ACHALASIA OF THE CARDIA AND MEGA-ESOPHAGUS

Report of Five Representative Cases

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ACHALASIA, defined as failure or inability to relax, when applied to the esophageal cardia most accurately designates a familiar clinical and pathologic disease entity. The term mega-esophagus refers to the presence or development of a dilated esophagus in association with achalasia of the cardia, and must be so restricted. Less acceptable in the light of present knowledge is the older term cardiospasm, which has been defined as a functional type of obstruction of the esophagus at or near the esophageal hiatus, usually associated with dilatation of the thoracic esophagus.1 Effler and Rogers2 differentiate cardiospasm from mega-esophagus, but the two conditions may be essentially the same, differing only in degree and duration. We have seen patients with achalasia without esophageal dilatation who later developed mega-esophagus.

The purpose of this paper is to review briefly the etiology, clinical features, and treatment of achalasia and mega-esophagus, and to present the reports of five cases that demonstrate the diagnostic findings, the treatment, and some of the complications of the condition.

Etiology

The etiology of achalasia is not known. It rarely is associated with gross organic disease of the esophagus. Most proposed explanations of the etiology have focused on the nervous control over the lower esophagus. Hurst3 in 1930 reported that some patients with achalasia had degenerative changes in Auerbach's plexus in the lower esophagus. He observed that the abdominal esophagus failed to relax in front of the contractional wave. Knight4a in 1934 experimentally reproduced this condition in cats by high bilateral vagotomy. He also was able to prevent or to relieve the condition experimentally by sectioning the sympathetic nerve supply to the cardiac end of the stomach and lower esophagus. He4b later employed left gastric sympathectomy in the treatment of some patients with achalasia and reported good results in a few. Mitchell5 and others

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have noted that sympathetic denervation of the cardia is an extensive and impractical procedure in human beings.

Etzel\(^6\) in 1942 reported that mega-esophagus frequently was associated with vitamin B\(_1\) deficiency. He found that other conditions, including megacolon, megaureter, pyloric achalasia, altered intracardiac conduction, low basal metabolic rate, and polyneuritis, frequently coexisted with mega-esophagus. He attributed these changes to degeneration in Auerbach's plexus involving the digestive and urinary tracts. He believed that this degeneration resulted from deficiency in vitamin B\(_1\) because that deficiency was present in 626 patients having mega-esophagus included in a survey.

Mega-esophagus and megacolon are similar in that each involves dilatation and atony of the respective organ proximally, and may involve degeneration of Auerbach's plexus. A true cardiospasm rarely is present in patients having mega-esophagus, and hypertrophy of the muscle at the cardiac sphincter, such as that which occurs in congenital hypertrophic pyloric stenosis, seldom is seen. The term achalasia describes the deranged neuromuscular mechanism in mega-esophagus better than does cardiospasm. The pathologic changes probably are not restricted to the distal esophagus, since extremely ineffective and tertiary peristalses and atony are present throughout the esophagus. Palmer\(^7\) states, "Achalasia is a disease of the esophagus as a whole. In a sense the abnormality at the cardia is least important. Atony of the upper esophagus with absence of peristaltic activity is of primary importance." He reports that achalasia is much more common than cardiospasm.

There are a number of factors that may result in degeneration of Auerbach's plexus and achalasia and mega-esophagus. These include congenital absence of the plexus, vitamin B\(_1\) deficiency as reported by Etzel\(^6\), syphilis and other neurologic lesions, emotional factors, circumscribed lesions in the lower esophagus, reflex irritation from ulcer and other diseases, and allergy.

**Clinical Features**

The diagnosis of achalasia of the cardia is based upon the history and the roentgen and esophagoscopic findings.

Initially the patient may complain of slight pain, pressure, discomfort or burning at or near the xiphoid, or a sensation of food lodging in the lower esophagus after swallowing. The symptoms initially may be intermittent. Occasionally there may be an abrupt onset. The symptoms become progressively more severe and frequent until the retrosternal pain becomes continuous and severe. Attacks of distress may be precipitated by rough foods, cold fluids, acute infections, and emotional tension.

The pain may be referred to the neck, jaw, or ears, and may closely simulate angina pectoris in character and distribution, but is not related to exertion. With balloon studies and distention of the esophagus, anginoid type of pain and electrocardiographic changes have been shown to be associated with esophageal spasm.
Late in the course of achalasia, usually after dilatation of the esophagus has occurred, there may be a great loss in weight, a hypochromic anemia, and symptoms of vitamin deficiencies due to poor nutrition. Regurgitation of undigested food may occur. Pulmonary symptoms of cough and dyspnea may result from the pressure of a greatly distended esophagus, or from aspiration pneumonitis and bronchitis that are caused by repeated aspiration of undigested food during sleep.

Roentgen examinations show varying degrees of dilatation of the esophagus with retention of food. The esophagus may be so greatly dilated that a diagnosis of mega-esophagus can be made on roentgen study of the chest alone. Barium swallow shows the esophagus tapering to a smooth constriction at the level of the hiatus; this constriction is characteristic of achalasia. The margins of the esophagus are smooth and regular. Fluoroscopic examination usually shows an atonic dilated esophagus with weak, ineffective tertiary waves and no primary peristalses. Esophagoscopy is indicated to exclude the possibility of organic lesions of the distal esophagus.

