THE CONGENITAL SHORT OESOPHAGUS

BY

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There are two closely related developmental abnormalities which are rare predisposing causes
of the common digestion oesophagitis. These are (a) congenital shortness of the squamous
oesophagus, and (b) congenital enlargement of the oesophageal hiatus of the diaphragm. This
paper presents an account of the pathology of 25 cases.

It is now generally agreed that many cases of short oesophagus with hiatal hernia were
previously described as congenital on insufficient evidence; by far the commonest cause is acquired
chronic cicatrizing reflux or digestion oesophagitis. There is, however, a risk that the pendulum may
swing too far the other way and the existence of the congenital lesion be generally denied or
forgotten.

Findlay and Brown Kelly (1931) collected seven post-mortem congenital cases from the literature,
the earliest by Richard Bright in 1836. In two of the cases (Bund's and Huffman's) other congenital
anomalies were present. Hume (1932) described a case and stressed the accompanying enlargement
of the hiatus.

In 1950 Barrett published three cases which are included in the present series; he suggested
that certain chronic oesophageal ulcers reported previously by Tileston (1906), Stewart and Hart-
fall (1929), and Lyall (1937) were "gastric" ulcers in congenital partial thoracic stomachs. He also
raised the at first sight startling question, "What is an oesophagus?" Barrett claimed that
any part of the alimentary tract above the dia-
phragm lined with gastric mucosa continuous with
the stomach proper must be regarded as partial
thoracic stomach whatever the gross morphology.
If not covered by peritoneum, it was an extension
of the bare area normally seen above the peri-
toneal reflections at the cardiac end. Allison
(1951) contested this view; if in gullet form, the
anomaly was best described as oesophagus with
atypical (gastric) lining, as gross morphology
could not be ignored. There is the point here,
however, that the normal gastric cardia, although
very short, is essentially a tube.

Both these interpretations were developed in
later papers (Allison and Johnstone, 1953; Bar-
rett, 1954). In addition, Allison and Johnstone
recorded islands of squamous epithelium in the
gastric-lined segment (see also Morson and Belcher, 1952), and oesophageal glands in the
submucosa. I made similar observations inde-
pendently (Peters, 1952). In the differential diag-
nosis, Allison and Johnstone suggested that heal-
ing of oesophagitis in a continuing acid medium
might be by gastric type epithelium rather than
by squamous epithelium. There is clear histo-
logical evidence, however, that squamous re-
geneneration from residual islets occurs readily
even in the distal oesophagus (Peters, 1955).

Barrett (1957) has recently modified his view. He regards the lower atypical segment as repre-
senting an embryonic maturation failure and sug-
gests the lesion be called "the lower oesophagus
lined by columnar epithelium."

The congenital short oesophagus is necessarily
associated with a large hiatus unless the gastric-
lined part is wholly tubular, but the congenitally
large hiatus can occur alone and appears to have
been first reported by Harrington in 1940, who
also described the embryological basis (vide infra).

EMBRYOLOGY

The first indication of the developing stomach
is very early in intra-uterine life. In the
fourth week (2–3 mm. embryo) a fusiform dilata-
tion of the entodermal foregut develops in the
cervical region, and this, the primitive stomach, is
separated from the future pharynx by a constric-
tion in the tube, the oesophageal anlage. At this
stage the foregut, slung on a dorsal mesentery, is
separated from the pericardium by the nearly
vertical septum transversum, into the caudal part
of which the liver bud from the lower end of the
foregut pushes cranially. During the sixth week
(5–10 mm. stage) the growth and development of

Thorax (1958), 13, 1.
the oesophagus are rapid. As the bulk of the heart, lungs, and liver increases and the neck differentiates, the entodermal oesophageal tube elongates and concentric condensation of mesoderm around it foreshadows the development of submucosa and muscularis, the outer longitudinal muscle layer being indicated after the inner circular and the muscularis mucosae later still (Hamilton, Boyd, and Mossman, 1952). The superficial cardiac glands of the oesophagus begin to develop at four months or earlier, but the deep glands not until much later (Arey, 1934). The entodermal epithelium of the oesophagus is composed of small columnar cells, ciliated at one stage, which are later replaced by cells of squamous type. Lewis (1912) pointed out that the characteristic shape of the different segments of the alimentary tract is determined by differential growth of the entoderm epithelium, the outer mesodermal layers being modelled round and conforming to the inner tube, which is itself eventually represented by the definitive mucosa.

