Anorectal Agenesis with Persistence of Cloaca

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The term cloaca conjures up memories of embryological studies or ideas of gross irremediable congenital deformities in the newborn. The latter, as I hope to show, is far from the truth; the embryological memories will have to be supplemented by a brief reiteration of the development of the hind end of the embryo.

The development of the tail fold leads both to the formation of the hind-gut and the cavity which receives the hind-gut, the developing mesonephric ducts and the allantois; in other words the cloaca. Embryologically the term cloaca refers to the cavity receiving alimentary, urinary and genital ducts. This is also sometimes called the endodermal cloaca for its epithelium is derived from that germinal layer, as opposed to the so-called ectodermal cloaca which develops as a depression on the ventral or ectodermal aspect of the cloacal membrane. Clinically one maintains the same criteria of definition, but the cavity itself does not necessarily represent exactly the same cavity as the embryological cloaca. It is, in fact, usually only a small part of it. It is important to stress this difference between the embryological definition of the term cloaca and the sense in which it is to be used as a clinical description. There is considerable variation of opinion regarding the nomenclature of congenital anorectal deformities, and the term cloaca has been used to describe more than one type of abnormality, as will be mentioned later.

To revert to the embryology, the cloaca now becomes divided by the growth, in the angle between the allantois and the hind-gut, of the urorectal septum: this extends caudally to divide the primitive rectum from the primitive urogenital sinus. The remaining small communication between the rectum and urogenital sinus, before the septum reaches and fuses with the cloacal membrane, is called the cloacal duct. The primitive urogenital sinus thus formed may be divided into the vesico-urethral segment above the openings of the mesonephric ducts, and the definitive urogenital sinus below these openings. The former is destined to become the bladder and posterior urethra, and the latter, according to sex, most of the distal urethra in the male or the vestibule in the female. The mesonephric ducts form the vasa in the male and virtually disappear in the female. At this time also, the tail gut—the most caudal part of the cloaca—regresses and disappears. The cloacal membrane then breaks down so that free communication exists between the ectodermal and endodermal cloaca.

The remaining aspect of embryology relevant to this discussion is the formation of the female genital tract. The müllerian ducts develop from the cœlomic epithelium just lateral to the mesonephric ducts, and caudally they cross in front of the mesonephric ducts and grow to meet and fuse with their fellow of the opposite side to form the uterovaginal canal. The caudal end of this canal meets the posterior wall of the urogenital sinus and produces there an elevation—the müllerian tubercle. Proliferation of cells from the region of the müllerian tubercle results in the growth of a solid cord of cells, the vaginal cord, which increases the distance between the uterovaginal lumen and the urogenital sinus. There is still some controversy about the origin of the lower vagina, the generally held view being that it is formed entirely by the recanalization of the vaginal cord. However, Koff (1933) postulates that the lower vagina is formed from the sino-vaginal bulbs which develop from the urogenital sinus, and McKelvey and Baxter (1935) pointed out that abnormalities found supported the view that the vagina was of composite origin. This is borne out by examination of some of the cases under discussion. The question of the origin of the vagina also affects that of the origin of the hymen. If the whole of the vagina develops from the vaginal cord, then the hymen represents the remains of the cloacal membrane; if partly from the sino-vaginal bulbs it represents the junction between these bulbs and the vaginal cord.

In describing anorectal abnormalities we meet the difficulties in nomenclature which I mentioned. Swenson (1958) groups all these abnormalities under the heading "imperforate anus". This is perhaps not wholly suitable although the word anus may be used in its purely embryological sense as the termination of the bowel, or in its usual clinical sense as the termination of the bowel complete with sphincter mechanism. I propose to call these cases rectal or anal agenesis with fistula formation, as appropriate. Nixon (1959) has classified the case of high rectal agenesis with rectovaginal, rectovesical or rectourethral fistula formation as a cloaca, using as a synonym "imperforate rectum". This nomenclature has advantages in that, used in conjunction with the terms vulval ectopic and perineal ectopic anus (Browne, 1955) it serves to divide these lesser abnormalities from the more serious ones where the bowel terminates *above* the pelvic floor. In the former, some form of minor surgery such as a perineal cutback will be

adequate for treatment, whilst in the latter some form of major surgery will be needed. This conception of a "cloaca" is not new and has been used by several writers in the past, including Pennock and Stark (1926) and Stone (1936).

The following cases may show that the term cloaca should be reserved for more serious abnormalities occurring from some arrest in development at a fairly early stage. One congenital abnormality may be accompanied by another, and the occurrence of an earlier fault in the embryo might be expected to be more frequently associated with other, often severe, abnormalities which may radically alter any plan of treatment. This is, in fact, what is found to occur.

The following examples of what I term cloaca have been collected from the records of children treated at the Hospital for Sick Children, Great Ormond Street, over the past ten years. A few of these seem to be little more than examples of the more usual anorectal anomalies with a somewhat abnormally situated urethra: in the examples a gradual regression towards the primitive form may be seen, some of them approximating to the normal conditions found in marsupials, amphibians and reptiles, as one descends the evolutionary scale.

