so-called 'cutback operation' one blade of a scissors was placed in the fistula and the other across the perineum; on closing the scissors the lower orifice was enlarged by a cut backwards in a midline of the perineum. This enlarged opening was subsequently kept wide by dilatation. Because the fistula passed through the limbs of the puborectalis sling, if a sufficiently wide channel was made by dilatation, the child was continent except when the rectum filled with faeces and faecal impaction led to spurious diarrhoea. This operation was not always successful in the long term; in some girls faeces were discharged at least partially into the vagina, and while this might be tolerated in the young child it became offensive to older children, especially after puberty. It was an operation which was designed at a time when the risk of a more major procedure, such as an abdominoperineal dissection, was greater because of poorer anaesthesia and postoperative treatment. The preferred abdominoperineal operation is now sufficiently safe to justify the abandonment of this type of cutback operation. (Some surgeons have modified this original cutback operation. They make a more careful and detailed dissection of the lower end of the fistula in the perineum which avoids any major division of the superficial muscles, but I have not used this operation.)

The common belief that lesions might be 'high'
or 'low' implies that there may be a difference in the level of the blind end of the rectum. Though during straining the blind end of the rectum may appear to be pushed closer to the perineal skin, it is still above the pelvic floor as represented by the sheet of the levator ani muscle. It is this idea of the blind end of the rectum being above the pelvic floor which is so important to the future continence of the child. Any attempt to reach the blind end of the rectum from the perineum by cutting upwards in the midline is almost certain to damage the puborectalis sling and the levator ani muscles, and in this way to destroy the future continence of the child, which depends on the bowel passing normally through the limbs of the puborectalis sling and then down to the perineum. It follows that the procedure suggested by Wangenstein and Rice (1930) of judging the type of lesion according to the level of the gas shadow in the blind end of rectum when the child was x-rayed in the inverted position is based on a misconception of the anatomy and is misleading. Moreover, gas does not always reach the blind end of the rectum within 6 hours of birth as was supposed (Wilkinson, 1944).

**Atresia in Middle Third of Rectum**

Atresia affecting the middle third of the rectum is the fourth and much the rarest type of anorectal anomaly. In this the anus, the anal canal, and the lower third of the rectum are normal and there is a gap at the site of the middle third of the rectum, the upper third ending blindly in the hollow of the sacrum. Fistulas are not associated with this type of atresia (Fig. 11).

**Associated Anomalies**

The pelvis must be x-rayed in all patients with anorectal anomalies. Abnormalities of the sacrum and coccyx are commonly associated with anorectal anomalies and may be accompanied by neuromuscular defects in the pelvic floor and anomalies of the urinary tract. It should be recognized also that in the presence of several bony abnormalities of the pelvis the chances of continence being good are reduced.

Intravenous pyelography should always be carried out before the child is discharged from the first admission to hospital.

Between September 1958 and December 1970, 178 patients with anorectal anomalies were treated in the Professorial surgical unit at The Hospital for Sick Children, Great Ormond Street (Table I). In two-thirds of the total the rectum ended blindly above the levator ani, and in most of these there was some type of fistula. The distribution between the various types in this series is similar to what has been found in a number of other published series in which there is enough anatomical detail to allow them to be reclassified in the same way.