During the stage of rapid caudal migration the stomach starts to differentiate from a simple fusiform swelling into a structure with defined curvatures and recognizable pars cardiaca and tubular pars pylorica (Lewis, 1912). At the same time the septum transversum descends rapidly and rotates so that it lies nearly horizontally, thus separating the ventral parts of the developing thoracic and abdominal cavities. The pleuroperitoneal ridges descend with the septum transversum, and both for a short time outstrip, so to speak, the elongation of the oesophagus, so that the stomach is temporarily cranial to the developing diaphragm. During the seventh week (11–18 mm.) the continued elongation of the oesophagus is such that the stomach rapidly overtakes and passes the septum transversum and comes to lie in the future abdominal cavity, and during the eighth week the membranous diaphragm is completed round the lower end of the oesophagus. Muscularization of the diaphragm closely follows the growth in membrane. Differentiation in the developing oesophagus is seen in Figs. 1–3, to obtain which two well-preserved embryos of the approximate stated ages were embedded in wax, in the left lateral position, and cut through in serial sections 5 μ thick. Fig. 4 shows a hernial sac formed by a greatly stretched membranous dome in a case where muscularization of the diaphragm had failed to take place.

If the elongation of the oesophagus is prematurely arrested, part or the whole of the stomach remains in the thorax and the diaphragm forms round it. If, as Harrington (1940) pointed out, the elongation of the oesophagus is merely delayed, even for a short time, the diaphragm again forms round the stomach and the resulting large hiatus persists, even though by subsequent growth of the

Fig. 4.—Failure of muscularization of the dome of the diaphragm. A. Oesophagus. B. Right crus. C. Aorta. D. Inferior vena cava. E. Hernial sac formed by membranous left dome.
Fig. 1.—Primitive oesophagus showing early condensation of mesoderm (arrows) round entodermal foregut. Section taken from a 11–12 mm. human embryo (approximately 40 days' fertilization time). Haematoxylin and eosin. x 22. A. Oesophagus. B. Septum transversum. C. Pharynx. D. Respiratory tubule.

Fig. 2.—Detail of Fig. 1. Haematoxylin and eosin x 61. A. Oesophagus. B. Septum transversum.

Fig. 3.—Further differentiation of oesophagus. Muscularization of membranous diaphragm (arrow). Section taken from a 30 mm. human embryo (approximately 56 days' fertilization time). Similar field and same magnification as Fig. 1, but about 16 days older. A. Oesophagus. B. Diaphragm. C. Liver. D. Adrenal. E. Heart.
oesophagus the stomach later becomes a wholly abdominal organ. Slightly asynchronous development may thus be responsible for some cases of large hiatus with sliding gastric hernia in postnatal life.

It is thus seen that the most important stages in the highly complex evolution of the oesophagus, stomach, and diaphragm occur from the fourth to the eighth weeks of development, and the malformations to be discussed in this paper presumably originate thus early in embryonic life.

**Present Material**

The following investigations are based on 25 cases collected at necropsy (21 of congenital short oesophagus, four of congenital large hiatus alone). The main findings are shown in Tables I and II.

**MORBID ANATOMY.—**In the congenital short oesophagus the external appearances depend on the form of the gastric-lined lower part and the type and degree of the digestion oesophagitis which is usually present in the short squamal part. If severe, the latter may show externally as an indurated stricture, sometimes as high as the bifurcation of the trachea, but in such heavily scarred cases the evidence for an underlying congenital abnormality needs to be very strong. The lower gastric portion varies in gross morphology: it may be (a) tubular, (b) conical or inverted funnel-shaped, (c) both successively, or (d) resembling normal stomach. These forms are usually fairly clear cut, but sometimes intermediate nondescript specimens are seen. A characteristic of all the forms, naturally least in the tubular type, is a soft bulkiness when inspected and felt from the outside; this may also be present in some cases of severe oesophagitis, because of inflammatory swelling, but only in very minor