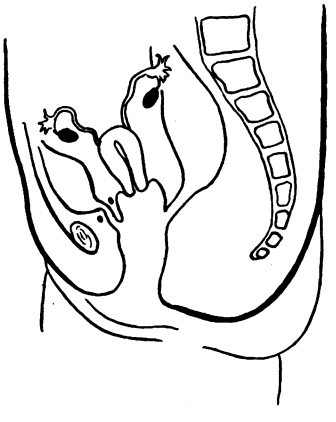

FIG. 1.

One example of a typical cloaca is diagrammatically represented in Fig. 1. This girl had a laparotomy, division of what I must call the rectocloacal fistula and a pull-through of the rectum. She is well apart from considerable residual trouble with colonic inertia, and it is interesting to note that she is fully continent of urine. By no means all of these children have a urinary sphincter, as is seen in Fig. 2 which shows a considerably more primitive state of affairs. Here there was a vesicocloacal orifice which

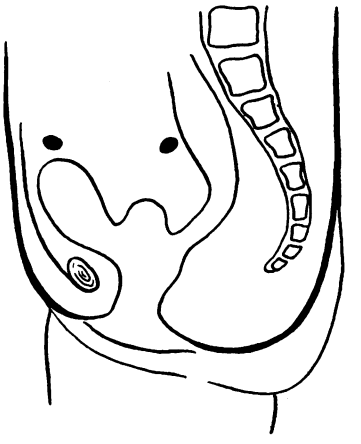

FIG. 2.

accepted two fingers and a rectocloacal orifice which accepted one finger. Neither orifice had any evidence of a sphincter and there was absence of the uterus and both fallopian tubes, although both ovaries were present. This girl had a defunctioning colostomy, isolation of the distal sigmoid loop and, later, a bilateral ureteric transplant into this isolated loop. She is alive and well in spite of the obvious disadvantages of a colostomy and a urinary diversion.

Further abnormalities may complicate the picture and Figs. 3–6 illustrate four such examples:

Fig. 3 shows two commonly associated anomalies, a double uterus and also a mucucolpos. The former occurs in the majority of the 18 cases of cloaca I have collected and is, in itself, of little importance with regard to treatment. The mucocolpos may be very large and may present as an abdominal tumour. It may be mistaken

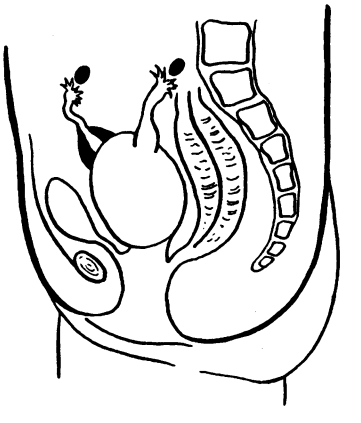

FIG. 3.

for an enlarged bladder and it will be difficult to disprove this because of the abnormal position of the urethra. An awareness of the occurrence of mucocolpos will be sufficient to prevent its inadvertent removal, along with a compressed and adherent bifid uterus, tubes and ovaries, as a dermoid or retroperitoneal cyst. This child died of peritonitis and septicæmia following drainage of the mucocolpos—which turned out to have been infected—and pull-through of the rectum (which incidentally was duplicated, this being found at autopsy).

Fig. 4.—This girl again had a double uterus, but the main point of interest is the presence of one

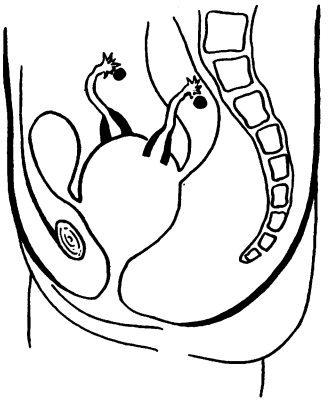

FIG. 4.

small external orifice which was taken at initial examination to be the external urinary meatus. At operation perineal dissection was performed and a pull-through completed of what appeared to be the rectum. Post-operatively she developed intestinal obstruction and laparotomy revealed that what had been pulled through was the cloaca itself, and following this the rectocloacal orifice had become obstructed. A transverse colostomy was then performed and she made a good recovery. She was readmitted at the age of 1 year for pull-through. This was successfully carried out, but she unfortunately died of gas gangrene following later closure of the colostomy.

Fig. 5. again shows a more primitive form. Here there is a double uterus and vagina, the bowel opens into the cloaca via the septum between the two halves of the vagina, and the bladder opens widely and directly into the anterior cloaca. There is a cystic reduplication of the terminal bowel, which alone represents large bowel. This child died in a cyanotic attack two days post-operatively. She had gross renal abnormalities and probably congenital heart disease also.

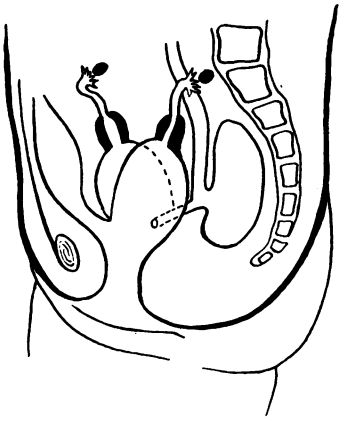

FIG. 5.

