Congenital Malformations of the Rectum, Anus and Genito-Urinary Tracts

BY

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WITH

CONTRIBUTIONS IN PAEDIATRIC PATHOLOGY, SURGERY AND SURGICAL ANATOMY BY RESEARCH FELLOWS OF THE DEPARTMENT

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FOREWORD BY

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FOREWORD

THE deformities of the lower end of the digestive canal have had curiously litt e attention given to them in medical writings and education; a marked contrast to the immense literature concerned with those of the upper end, such as harelip and cleft palate. The reasons for this comparative neglect are many. These deformities are not very common; they are out of sight though far from out of the mind of the sufferer, and they come under the universal taboo on the mention of the " shameful parts " of the human body in polite society. The medical profession itself is far from free from this split-mindedness—I had an interesting example of this recently when a line drawing of the technique of washing out the rectum aroused in a surgical audience the type of laughter which greets the breaking of a taboo.

Another factor in this neglect is the present division of the study of the human body into the anatomists who study its structure and the physiologists who study its working. What the opinion of the great John Hunter would have been on this disastrous dichotomy is a solemn thought; but its influence can be seen in the way that both sides leave the study of sphincters in neglect. Yet sphincters are one of the essential mechanisms of the body, and this book is mainly concerned with them, their imperfections or their absence.

The result of this lack of knowledge and teaching is a considerable number of quite unnecessary bad results from surgery, involving years of suffering or a completely ruined life, from a condition which the right treatment at the right time could have cured completely. Anyone who doubts the inadequacy and vagueness of the usual teaching on " imperforate anus " should examine a few works on general surgery, from which he will gain the impression that the treatment is to dissect up into the perineum in the hope of finding a manageable end of the lower bowel. The success of this proceeding in undiagnosed covered and microscopic anuses accounts for its use in other cases in which it destroys utterly the muscles of the floor of the pelvis on which the only hope of continence depends.

The remedy for this state of affairs is the analysis and classification of the different varieties of these malformations, and I consider this to be one of the most important accomplishments of paediatric surgery. It is because of the part I took in this study with Douglas Stephens that it gives me particular satisfaction to introduce this work in which he has taken the analysis further on, in both theory and practice.

He arrived at the Hospital for Sick Children in Great Ormond Street from Boston, where he had had the good fortune to see Orvar Swenson's work on Hirschsprung's disease at a time when it was hardly known. To myself and my fellow surgeons, David Waterston and Harold Nixon, and to our pathologist Martin Bodian, this solution of the ancient problem of an intractable malformation was a wonderful stimulus. For several years from that date we worked together on the subject of this book, not as a formal team, but without any of the jealousy and secretiveness which bedevil so much surgical work. There were many arguments and disputes—in fact this book will produce a few between me and its author—but there were no quarrels and no standing upon our respective dignities. It was a fruitful and exciting spell

of work of a kind which occurs all too seldom in the present conditions of surgery. The list of contributors to this book, and the unity of approach in all of them, shows that a similar enviable spirit has been present during its genesis.

A last question is raised by a book as full of wisdom and experience as this one—what is the mechanism by which the benefits it could bring to many sufferers may actually reach them, and how long will this process take? Surely here is a problem to be tackled by the profession as a whole. The fact that not all problems have been solved should not be taken as an excuse for delaying recognition of solutions which are theoretically sound and clinically satisfactory.

DENIS BROWNE.

LONDON, 1963.

PREFACE

IN this volume are presented a series of Essays by members of the Staff of the Department of Surgical Research, established in the Royal Children's Hospital, Melbourne, as an activity within the Royal Children's Hospital Research Foundation. In their preparation earlier papers, which have been published in the *Australian and New Zealand Journal of Surgery*, the *Medical Journal of Australia*, and in the *Australasian Annals of Medicine*, have been freely drawn upon, and in some instances considerably modified, by permission of the Royal Australasian College of Surgeons and the Australasian Medical Publishing Company. Chapters IX, XIII and XVI are included by courtesy of the publishers of the *Journal of Urology*, the Williams and Wilkins Company, Baltimore 2, Maryland, U.S.A. As originator of the book and principal contributor, Mr F. Douglas Stephens records his appreciation of the courtesies extended him by the Editors of the journals named, particularly with respect to the reproduction of the numerous illustrations.