There are associated congenital abnormalities in about half of the patients in any large series of anorectal anomalies, the genitourinary (26%), skeletal (21%) and cardiovascular systems (17%) being most commonly affected (Cozzi and Wilkinson, 1968). These associated anomalies are often severe in degree and threaten survival in about half the patients who have them. In this series oesophageal atresia occurred in 21 patients (12%) and only 4 survived. In 118 neonates in the series the risk to survival was graded according to birthweight, complications, and other associated anomalies (Table II). The mortality was low in groups A and B and most of the deaths occurred in group C. There was little to choose between the severe associated anomaly and low birthweight, as factors contributing to death; and of 5 babies who weighed less than 1·8 kg (4 lb) at birth, 4 died. Of 12 babies with rectal atresia who died after colostomy, 5 also had oesophageal atresia; 2 died of respiratory failure associated with prematurity, 3 of cardiac failure secondary to congenital heart lesions, 1 of *Esch. coli* meningitis and peritonitis, and 1 of *Esch. coli* gastroenteritis and a congenital heart lesion. 5 died before any treatment of the rectal atresia could be started, 2 after the first stage of treatment for oesophageal atresia, and 3 so soon after admission that no treatment was possible. None of the deaths was due directly to the making of a colostomy, though there is no doubt that sometimes this procedure contributed to death.

**Treatment**

Anorectal stenosis was treated by repeated
dilatation, at first under general anaesthesia. Covered anus was treated by excision of the lid of skin over the anus and any associated folds or fistulous tracts leading to the skin on the midline of the scrotum. The mucous membrane was sewn to the cut edge of the skin with interrupted stitches of fine black silk, and subsequent dilatation was carried out as necessary. Rectoperineal fistula was treated by repeated dilatation. It is my practice to treat all the other lesions by a colostomy in the neonatal period to relieve the intestinal obstruction, followed by an abdominoperineal type of pull-through operation at the age of 1 year or when the child has attained a weight of about 9 kg (20 lb). Earlier attempts at dissecting the low type of vaginal or vestibular fistula and transplanting it backwards across the perineum to the normal site of the anus, the anal dimple, resulted in such frequent breakdown of the new attachment of the fistula to the skin with recurrence of the fistula in the lower end of the vagina that this procedure has now been given up.

A transverse colostomy is preferable to a left iliac colostomy because it allows much greater freedom during the pull-through operation, at which it may often be necessary to mobilize the rectum fairly extensively before it can be brought to the perineum; this is hindered by the presence of a left iliac colostomy and it may be impossible to get sufficient viable rectum to the perineum without first taking down the left iliac colostomy. Whether the colostomy is opened at the time it is made or 24 hours later depends entirely on the degree of distension of the bowel and the abdomen and the effect this has on diaphragmatic respiration. Interference with respiratory exchange and the consequent increased tendency to respiratory acidosis is of special importance in babies with pneumonia or associated cardiovascular anomalies. Sometimes it is necessary to aspirate the gas and meconium from the colon before the colostomy can be completed.

The introduction of plastic bags, worn on a flange and held in position by adhesive and a belt has considerably reduced the complications and difficulties formerly associated with a transverse colostomy in an infant. A colostomy at this level results in a discharge of thin and often very irritant faeces compared with those which come from a left iliac colostomy, though in a child even these are more liquid and more irritant than in the adult. In the past, severe irritation of the skin and repeated minor damage to the mucous membrane exposed at the colostomy have resulted in secondary anaemia, which has required the administration of iron throughout the life of the colostomy. Moreover the irritation of the skin becomes sufficiently severe to cause the child considerable discomfort and requires the use of large quantities of ointment and dressings which must be changed every time the colostomy is cleaned. The introduction of plastic bags has almost entirely prevented both the excoriation of the skin and secondary anaemia, and it is almost easier to look after a colostomy in this way than to care for a normal bowel opening during the first year of life. The cost, however, is high, about £95 per annum for bags and flanges, but when this is set against the reduction in discomfort, excoriation of the skin, and anaemia, and the difficulties these and the care of the colostomy present to the child's mother, as well as the hours of nappy washing which she is saved, the benefit is cheap at the price. Moreover, the use of bags greatly reduces the emotional disturbance which some women find unavoidably associated with the care of a colostomy in a small baby. This may be so severe that the pull-through operation has to be undertaken earlier than would otherwise be the case because of emotional distress of the mother.