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### Table I

**CASE ANALYSIS**

<table>
<thead>
<tr>
<th>Case</th>
<th>Age</th>
<th>Sex</th>
<th>Congenital Short Oesophagus</th>
<th>Morphology of Gastric Part</th>
<th>Anomalous Microscopic Organization in Gastric Part</th>
<th>Congenital Large Hiatus Only</th>
<th>Ulceration</th>
<th>Diam. of Hiatus (cm)</th>
<th>Primary Carcinoma</th>
<th>Other Major Malformations</th>
<th>Severe Mental Disorder</th>
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<tr>
<td><strong>Average</strong></td>
<td><strong>62.4 years</strong></td>
<td><strong>M:</strong> 12M, F:** 12F**</td>
<td><strong>Recorded total 18</strong></td>
<td><strong>Tubular conical 6 (55.5%) and conical 6 (55.5%)</strong></td>
<td><strong>Recorded total 19</strong></td>
<td><strong>12 (62% of recorded total)</strong></td>
<td><strong>4</strong></td>
<td><strong>20</strong></td>
<td><strong>9</strong></td>
<td><strong>50%</strong></td>
<td><strong>3 (14.3%) excluding 1 possible</strong></td>
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</table>
degree. In the more differentiated types distinction from the far more common acquired fixed hiatal hernia with cicatrising oesophagitis (see Table III) may be difficult, but in the latter condition the intrathoracic gastric cone is usually much smaller. The visceral attachment of the elastic hiatal ligament may be well below the level of the squamo-glandular junction, and is then good evidence of a congenital anomaly. Similarly the reflections of the peritoneum do not usually follow the thoracic course of the abnormal stomach.

Internally the combined viscus shows mucous membrane of two types. Proximally, for a length varying in the adult from 6 to 18 cm., is the grey-white squamous epithelium, usually with general or focal leukoplakia and irregular superficial ulceration towards the lower limit. The amount of fibrosis is very variable. Below the squamous length is the atypical gastric-lined part, the junctional zone sometimes jumbled and not clear cut (Fig. 8). The quite distinct brownish velvety mucosa is continuous with that of the main part of the stomach. As would be expected, there is some correlation between gross morphology and type of mucosa. Tubular forms and the proximal tubular parts of more differentiated forms have a smooth lining, microscopically cardiac in type; in the junctional zone proper, however, an artefact to be borne in mind is post-mortem desquamation with exposure of normal oesophageal cardiac glands (Peters, 1955). In the expanded more differentiated (conical) parts the mucosa is rugose and microscopically may contain oxyntic cells (sometimes abundantly) and occasionally the gastric feature of two crosswise layers in the muscularis mucosae (Fig. 15).

Ulcers in the gastric part are sometimes shallow, necrotic and haemorrhagic (Figs. 5 and 12) and sometimes of classical excavated and fibrous form (Figs. 9 and 14). Metaplasia to epithelium of intestinal type may occur round the ulcers (Morton, personal communication).

The microscopic organization may be atypical in two particulars (vide supra). Oesophageal mucous glands with typical periductular lymphoid aggregates may be seen in the submucosa below frank gastric epithelium (Fig. 17), and islets of well-differentiated squamous epithelium may be seen embedded in or focally replacing the gastric epithelium (Fig. 16). Both these features, in my belief, are of positive significance in the diagnosis. The squamous islets may be just visible macro-