The first section of the book is concerned with studies of Malformations of the Rectum and Anus by F. Douglas Stephens, who focuses attention on the cloaca as the embryological rendezvous of the alimentary, urinary and genital systems, and a frequent participant in developmental defects. Malformations may involve one, two or all three systems, and the complications are many and baffling. The author's objective has been to clarify the confusion which has hitherto existed regarding the embryological deviations upon which rectal and anal malformations ensue, and to apply ordered and systematized knowledge of the problems they present to the care of patients.

In the second section F. Douglas Stephens deals with abnormalities of the urinary tract, intensive study of the enigmatic nature of which has enabled him to formulate some clearly defined principles of management. Much attention is given to mega-ureter, the phenomenon of vesico-ureteric reflux, ureterocele and urethral obstruction; opinions are recorded regarding the vexed question of bladder-neck obstruction.

Section III is comprised of a group of studies contributed by members of the Staff of the Department of Surgical Research, as detailed in the Table of Contents. The book is designed for post-graduate study; it makes no claim to encyclopaedic authority, but is presented as an account of systematic investigation of important problems in paediatric proctology, urology, pathology, radiology and anatomy during the past twelve years.

F. DOUGLAS STEPHENS.

MELBOURNE, AUSTRALIA, 1963.

ACKNOWLEDGMENTS

WORK during the first three years was carried out at the Hospital for Sick Children, Great Ormond Street, London, where Mr F. Douglas Stephens profited much from stimulating contacts with Sir Denis Browne, Mr T. T. Higgins, O.B.E., Dr Martin Bodian, and many other members of the staff. On his return to Australia Mr Stephens was supported by the Royal Children's Hospital in the continuance of the work initiated in London, and in the development of the Department of Surgical Research; this Department is now an integral unit under the Royal Children's Hospital Research Foundation. At all times the author has derived much assistance from the cordial co-operation of his teachers and colleagues.

Substantial assistance from the Nuffield Foundation consisted in the grant to Mr Stephens of an Overseas Travelling Fellowship to the United Kingdom immediately after the war, and later, on his return to Australia, a Dominion Nuffield Fellowship. More recently the Trustees of the Nuffield Foundation have contributed generously to the cost of the illustrations.

Onerous secretarial duties have been capably discharged by Miss S. Hitchcock, Mrs S. Smith and by Mrs J. Williams (a member of the Volunteer Service of this Hospital). An invaluable contribution has been that of Mr Eric Thake of the Department of Visual Aids of the University of Melbourne, whose artistic talent has been effectively applied in many of the illustrations. One diagram, Figure 137, was drawn by Mr A. Strohlein, also a member of the same department. Mr C. Murphy and Miss Omond of the Department of Medical Illustration of this Hospital have worked with kindly co-operation and great patience to produce the numerous photographs which appear in this book.

Throughout the book radiographic methods of investigation figure prominently, and have involved many calls on the services of Dr H. Hiller, Radiologist to the Hospital, and his staff, whose sustained co-operation enabled the maintenance of a close liaison between the Department of Surgical Research and the Department of Radiology; incursions into the Department of Pathology have been welcomed and assisted by Dr John Perry and his successor as Director of Pathology, Dr Alan Williams. Much pathological material relevant to the work was obtained from the Royal Women's Hospital by courtesy of Dr H. F. Bettinger.

In the past twelve years, specimens of a large collection of proctological and genito-urinary deformities and diseases have been accumulated, thanks to the watchfulness and faithful efforts of the mortician, Mr Albert Winther, in notifying myself or members of the research team of their occurrence. These cherished examples provided the anatomical basis of many of the studies recorded in this book.

Many of the original observations stem from thousands of serial sections of material carefully and patiently prepared by three expert histological technicians, namely Messrs Ian Hine, Cyril Bird and John Komissarov.

Until his death in 1961 the late Sir Hugh Devine evinced a stimulating interest in the work, lending valuable assistance in the preparation of the earlier articles.

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MELBOURNE, AUSTRALIA, 1963.

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Part I

MALFORMATIONS OF THE RECTUM AND ANUS

CHAPTER I

CONGENITAL DEFORMITIES OF THE RECTUM

F. DOUGLAS STEPHENS

A CLEAR understanding of the deviations from the normal course of embryological events, and their ultimate effect on the structure and relations of the pelvic viscera, is essential to the proper appreciation of the distinction drawn by Wood Jones (1904) between anal and rectal deformities.