Once the colostomy has been made and is working
satisfactorily, the bowel beyond the colostomy should be washed out in order to remove all meconium and prevent the formation of hard scybalous masses which will distend the lower end of the rectum. When there is an associated rectourethral fistula, washout of the distal bowel is essential to prevent recurrent urinary infection. Even so, urinary infection is occasionally so persistent that it is necessary to undertake the pull-through operation before the child reaches the age of 1 year or 9 kg (20 lb) in weight.

In the past many surgeons have carried out the abdominoperineal operation soon after birth and some still do. It should be remembered, however, that while the mortality rate for colostomy alone in the neonatal period is 20 to 25%, which compares with the overall mortality rate of 22-4% in 1970 in this hospital for all emergency surgical admissions within 4 weeks of birth (J. A. S. Dickson, personal communication, 1971), primary abdominoperineal operation at this time probably carries a mortality rate of well over 30%. Moreover, because of the size of the tissues it is much more difficult to make a clean dissection deep in the pelvis and to be certain that the bowel has been pulled through the puborectalis sling on which in the end continence will so largely depend. Only 2 children in this series have died after 78 abdominoperineal operations at the age of 1 year; 1 on the third postoperative day of cardiac failure secondary to Fallot's tetralogy for which a Blalock operation had been done at the age of 6 months; the other 5 days after operation for a persistent circulatory failure for which no explanation, either clinical or at necropsy, could be found.

The abdominoperineal operation is unavoidable in patients with high level rectovaginal, rectourethral, or rectovesical fistulas. These cannot be satisfactorily dealt with by any form of perineal approach. Several patients have been treated who had had a perineal operation in other hospitals for such fistulas, and in the case of boys with rectourethral fistulas the damage to the puborectalis sling and the median raphe of the levator ani muscle was such that they were incontinent of both urine and faeces. In dividing the rectourethral fistulas it is important to do so flush with the posterior surface of the urethra, so that there is no remaining pouch from the urethra which could become infected and be the source of repeated bouts of urinary infection.

To emphasize the futility of the perineal operation for rectal atresia it should be said that it is often necessary, after mobilizing the rectum completely out of the pelvis, to divide the superior rectal vessels to obtain sufficient length to bring the rectum to the perineum; and sometimes it is necessary to sacrifice a third or a half of the rectum because mobilization and division of the superior rectal vessels results in impairment of the blood supply to the distal rectum. This may not, however, be much of a disadvantage because if the atretic rectum is similar to the atretic small bowel, then the dilated hypertrophied blind end has abnormal peristaltic function in any case and would be better excised. Indeed, much of the difficulty in getting the child to move the bowel satisfactorily after the abdominoperineal mobilization might be avoided if the whole of the dilated hypertrophied rectum was excised and the upper third of rectum or the distal end of the pelvic colon was brought down to the perineum instead.

The colostomy should be closed as soon as possible after a satisfactory new opening has been obtained in the perineum. It is probably better to delay closure of the colostomy, however, until this opening is in its final stage. Prolapse of mucosa at the new anus is not uncommon and should be trimmed before the colostomy is closed. Similarly the opening is sometimes too narrow and it should be widened before closing the colostomy. Closure of the colostomy may be associated with a good deal of bleeding and blood must be crossmatched and available for transfusion. It is usually better to dissect out the colostomy and excise it completely and make a formal end-to-end anastomosis of the two stumps of colon with a single layer of interrupted fine black mattress sutures than to attempt any less radical closure which is much more likely to break down.

**Functional Results**

The results of treating anorectal stenosis by dilatation are usually good (Tables III, IV). Control of faeces is ultimately normal though there may be some difficulty in the early stages; sensation of fullness of the rectum, the warning period, and the control of flatus are all normal. There is seldom any difficulty with urinary function apart from what one might expect to occur in the proportion of children who are temporarily enuretic. If medicine is needed to move the bowel, that suggests the stenosis may have recurred.