<table>
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<tr>
<th>Kidney</th>
<th>Skeletal</th>
<th>Cardiovascular</th>
<th>Gastro-intestinal</th>
<th>Miscellaneous</th>
<th>Doubtful or Doubtfully Significant</th>
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<tr>
<td>Hypoplasia</td>
<td>Dwarfism with gross asymmetry</td>
<td>Fallop's tetralogy</td>
<td>Ectopic gastric mucus in proximal oesophagus</td>
<td>Mesenteric cyst</td>
<td>Angioma of liver</td>
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<tr>
<td>Double pelvis</td>
<td>Rudimentary digits and meta-carpals; multiple symmetrical hyperostoses</td>
<td></td>
<td>Ectopic pancreas in duodenum</td>
<td>Congenital cataracts</td>
<td>Cysts of liver</td>
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<tr>
<td>Polyostic</td>
<td>Premature union of cranial sutures</td>
<td></td>
<td></td>
<td>Angiomatous hamartoma of brain</td>
<td>Diverticula of small intestine (including one Meckel)</td>
</tr>
<tr>
<td></td>
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<td></td>
<td></td>
<td>Malformation of liver (both lobes equal in size and shape)</td>
<td>Congenital peritoneal bands</td>
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<td></td>
<td></td>
<td></td>
<td>Endocrine hypoplasia</td>
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<td></td>
<td></td>
<td>Abnormal lobation of lungs</td>
</tr>
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<td></td>
<td></td>
<td>Diverticula of urinary bladder without urinary obstruction</td>
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<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>&quot; Lipoma &quot; of central tendon of diaphragm</td>
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### Table III

**DIFFERENTIATION OF CONGENITAL AND ACQUIRED SHORT OESOPHAGUS**

<table>
<thead>
<tr>
<th>Congenital Short Squamous Oesophagus (All Morphological Types)</th>
<th>Acquired Short Oesophagus with Fixed (Gastric) Hiatal Hernia</th>
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<tbody>
<tr>
<td>1. Gastric cone in chest usually high and bulky (least in simple tubular forms)</td>
<td>Gastric cone in chest small (up to 3 cm. on average)</td>
</tr>
<tr>
<td>2. Associated digestion oesophagitis in squamous part often not of excessively fibrous type, i.e., insufficient fibrosis to fix an acquired gastric thoracic pouch</td>
<td>Sufficient fibrosis present for fixation of hiatal hernia with permanent shortening (usually with stricture)</td>
</tr>
<tr>
<td>3. Gastric cone may not be covered with peritoneum or only in part</td>
<td>Gastric cone has normal peritoneal covering</td>
</tr>
<tr>
<td>4. Diaphragmatico-oesophageal elastic ligament may be attached well below the squamo-glandular junction</td>
<td>Diaphragmatico-oesophageal ligament attached roughly at or above the squamo-glandular junction</td>
</tr>
<tr>
<td>5. Hiatus often very large and circular</td>
<td>Hiatus usually only slightly enlarged and elliptical shape usually preserved</td>
</tr>
<tr>
<td>6. Anomalous microscopic structure may be demonstrable</td>
<td>No anomalies of microscopic structure. Acquired morbid changes only</td>
</tr>
<tr>
<td>(a) May be deep oesophageal mucous glands in submucosa below gastric epithelium</td>
<td></td>
</tr>
<tr>
<td>(b) May be squamous islets embedded in gastric epithelium</td>
<td></td>
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<tr>
<td>(c) Glandular epithelium (above the insertion of the elastic ligament) usually cardiac in type, sometimes fundal with oxyntic cells</td>
<td></td>
</tr>
<tr>
<td>7. Commonly associated with major malformations elsewhere</td>
<td>No special association with malformation</td>
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</table>
FIGS. 5–9.—Congenital short squamous oesophagus: gastric part in tubular form.

**FIG. 5.**—A. Squamous-lined proximal part. B. Chronic digestion ulceration of lower end of squamous part. C. Gastric-lined distal part. D. Shallow chronic "gastric" ulcer. Specimen reproduced by kind permission of Mr. Barrett and the editor and publishers of the *British Journal of Surgery.*

**FIG. 6.**—A. Squamous-lined part, ulcerated at lower end. B. Gastric-lined part. C. Diaphragm.

**FIG. 7.**—A. Squamous-lined part with ulceration and leukoplakia in the lower end. B. Gastric-lined part. C. Diaphragm.

**FIG. 8.**—Snakeskin leukoplakia above jumbled epithelia at junctional zone (detail of Fig. 7). A. Squamous plaques. B. Gastric mucosa. C. Ulcerated areas.