Subdivisions of the rectum and anal canal

ANATOMICAL.—The rectum commences at the level of the third sacral vertebra and extends distally to the apex of the prostate (Gray, 1938). The anal canal commences at the apex of the prostate, or anorectal ring (Milligan and Morgan, 1934) and terminates at the orifice of the anus. The anal canal is that part which is surrounded by the sphincters.

EMBRYOLOGICAL.—The rectum and anal canal develop from the hind gut, the cloaca, and the proctodeal pit, the proctodeal segment being delimited by the line of the anal valves. The rectum above this level, to its origin at the third sacral vertebra, is derived from the cloaca and hind gut. The embryological dividing line between the cloacal rectum and that formed directly from the hind gut is not certain. Wood Jones (1911) described the hind gut contribution as ending at the level of the peritoneal reflexion, or Houston's third fold, where anatomical changes in the bowel suggest a different embryological origin. The teaching of Wood Jones that the mid-portion of the rectum is the product of a post-allantoic outgrowth from the hind gut now appears as minority opinion in view of the adherence of most embryologists to the theory of cranio-caudal subdivision of the cloaca, later to be enunciated. Nevertheless, it remains true that many congenital rectal deformities, particularly the common recto-urethral fistula, occur in situations which conform to Wood Jones's embryological rectal boundaries.

Throughout these essays, therefore, the nomenclature of abnormalities adheres to this major embryological grouping, incorporating the prefixes recto- or ano- (or the words rectum or anus). These terms are strictly definitive.

The anal group of malformations is referable to developmental aberrations affecting the proctodeal pit and membrane, the perineum and genital folds. The deformities comprising this group are the more common, the more easily treated, and provide the greater number of satisfactory surgical cures; they are described in Chapter II.

The rectal group consists of those abnormalities in which the termination of the hind gut is found at the level of the verumontanum or higher in the male, and in the female lies at a corresponding level in the pelvis, perhaps opening into the cloacal canal or the posterior wall of the vagina. The term "imperforate rectum" or rectal agenesis covers this group of visceral deformities, individual components of which may or may not exhibit a fistulous communication with the urinary tract or vagina; their discussion involves of necessity consideration of associated defects of the spine, sphincter and levator ani muscles.

The schemata in Figs. 1 and 27 correlate the usual malformations with their underlying defects in development, and the stages at which such lapses occur in males and females respectively.

Embryological data

To clarify the several steps in the development of the pelvic viscera, and the manner in which imperfect achievement of one or more stages results in familiar anal and rectal deformities, it is necessary to recall the conditions which exist in the 4-mm. embryo, when the cloaca, or slightly dilated caudal portion of the hind gut acts as a reservoir into which open three endodermal tubes, namely, the hind gut, the allantois and the tail gut (Hamilton, Boyd and Mossman, 1946).

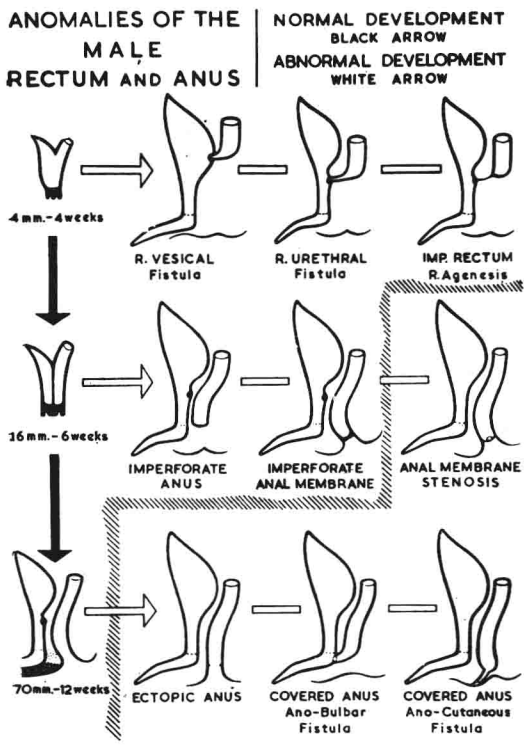

FIG. 1

Embryological Classification.

Upper Row: Rectal deformities arising from failure of subdivision of cloaca.
Middle Row: Defects in the formation of the anal pit.
Lower Row: Mixed malformations derived from concurrent faulty development of the perineum (dotted area) causing irregular migration of the anus, and of genital folds (cross hatching) causing anterior projection of anal fistula.
Hatched line—Diagnostic inversion radiographs are required to define groups shown above the line. Diagnosis of deformities shown below the line is made by local inspection.