In covered anus, the control of faeces is usually good but there is commonly a leakage of faecal-stained mucus from the anal canal which slightly stains the underclothing. This usually persists for years. Sensation is usually good, the warning period is normal, and so is the control of flatus and urine.

In rectoperineal fistula the picture is much the
same as for covered anus. Most of the functions are present and normal unless there is some associated defect in the sacrum or urinary tract.

Anorectal atresia treated by the abdominoperineal pull-through operation gives variable results, but when allowance is made in a few patients for associated abnormalities such as ectopic bladder, sacral defects, or myelomeningocele, the results are surprisingly good, but in any particular individual depend on the age of the child at which the situation is assessed. Comparatively few children achieve control of urine or faeces after this operation as soon as normal children. The sensation of fullness of the bowel though present is not normal, and it takes most children some time to realize the significance of what sensation they have. Even then, and no matter how willing they may be to keep themselves clean, the warning period is often so short that they are unable always to reach the lavatory in time. In some children, however, for several years there seems to be an aversion to cleanliness, and others, preoccupied with their play or with work, do not act soon enough on what warning they have and are persistently dirty.

After the pull-through operation slight soiling of the underclothing with faecal-stained mucus is common because the superficial and subcutaneous parts of the external sphincter do not close off the new anal opening. Though these two parts of the external sphincter can be shown to function by stimulating the perineal skin at the centre of the anal dimple before the pull-through operation is done, these two sphincter rings are seldom able to close the opening after the rectum has been brought to the perineal skin.

Even in the best patients there is variation in function from time to time. These children are more liable to disturbance and malfunction as a result of injudicious consumption of fruit and vegetables, and are also more liable to severe constipation. In some, constipation is most persistent and is mainly related to the abnormal dilated and hypertrophied rectum in which peristalsis is abnormal and allows faeces to accumulate. Good function is not related particularly to the type of lesion and occurs as often with rectourethral fistula as it does with high or low rectovaginal fistula.

In the past the treatment of these anomalies of the anus and rectum has received less than its proper share of respect and care from surgeons. Only those whose bowel is not continent, or who have stained underclothing, or are the parents of a child in this state have any real idea of the social and economic burdens these entail. Successful treat-

### TABLE III

Functional Results of Treatment of Anorectal Anomalies; Control of Faeces, Flatus, and Urine in 76 Patients

<table>
<thead>
<tr>
<th>Primary Lesion</th>
<th>Faeces</th>
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<th>Urine</th>
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<tr>
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<tr>
<td>Rectal atresia lower third</td>
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<td>4</td>
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<td>—</td>
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<tr>
<td>Total (76)</td>
<td>38</td>
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### TABLE IV

Functional Results of Treatment of Anorectal Anomalies; Warning Period, Sensation or Fullness, and Need for Laxative in 76 Patients

<table>
<thead>
<tr>
<th>Primary Lesion</th>
<th>Warning Period</th>
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<td>Covered anus</td>
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<td>Rectal atresia lower third</td>
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<td>Rectal atresia middle third</td>
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<td>Total (76)</td>
<td>41</td>
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There is much controversy about the treatment of those children in whom partial absence of the sacrum suggests that innervation of the pelvic floor or the musculature may be abnormal. It is possible that the best course may be to carry out the pull-through operation in the hope that it may give a satisfactory result, but in such patients it is important first to warn the parents that control may be imperfect and function poor. This is probably better than to advise an iliac colostomy as an initial and permanent treatment.

When the pelvic floor has been damaged at either a primary perineal operation in an attempt to bring the rectum to the surface or during a pull-through operation, it is often still possible, and nearly always worthwhile, to attempt to improve the situation by a pelvic floor repair rather than to abandon all hope of improvement and advise a permanent iliac colostomy.

I am indebted to Mr. J. A. S. Dickson and Mr. A. J. Dougall for their help during the preparation of this paper; and to the Editor, Proceedings of the Royal Society of Medicine, for Fig. 3(a), (b), 6, and 7.

REFERENCES


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