CONGENITAL ABNORMALITIES OF THE ANUS AND RECTUM*

BY

MALCOLM H. GOUGH†

From The Hospital for Sick Children, Great Ormond Street, London

This paper is based on a study I have made with John Partridge, until recently Resident Assistant Surgeon at The Hospital for Sick Children, of 323 cases of anorectal abnormality seen here in the 10-year period January 1949 to December 1958.

We feel that the most important fact with regard to treatment and prognosis in these cases is whether the bowel terminates above or below the pelvic floor. We accordingly make a primary distinction between high and low abnormalities. In the low abnormalities the bowel passes through a normal puborectalis sling and pelvic floor, and continence is assured even in the presence of an abnormal external sphincter. Treatment is therefore easier and less prolonged than in the high cases where there is usually an abnormal pelvic floor and deficient internal and external sphincter mechanisms.

The low abnormalities are subdivided into:

- (1) The covered anus;
- (2) The ectopic anus;
- (3) The stenosed anus;
- (4) Anorectal membrane.

1. *The covered anus* is common in males and rare in females. The embryological explanation of this deformity is that the anus is covered by excessive posterior fusion of the genital folds. On examination there is usually a ^-shaped bar of skin at the expected site of the anus, or the midline perineal raphe is thickened and may contain a meconium-filled track leading back to the anal canal (Fig. 1). In this and succeeding Figures the important recto-pubalis muscle is diagrammatically represented. If both these distinguishing characteristics are absent diagnosis must be made from the common high anomaly.

2. *The ectopic anus* is common in females and rare in males and results from defective migration of the embryonic bowel. According to the site at

which the bowel terminates below the pelvic floor this group is divided into the vaginal, the vestibular, the vulval and the perineal ectopic anus (Figs. 2, 3 and 4).

3. *The stenosed anus* may be easily visible but reduced in calibre or it may be of pin-hole size (the microscopic anus described by Denis Browne), and here I would like to say how many of our ideas concerning these low abnormalities have resulted from the work of Denis Browne (1951) and Douglas Stephens (1953a, b). Embryologically this abnormality is caused by excessive fusion of the anal tubercles.

We have also included in this category the less usual type where the stenosis occurs within the anal canal at the level of the anal valves. This is due to incomplete breakdown of the anal membrane. Should this membrane remain imperforate our fourth division results.

4. *Anal membrane* (Fig. 5) is a rare anomaly. It has been confused with the covered anus when the covering is very thin, and sometimes with Hirschsprung's disease when withdrawal of the examining finger is followed by a gush of meconium.

The high anomalies are subdivided into:

- (1) Anorectal agenesis, with or without fistula;
- (2) Rectal atresia;
- (3) Cloaca;
- (4) Multiple abnormalities, e.g. ectopia cloacae.

1. *Anorectal agenesis* is more common in the male than the female and in over 75% of our male cases there has been an associated fistula into the prostatic urethra (Fig. 6). We have not encountered a rectovesical fistula.

2. *Rectal atresia* (Fig. 7) has previously also been known as the Ladd type IV anomaly or imperforate rectum; the anal canal ends blindly below the pelvic floor and the upper rectal pouch ends at or above this level.

* A paper read at a meeting of the British Association of Paediatric Surgeons held in London in July 1960.

† Present address: c/o Peter Bent Brigham Hospital, 721 Huntington Avenue, Boston, Mass., U.S.A.

FIG. 1.

FIG. 4.

FIG. 2.

FIG. 5.

FIG. 3.

FIG. 6.

FIG. 7.

We have been accused of bandying words in applying the term agenesia to the first abnormality and atresia to this. Following the definition of these words in the Shorter Oxford Dictionary (3rd edition, reprinted in 1955) we have used agenesia to indicate imperfect development of the functional unit (as it is used in renal agenesia) and atresia to indicate occlusion (as in duodenal atresia). We have avoided using the term 'imperforate anus',

which at times has been used to describe the whole group of anomalies and at others to describe certain types within this group. We have also reserved the term 'fistula' for use in the description of high anomalies, using 'anus' to describe the ectopic or abnormal termination of the bowel in the low abnormalities. We hope that this will stimulate the idea that a low ectopic opening can be encouraged to function as an anus, whereas this is not the case with the fistula in the high cases.

3. *Cloaca* (Figs. 8 and 9) is used to describe the anomaly in females when the urinary, genital and alimentary tracts all open into a common wide cavity (Gough, 1959).

4. Our fourth heading, *Multiple abnormalities*, includes ectopia cloacae (vesico-intestinal fissure) and other severe malformations.