The 5-mm. stage witnesses, in the form of a groove which appears in the angle between the allantois and the hind gut, the initiation of the growth of the urorectal septum, a structure of prime importance in ensuring the orderly development and ultimate integrity of the rectum, bladder and urethra. The urorectal septum grows caudally in a coronal plane and divides the cloaca, incompletely at first, into a smaller dorsal part—the primitive rectum—and a larger ventral part—the primitive urogenital sinus. As the urorectal septum progresses caudally the separation of the rectum from the primitive urogenital sinus is gradually advanced until, at the 16-mm. stage, the urorectal septum reaches and fuses with the endoderm of the cloacal membrane, and the separation of the rectal part of the cloaca from the urogenital sinus portion is complete (Fig. 2).

Such is the conception most favoured in textbooks of embryology of the manner in which the subdivision of the cloaca in the embryo is brought about, but another school of opinion, to which the author adheres, (Stephens, 1953) maintains that the urorectal septum effects the cleavage of the cloaca only as far as Muller's hillock and the pubococcygeal line. In the interval between this level and the cloacal membrane, a second mechanism, in the form of an inward surge of mesenchyme from the sides of the embryo towards the midline, operates to complete the cleavage of the urinary tract from the gut. The lateral mesenchyme indents the sides of the cloacal cavity in the manner described by Keibel and Mall (1912), and illustrated by Bradley Patten (1947) (Fig. 2).

If downgrowth of the urorectal septum were the only mechanism involved in cloacal division, it would be expected that the commonest form of fistula in the male would be

located at the level of the triangular ligament, and the site of the cloacal duct, a structure which represents the last part of the cloaca to be sealed off from the urogenital sinus, and which occasionally persists. The extreme rarity of such a fistula at this level, and in the membranous urethra, and the frequency of a fistula in the plane of the pubococcygeal line, suggest that the subdivision of the cloaca below this line is effected by a different process from that which defines and separates the gut from the urinary tract cranial to this point. The frequency with which a recto-urethral fistula occurs at the level of the verumontanum, and the rarity of fistulae below this level are explained by the complete failure of the supplementary mechanism. The cloaca remains undivided below the level of lowest descent of the urorectal septum and, together with the hind gut communication, remains narrow in calibre relative to the surrounding structures.

As has been emphasised, failure of cloacal subdivision by the lateral ingrowths accounts for the common abnormality of recto-urinary fistula in the male at the level of the pubococcygeal line (Fig. 3). The urethra below the fistula is actually the undivided cloaca. In the female this fistula, being incorporated in the actively proliferating and migrating sinovaginal bulbs, is often carried in the posterior wall of the vagina to a lower level (Fig. 3).

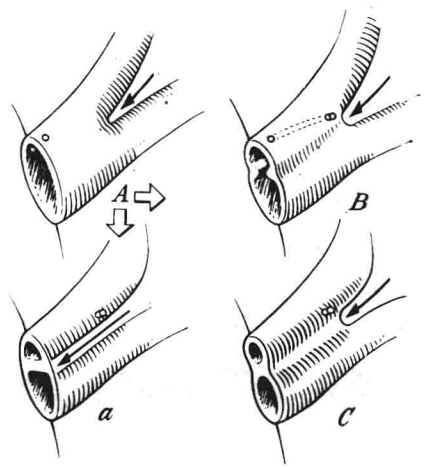

FIG. 2

Methods of subdivision of the Cloaca.

A–a: Subdivision by downgrowth of the anorectal septum to meet the cloacal membrane.

A–B–C: Subdivision by downgrowth of urorectal septum as far as Muller's hillock (verumontanum), and by lateral ingrowths of mesenchyme caudal to this level. (The dotted lines in *B* represent the line of retraction of the orifices of the Wolffian ducts from sites near the cloacal membrane at the earliest stage, to the later attained level of the verumontanum.)

DEVELOPMENT IN THE FEMALE.—The situation created by imperfect division of the cloaca is complicated in the female by the interposition of the Mullerian duct system (Fig. 3). The sexual duct system in the male, by what Patten has happily termed "developmental opportunism," is appropriated from the mesonephric ducts, from which are derived the vas deferens and epididymis of each side.