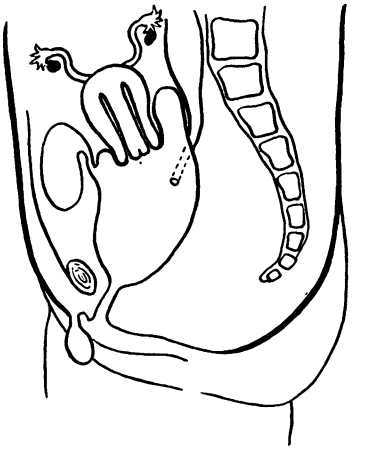

FIG. 6.

Fig. 6.—This child has a cloaca with an apparent pseudohermaphroditism.

An easily recognizable deformity seen in 2 cases has been the presence of a tube-like fold of skin representing the phallus. Birnbaum (1912) referred to the fact that complete absence of the genitalia occurs only in conjunction with severe abnormalities such as cloacal formation or sireniform monsters. I have not seen any reference to this tube-like phallus, but its presence should suggest the presence of a cloaca. In one child it overlay a single orifice, and in the other it provided an outlet for the cloacal orifice.

All the examples of true cloaca which I have shown are females; in these children the existence of the genital cavity makes the decision as to

whether or not a cloaca is present more easy. Whether anorectal agenesis with recto-urethral or rectovesical fistula in a male should be termed a cloaca is, from the practical point of view, unimportant. This problem, strictly speaking, rests on the site of the uterus masculinus and openings of the vasa.

Several points arise from these 18 cases—six, or one-third, are alive; 11 have died; and one I cannot trace. I have arbitrarily divided the deaths into those which were probably inevitable due to other serious abnormalities and those which were possibly avoidable taking our present surgical armamentarium into account.

Of the 11 deaths, 6 were probably inevitable and 5 might have lived apart from the onset of some form of sepsis. 5 children died in cyanotic attacks in cardiac failure or bronchopneumonia, 4 died of septicæmia, peritonitis or gastroenteritis, and 2 died directly of gross renal anomalies.

The associated abnormalities most frequently found were: (1) Œsophageal atresia, (2) upper renal tract abnormalities, (3) congenital heart disease and vascular anomalies, (4) anomalies of vertebral, especially sacral, development. It is probably always worth while passing a nasogastric tube on these infants; in 1 of these 18 an œsophageal atresia was missed until after the operation for the cloaca. Should the infant survive it is wise to perform an intravenous pyelogram to exclude upper renal tract abnormalities which may require attention, although in the absence of urinary infection this investigation is probably better done, from the point of view of good pictures and decreasing the radiation risk, at about 1 year of age. The incidence of such abnormalities in straightforward cases of anorectal agenesis is surprisingly high. It is even higher in these more unusual children.

The first aim of management is to discover, as far as possible, exactly what the anatomy is. From this series an associated abdominal tumour is more likely to be a mucocolpos than an enlarged bladder. The usual X-rays to show the level of the rectal gas bubble will help and, in addition, will show any bony deformity of the sacrum. An examination under anæsthesia of a single orifice or cavity may help, although the presence of meconium and mucus may make interpretation difficult. It may be possible to catheterize the abnormally placed urethral orifice and perform cystography. A mucocolpos should be drained. Whether an immediate abdominoperineal operation for rectal pull-through should be performed depends on individual circumstances: in the absence of other abnormalities this is the ideal treatment.

In some children, such as my second example, there can be no chance of urinary continence; in others it will have to be left to time to establish whether control exists. The possibility that an operation for urinary diversion on to the abdominal wall may be needed at some later time should be borne in mind. One of the serious complications of the abdominoperineal pull-through operation is damage to the pelvic splanchnic nerve plexuses supplying the bladder and its sphincters. These plexuses lie lateral to the normal rectum and probably lie close to each other in the mid-line in cases of anorectal agenesis. They are, therefore, especially at risk during the perineal dissection in the male; in the female the presence of the mid-line vagina may protect them. Whilst the actual operation is easier in the neonate, due to the relative shallowness of the pelvis, it may be that further deforming abnormalities such as a mucocolpos should be reason for performing an initial transverse colostomy.

It may be much easier to plan a surgical approach to the problem of urinary incontinence and anorectal agenesis when the child is a little older. Should continence exist it may be easier to visualize and avoid the important splanchnic nerves. Whenever laparotomy is performed a full note should be made of the state of the internal genitalia. Questions by the patient in later years regarding getting married and having children will be difficult to answer unless operation notes are complete.

I hope I have been able to show that the condition of persistence of the cloaca is not such a hopeless one as might be thought. Reports in the literature are few; it is possible that in the past some of these cases were classified as "fœtal ascites"; I have certainly found one such in the literature of fifty years ago. With the aids to surgery available to-day there seems no reason why the lives of the majority of these children should not be saved.

Acknowledgments.—I would like to thank the surgeons at Great Ormond Street for permission to refer to cases admitted under their care, and especially Mr. D. Innes Williams who first drew my attention to this interesting condition.

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