Incidence

Of the total number of 323 cases, 60%, or 192, were low anomalies and 40%, or 131, were high; 57% of the children were males.

Table 1 shows the numbers of each type of the low anomaly. The common male anomaly is the covered anus, and the common female anomalies are the perineal, vulval and vestibular ectopic anuses.

Stenosed anus is more common in the male (29 of the 38 cases); the figures stress again the relative infrequency of the anal membrane.

FIG. 8.

FIG. 9.

TABLE 1
INCIDENCE OF LOW ANOMALIES

Low Anomaly	No. of Cases
Covered anus	53
Ectopic anus	98
perineal and vulval	57
vestibular	33
vaginal	8
Stenosed anus	38
Anal membrane	3
Total	192

TABLE 2
INCIDENCE OF HIGH ANOMALIES

High Anomaly	No. of Cases
Anorectal agenesis	114
male	103
female	11
Rectal atresia	5
Cloaca	9
Multiple abnormalities, e.g. ectopia cloacae	3
Total	131

Table 2 shows the incidence of the high anomalies and among these the great majority were anorectal agenesis in the male.

Our findings also confirm the high incidence of associated congenital abnormalities; 64% of the high and 25% of the low anomalies had other, often multiple abnormalities. The commonest associations were of skeletal, cardiac and genito-urinary malformations and we now consider that all these children should have an intravenous pyelogram performed. In the absence of urinary infection we prefer to delay this investigation until 1 year of age; the radiographs are more satisfactory then and the radiation risk is diminished. We have regarded the presence of a fistula in the high cases as part of the anomaly rather than an associated anomaly.

Treatment and Results

Low Abnormalities. The covered anus has been treated by the cutback operation described by Browne (1951) or if there has been no track, the anus has been uncovered by excision of a circle of skin. The perineal and vulval ectopic anuses have been similarly treated by cutback; the vestibular ectopic anus has been treated by dilation only, by cutback or by transplantation. We now favour transplantation. The vaginal ectopic anus has been either transplanted or pulled through by the

abdomino-perineal method. Treatment of the stenosed anus has been dilation, and of the anal membrane, rupture and dilatation. Some children were referred having been treated initially elsewhere; many of these were sent for advice about colonic inertia (which used to be known as acquired mega-colon), which had developed secondarily to stenosis of the anus, and long periods of bowel re-education have been required.

The results of treatment are shown in Table 3. Under the heading of the anomaly the total number of cases is repeated, followed by the number of children who have died; the third column represents children who cannot be traced, or for whom there is inadequate follow-up, and the fourth column therefore gives the number available for study; this is followed by the division of results into good, fair or poor. A *good* result indicates normal bowel control without regular laxatives; *fair* indicates the need for continual supervision to avoid recurrent constipation or anal stenosis; these children usually are being maintained on laxatives and suppositories, and *poor* indicates that a state of incontinence or severe colonic inertia still exists. In the final column we have inserted the average length of follow-up; this, with the fair and poor results gives some idea of the anomalies which have proved the most troublesome to manage. There is an overwhelming majority (86.5%) of good

TABLE 3
RESULTS OF TREATMENT OF LOW ANOMALIES

Type	Total	Deaths	Not Followed Up	No. Seen	Good	Fair	Poor	Average Follow-up (yrs)
Covered anus	53	3	8	42	37	5	—	3.1
Ectopic anus:								
Perineal and vulval	57	2	13	42	39	1	2	3.6
Vestibular	33	3	3	27	20	6	1	5.6
Vaginal	8			8	7	1		4.25
Stenosed anus	38	3	7	28	24	4		3.0
Anal membrane	3			3	3			2.4
	192	11	31	150	130	17	3	

TABLE 4
RESULTS: HIGH ABNORMALITIES

Type	Total	No. Seen	Good	Fair	Poor
Pull-through at birth	24	19	7	9	3
Colostomy at birth, pull-through later	29	29	7	14	8
Colostomy at birth, perineal dissection later	8	8	4	2	2
Perineal operations at birth and later	7	6	2	2	2
Colostomy at birth, sacral operation later	1	1	1		
	69	63	21	27	15

results. Several of these 'good' cases have spent a long time in the fair category before bowel habit has been restored to normal, and this stresses the most important factor in treatment. These children *must* be watched carefully postoperatively. Dilatation of the new anus must be adequate but not excessive, or scarring and restenosis occurs. If constipation develops it must be treated energetically to avoid colonic inertia. Of the 20 'fair' or 'poor' results, 10 can be directly attributed to the anomaly not being recognized until the age of 6 months or later. The higher ectopic openings, i.e. the vaginal and vestibular are less stenosed at birth and so may remain unrecognized, presenting later with overflow incontinence or severe constipation. Another fact that emerges is that the stenosed anus (in our classification) in the normal site, tends not to restenose after adequate dilatation whereas the cutback anus often does.