In the female, special ducts, heralds of the Fallopian tubes, uterus and vagina, termed paramesonephric, but better known as Mullerian ducts, are initiated in the 10-mm. embryo by an invagination of the coelomic epithelium into the mesenchyme on each side. Such invaginations occur laterally to the cranial extremities of the mesonephric ducts, and the paramesonephric or Mullerian ducts, formed by the caudal burrowing and later canalization of the invaginations, follow a course parallel with the mesonephric ducts as far as the caudal extremity of the mesonephros; at this point the Mullerian ducts cross in front of the mesonephros, fuse in the midline and continue their caudal progress until they establish contact with the dorsal aspect of the urogenital sinus. At this point the vaginal cord is formed by the fusion of the Mullerian ducts but lacks a lumen; it is eventually channelled and takes shape as the uterovaginal canal.

While the caudal tip of the uterovaginal canal is as yet solid, cells growing back from the epithelial lining of the urogenital sinus merge with it to form a projection into the lumen of the urogenital sinus, an elevation known as the Mullerian tubercle (or hillock). The uterovaginal canal and cells derived from its lower end give rise ultimately to the

vagina, but whether to the whole or only portion is a matter in dispute. Those that support the opinion that the vagina is of composite origin believe that approximately the caudal one-fifth of the vagina is derived from the sinovaginal bulbs; these formations appear at the 63-mm. stage of development, and are described as bilateral posterior endodermal