**FIG. 9.**—A. Squamous-lined part, lower half ulcerated. B. Gastric-lined part. C. Chronic "gastric" ulcer (Barrett's ulcer). D. Diaphragm.
Fig. 10.—Congenital short squamous oesophagus. Tubular and conical gastric part, with primary adenocarcinoma. A. Squamous part. B. Carcinoma. C. Non-ulcerated gastric mucosa, rugose in expanded part. D. Diaphragm. E. Approximate insertion of stretched elastic diaphragmatico-cesophageal ligament. Arrow, compare Fig. 11.

Fig. 11.—Typical cardiac (junctional) arrangement, in this case high in the thorax. Haematoxylin and eosin × 54. A. Squamous epithelium. B. Cardiac glands. C. Oesophageal submucosal gland. D. Carcinoma. (Compare arrow in Fig. 10.)
scopically (Fig. 14) or occasionally be in larger plaques (Figs. 8 and 14), and they not infrequently occur in the margins of ulcers (Fig. 13). This raises the possibility that such ulcers may in some cases actually form through these squamous foci; in any fully established ulcer it is of course impossible to have certain knowledge of the type of epithelium which has disappeared; this can only be inferred. As the epithelium proper to the gullet is squamous, neither "metaplasia" nor "ectopic islet" appear appropriate terms for such foci, which are best regarded as areas of normally differentiated epithelium surrounded by aberrant mucosa.

The main muscular coats of the anomalous viscus consist of normal and well-defined inner circular and outer longitudinal layers and do not contribute to the differential diagnosis. In two cases of this series, both of simple tubular form, there appeared to be general hypoplasia of the whole wall. In the adventitia, the insertion of the elastic hiatal ligament far below the squamo-glandular junction may be confirmed microscopically.

FIG. 12.—Congenital short squamous oesophagus, showing tubular and conical gastric part. (The whole specimen was supra-diaphragmatic.) A. Lower end of squamous part. B. Gastric mucosa, rugose in expanded part. C. Haemorrhagic ulcers. Specimen reproduced by kind permission of Mr. Barrett and the editor and publishers of the British Journal of Surgery.

FIG. 13.—Margin of ulcer with squamous islet (arrow) included in gastric mucosa (detail of Fig. 12). (See also Figs. 15 and 16.) Haematoxylin and eosin. × 60.

Points in the pathological differentiation of the congenital and acquired short oesophagus are listed for convenience in Table III.

As to the congenital large hiatus without shortness of the squamous oesophagus, this may be difficult to distinguish from the common atrophic or toxic lax hiatus; therefore the four present examples are advanced as probables rather than certainties. The criteria used were the size and shape of the opening, supported indirectly in two cases by the occurrence of major malformations elsewhere.

The average diameter of the hiatus (recorded in three of four cases) was 4 cm., compared with the average of 4.5 cm. in the six adult congenital short oesophagus cases in which it was recorded. In one specimen (5 cm. diameter) the hiatal organization was dissected fully and the opening was found to lie as usual in the right crus. The large hiatus appeared circular and not elliptical and was spanned by a hypertrophied elastic ligament (Figs. 20 and 21). This circular shape was thought to be important as well as the size, because in the common acquired hiatal deficiencies the normal elliptical shape seems to be surprisingly long retained. In all four cases chronic digestion oesophagitis was present.
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Fig. 14.—Distal part of congenital short squamous oesophagus.
A. Jumbled epithelia at junctional zone. B. Rugose gastric mucosa. C. Large chronic "gastric" ulcer (Barrett's ulcer). D. Minute squamous islets.

Fig. 15.—Margin of chronic "gastric" ulcer. Note two-layer muscularis mucosae. A. Gastric mucosa. B. Oesophageal glands in submucosa. C. Fibrous ulcer floor. (Inset see Figs. 16 and 17.) Haematoxylin and eosin x 4.

Fig. 16.—Squamous islet in gastric mucosa. Haematoxylin and eosin. x 51.

Fig. 17.—Oesophageal mucons gland in submucosa. Haematoxylin and eosin. x 61.

Figs. 15, 16, and 17.—Microscopic details of Fig. 14.
Fig. 18.—Congenital short squamous oesophagus. Gastric part in differentiated stomach form. A. Squamous oesophagus. B. Gastric fundus. C. Diaphragm. D. Pancreas.