High Anomalies. Rectal atresia has been treated by pulling the blind proximal bowel through the distal pouch; the treatment of cloaca has varied according to the anatomical abnormality, and there have been no survivors amongst the three cases of ectopia cloacae. The treatment of the cases of anorectal agenesis has been more difficult to analyse. 38 children were referred here having had a previous operation, the majority (27) with a simple colostomy (quite often this was an iliac colostomy which interfered with the later pull-through operation). All but two of these 38 required further operations and in 17 cases this was an abdomino-perineal and in 14 a perineal pull-through; two were pulled down by a sacral approach, two were eventually left with a permanent colostomy and one required only refashioning of the anus. In the neonatal period 76 children were seen and the two main forms of treatment were (1) immediate abdomino-perineal pull-through and division of fistula if present

TABLE 5
MORTALITY RATES

Anomalies	No. of Cases	Deaths
Low:		
Covered anus	53	3
Ectopic anus	98	5
Stenosed anus	38	3
Anal membrane	—	—
High:		
Anorectal agenesis	114	36*
Rectal atresia	5	2
Cloaca	9	5
Multiple abnormalities	3	3

* Corrected mortality anorectal agenesis alone is 46%. (See text).

(33 cases), and (2) simple colostomy with or without division of a fistula (36 cases). This second routine was usually adopted in children in whom there were other abnormalities or who were unfit for a major operation. Of the total of 114 cases of anorectal agenesis 69 survivors have had operations providing an anus at the normal site; the results of these operations have been assessed in Table 4.

A result has been regarded as *good* when the child is continent of faeces and urine, has his bowels open spontaneously and regularly and appears to have some degree of rectal sensation in that a full lower bowel is recognized, and soiling is only an occasional accident. A *fair* result is one in which the child is continent of faeces except when these are unusually loose, although soiling may commonly occur, and in whom laxatives or suppositories are often required. A *poor* result signifies virtually a perineal colostomy.

There is, at first glance, little to choose between the results of the two principal forms of treatment. In the initial abdomino-perineal pull-through, however, there are still five children too young to assess. In the absence of other complications this is the operation which we now favour. If this is not possible we prefer initially to perform a colostomy and divide a fistula if present. The perineal and sacral operations have been abandoned.

There remain the mortality rates which are stated in Table 5. The overall mortality rate for the low anomalies is 5.7% and for the high 35.9%. Of rather more interest, however, is the mortality rate amongst the 76 cases of anorectal agenesis first seen and treated at this hospital (the remaining 38 of the total of 114 cases had been initially treated elsewhere). There were 35 deaths amongst these 76 infants, giving a mortality rate of 46%. This formidable figure is largely attributable to the high incidence of severe associated anomalies which were themselves the cause of death: no less than 24 of these 35 children were so affected.

Summary

The division of these anomalies into high and low types is of assistance both in treatment and prognosis, and is justified by the figures we have given and which bear repeating. Results of treatment show 86% good results in the low cases compared with 30% in the high; 25% of the low cases have associated anomalies, compared with 62% of the high, and the latter are usually of a more severe nature. The mortality rate for the low cases is 5.7% and for the high cases 35.9%, and this figure if corrected for anorectal agenesis seen and treated in the neonatal period rises to 46%.

This is the largest series of these cases yet recorded. Well-documented series have been reported from the United States, but unfortunately comparison with our results is impossible owing to differences

of classification, e.g. the covered anus in our classification is a low anomaly, in Ladd's classification it would be Type 3 and be grouped with the high anomaly we call anorectal agenesis. There is certainly a place for standardization of nomenclature and we hope that this system will recommend itself.

We would like to thank the Surgeons of The Hospital for Sick Children, Great Ormond Street, for permission to report their cases. My own thanks I gratefully offer to Mr. H. H. Nixon, F.R.C.S. for helpful criticism and advice, and to Mr. Geoffrey Lythe for the illustrations and diagrams.

REFERENCES

- Browne, Denis (1951). Some congenital deformities of the rectum, anus, vagina and urethra. *Ann. roy. Coll. Surg. Engl.*, 8, 173.
Gough, M. H. (1959). Anorectal agenesis with persistence of cloaca. *Proc. roy. Soc. Med.*, 52, 886.
Stephens, F. D. (1953a). Congenital imperforate rectum, recto-urethral and recto-vaginal fistulae. *Aust. N.Z. J. Surg.*, 22, 161.
— (1953b). Malformations of the anus. *Ibid.*, 23, 9.