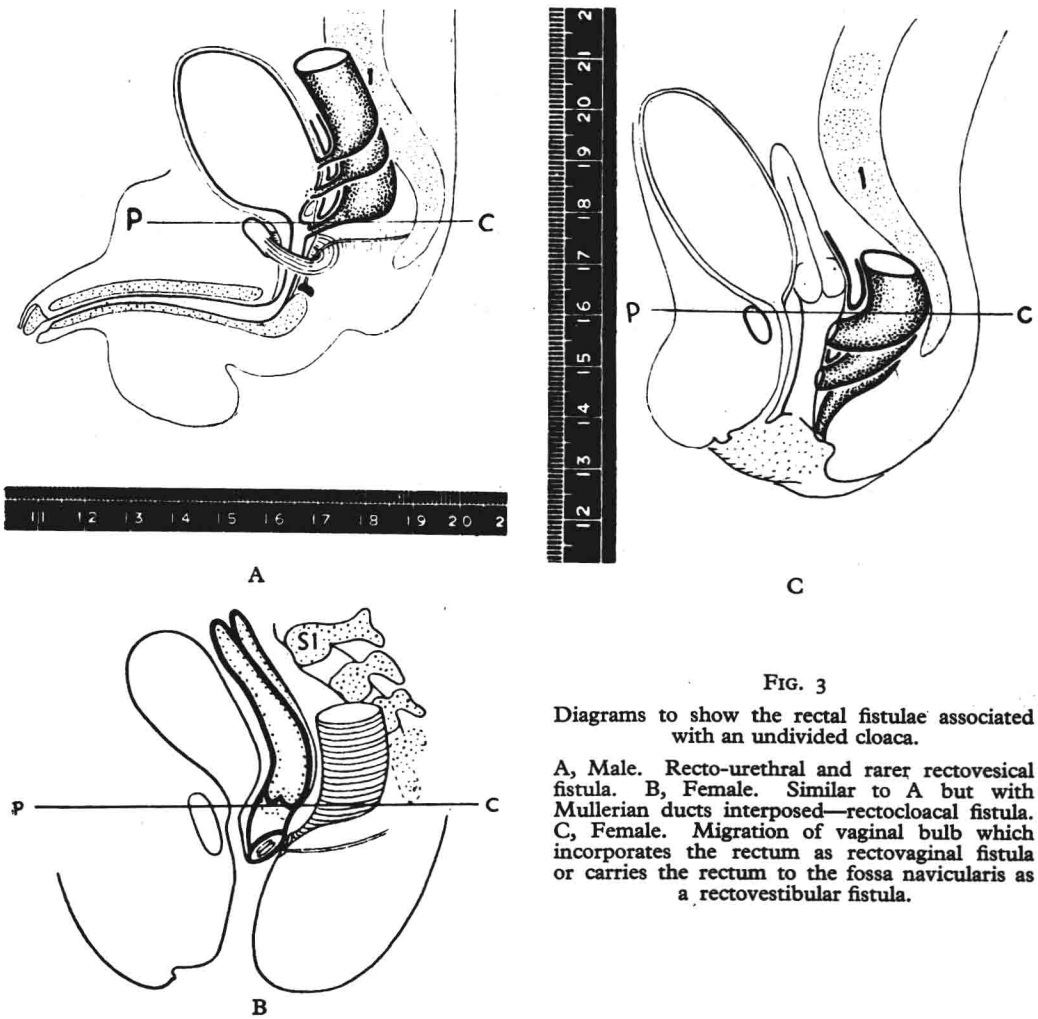

FIG. 3

Diagrams to show the rectal fistulae associated with an undivided cloaca.

A, Male. Recto-urethral and rarer rectovesical fistula. B, Female. Similar to A but with Mullerian ducts interposed—rectocloacal fistula. C, Female. Migration of vaginal bulb which incorporates the rectum as rectovaginal fistula or carries the rectum to the fossa navicularis as a rectovestibular fistula.

evaginations from the urogenital sinus, situated close to the attachments of the mesonephric ducts.

By cellular proliferation these endodermal evaginations, or incredibly minute diverticula, become solid, and as they increase in size obliterate the Mullerian tubercle. At the same time that part of the urogenital sinus immediately cranial to the sinovaginal bulbs becomes narrowed and elongated to form the female urethra, and the sinovaginal bulbs, exhibiting exceptional activity in cell multiplication, soon reach their maximum development, move caudally and fuse with the vaginal cord. Canalization is effected by extension of the uterine

canal from above, and by the breaking down of the epithelium of the now fused sinovaginal bulbs from below; when this phase is completed the embryo has attained a length of 162 mm.

RECTAL DEFORMITIES

The ensuing discussion is based on study of the pelves of sixteen male and thirteen female infants who exhibited rectal deformities such as result from failure of subdivision of the cloaca. From this cause the termination of that portion of the rectum contributed by the hind gut lies at or about the level of the third fold of Houston, attached by fibrous tissue, in which perhaps is a fistulous track, to the undivided cloaca or to the vagina.

In the male the rectum may end blindly, remaining attached to the cloaca by fibrous tissue in close proximity to the prostate (Fig. 3). The term imperforate rectum or rectal agenesis has been applied to this particular anatomical defect. Communication with the urinary system may persist as a fine fistula opening in immediate proximity to the normal orifices of the ejaculatory ducts and the utriculus masculinus—the so-called recto-urethral fistula (Fig. 3). More rarely, the fistula and Wolffian ducts together open into the urinary system at a higher level, with the production of the rarer forms of recto-urethral and rectovesical fistulae (Fig. 3).

In the female the rectal attachment or fistula to the urinary tract, at the level of Muller's tubercle, becomes incorporated later in the actively proliferating and migrating epithelial vaginal bulbs (Fig. 3). In changing their location from the original level of Muller's tubercle below the bladder neck to the vestibule, these bulbs carry the rectum distally a varying distance. Sometimes the fistula is carried before the bulbs to the fossa navicularis in the vestibule, in which event it is herein described as a rectovestibular fistula; sometimes, however, it is incorporated in the bulbs, when it may form a fistula at any level into the posterior wall of the vagina (Fig. 3). The orifice of a fistula between the rectum and posterior vaginal wall is usually of wide calibre, whereas that of the fistula into the fossa navicularis is usually small. These vaginal deficiencies are readily recognized by the naked eye, though imperforate conditions of the rectum and the deeply situated fistulae require the assistance of radiology and the examining telescope.

SURGICAL ANATOMY AND PATHOLOGY

Essential to the accurate appraisal of these deformities and to the design of efficient surgical technique for their correction, is a precise knowledge of the anatomy of the levator ani muscle, and the deviations from normal development which it may exhibit. Another necessary requisite is familiarity with the pubococcygeal line, which as an indicator of an embryological plane is an important level of reference. Needless to state, the sphincters of the rectum are matters of particular concern in devising operative technique. These prime considerations will now be discussed in detail in the order in which they have been mentioned.

The normal levator ani muscle

From the posterior surface of the body of the pubis and the white line as far back as the ischial spine, fibres of the levator ani arise and sweep obliquely backwards medially and caudally in three sections (Fig. 4, A and B).

One thin section pursues an oblique course across the membranous urethra and meets