Fig. 19.—Congenital enlargement of hiatus and chronic ulcerated oesophagitis with slight shortening. A. Diaphragm. B. High lesser curve chronic gastric ulcer.

Fig. 20

Fig. 21

Figs. 20 and 21.—Hypertrophied elastic diaphragmatico-oesophageal ligament spanning a large hiatus. The sections were taken from the specimen illustrated in Fig. 19. Haematoxylin and van Gieson stain and elastic. Fig. 20 × 1] approximately and Fig. 21 ×4 approximately. A. Ulcerated oesophagus. B. Stomach. C. Elastic ligament. D. Diaphragm.
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Discussion

Although tables should speak for themselves, a few points of interest may be abstracted. The cases are too few to have much statistical weight.

(1) Nearly all the specimens showed chronic ulcerative oesophagitis in the squamous part. In the short oesophagus series, there was ulceration in the gastric part (Barrett’s ulcer) in about half.

(2) There was primary adenocarcinoma of the gastric part (Fig. 10) in 14.3% of the short oesophagus cases. From the latest available Registrar-General’s statistical review, approximately 2.8% of all deaths in England and Wales in 1954 were due to gastric neoplasm. Smithers (1950) has recently published a detailed analysis of 31 cases of adenocarcinoma of the oesophagus. The condition has also been discussed by Barrett (1957).

(3) In about half the cases (45.8%) other major congenital malformations were present; in further 20.8% there were doubtfully significant or doubtful congenital abnormalities (total 66.6%). A curiosity is two cases of short oesophagus having very similar skeletal abnormalities, including rudimentary digits and symmetrical hyperostoses.

(4) In six cases (27.3%) gross mental disorder had been present in life. In one (Case 14) it could probably be accounted for by age and anaemia; of the remaining five, three were lifelong mental defectives and two were chronic psychotics (one of them schizophrenic since puberty).

General Incidence.—The present cases occurred in a series of approximately 25,000 necropsies, giving the following figures for overall incidence:

- Congenital short oesophagus ... ... 0.84%  
- Congenital enlargement of hiatus only ... 0.016%  
- Combined total of about 1 in 1,000 necropsies

Age Incidence.—The average age at death was about 62 years so that, although early death may be caused in individual cases, e.g., Case 2, on the whole the malformations and resultant lesions do not appear seriously to shorten life.

Sex Incidence.—No significant preponderance of either sex was found (13 female, 12 male).

Pathogenesis and Terminology.—It is relevant at this point to reiterate the view of Lewis (1912), who pointed out that it is the differential growth in the embryo of the entodermal foregut (ultimately represented by the definitive mucosa) which governs the shape of the final viscus. In the congenital short oesophagus it seems that where the atypical gastric epithelium is of fundal type the gross viscus resembles stomach in shape, whereas if it is of cardiac type the form tends to be tubular, i.e., as a much drawn-out gastric cardia or junctional zone. Both of these forms may show the oesophageal feature of mucous glands below the muscularis mucosae, that is to say, the wall of the anomalous organ, whatever its shape or mucosa, may retain a microscopic oesophageal characteristic. It is an atypical structure, neither oesophagus nor stomach (see also Barrett, 1957), or alternatively both. There would seem, however, no fundamental objection to the old-established term “congenital short oesophagus,” especially if qualified by the word “squamous.”

The evidence, such as it is, suggests that the essential abnormality is a relative failure of differentiation of the oesophageal part of the primitive entodermal foregut; the squamous anlage appears to be inadequate, and this, associated with insufficient “caudal migration” in the early embryo, is eventually revealed as shortness of the proper epithelium in the final organ. This view corresponds in some degree with the conception of a graded failure of normal transformation or maturation of the embryonic oesophageal epithelium advanced by Smithers (1956) and Barrett (1957).

Summary

The congenital malformations directly related to digestion oesophagitis are: (a) Congenital shortness of the squamous oesophagus with or without a large hiatus; (b) enlargement of the hiatus alone.

The incidence, morbid anatomy, pathological diagnosis, and pathogenesis of these two conditions are discussed on the basis of 25 cases.

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