Complications that may result from achalasia include esophagitis which usually is present with mega-esophagus, stasis of food, ulcerations and erosions in the esophagus, aspiration pneumonitis, and pulmonary abscess. Spontaneous pneumothorax following the Valsalva maneuver, pleural effusion, bronchiectasis, and fistulous communication with a bronchus also have been reported. Carcinoma of the esophagus is uncommon in patients with achalasia, although three such cases have been reported.

**Treatment**

The treatment of achalasia is essentially medical with esophageal dilations, bland diet, and supportive measures. Surgery is reserved for the patient whose condition does not respond to medical treatment. We agree with Bockus, that “All physicians who have had some experience with cardiospasm, believe that surgical operation of any type is contraindicated unless all methods of peroral dilation have failed.”

The basic principle of the medical treatment of achalasia is dilation of the cardiac sphincter. Various types of dilators are used, including hydrostatic and pneumatic dilators and mercury bougies. The dilators can be placed in position under fluoroscopic control; sufficient pressure must be used to disrupt some of the muscle fibers. One dilation may suffice, but frequently several are required. Some of our patients have learned to pass dilators themselves, and to perform dilation whenever their symptoms recur. One of Sippy’s principles was that where water would pass, a string could be passed, and where a string would pass, a dilator could be passed over the string. The first dilation may be done at the time of esophagoscopy examination.

Supportive measures are important and may be quite helpful. A bland, high-caloric, high-protein diet is recommended, and excessively hot and cold fluids should be avoided. Sedation may be helpful, but atropine, belladonna, and
Anticholinergic drugs are contraindicated, because Knight reproduced this condition in cats with a high vagotomy. Adrenolytic drugs may be of some help. Local anesthetic agents, such as procaine or Probutylin-Rorer, before meals have been of help in some patients. Large doses of vitamin B should be given not only because of the possibility that deficiency in B₁ is a factor in etiology but also because many of these patients are deficient in the entire vitamin B complex. Aminophylline and nitroglycerin have been used with beneficial effect in a few patients.

Sifers and Crile reported a follow-up study of 92 of 100 patients with cardiospasm and found only 18 failures on medical treatment, including dilations. Four of these 18 patients were successfully treated by surgical intervention.

Most of the patients in whom surgery is necessary have severe symptoms of mega-esophagus. Regurgitation, particularly if it occurs during sleep, with resulting cough, dyspnea, and aspiration pneumonitis, is an indication for surgery. Persistent anemia and malnutrition that do not respond to medical treatment are indications for surgery. It should be pointed out that surgical myotomy is very similar to medical dilation in that the circular muscular fibers are interrupted in both procedures. The size of a dilated esophagus rarely returns to normal after any surgical procedure, which is another indication that achalasia is a disease affecting the entire esophagus.

Effler and Rogers believe that linear myotomy is the preferred operation and report good results for 20 patients who underwent that procedure. Sweet reported his findings in a series of 48 patients who had undergone surgery for this condition. He divided his patients into two groups. Patients in group I did not complain of pain, but had dysphagia, malnutrition, and severe mega-esophagus; Sweet reported 80 to 100 per cent improvement in 13 of the 20 patients in this group treated by esophagoplasty. Patients in group II complained of substernal pain and had only moderate dilatation of the esophagus; at operation the circular muscle of the lower segment in these patients may be found to be thickened. In group II, Sweet prefers a linear myotomy, and reports a 90 to 100 per cent improvement in seven of seven patients after this procedure. Microscopically, degeneration of Auerbach's plexus was apparent in the patients in both of Sweet's groups.

Case Reports

The following selected cases demonstrate many of the clinical manifestations and complications of achalasia, and the indications for and results of medical and surgical treatment.

Case 1. A 24-year-old woman was first examined here in September 1949. She had a four-year history of substernal pain after meals which was frequently relieved by the drinking of water. For the six months preceding examination she had some regurgitation of undigested material, particularly during sleep. The patient volunteered the information that the symptoms were aggravated by rough foods and by emotional strain.
The findings on a complete physical examination were normal. Laboratory studies showed that the hemoglobin content was 12.2 grams per hundred milliliters, the serum albumin 4.8 and the serum globulin 3.6 grams per hundred milliliters. The remaining laboratory studies including urinalysis, serology, and determination of blood sugar level and basal metabolic rate were normal.

Roentgenograms of the chest were normal. Barium swallow revealed a typical cardiospasm with a funnel-like deformity at the cardiac end where the esophagus was approximately twice its normal width. No primary peristaltic waves were present.

On September 13, Hurst dilators, numbers 36 to 60, were passed and then a mercury bag was passed and inflated to 15 pounds for five minutes. After the dilation, the patient was given a bland diet, sedation, and vitamin B.

Subsequently, the patient's condition progressed satisfactorily. On April 10, 1956, approximately six years after initial examination, the patient's family physician reported that the patient had little or no dysphagia and only occasional episodes of epigastric burning; additional dilations had not been necessary.

Comment: This patient's condition responded satisfactorily to one dilation; however, several dilations frequently are required. The satisfactory progress of this patient's condition for a period of six years certainly indicates that surgery was unnecessary.

Case 2. A 33-year-old woman was first examined here on April 26, 1947, because of a one-year history of nausea, vomiting, and loss of 65 pounds in weight. For the year preceding examination, she also had difficulty in swallowing bread and other solids, and noted flatus, belching, bloating, retrosternal pain, dyspnea, and regurgitation of undigested food during sleep.

On physical examination the patient's weight was 118 pounds; the tongue was coated and dry and the buccal mucosa showed some desquamation. The remainder of the findings on physical examination and laboratory studies were normal. Barium swallow showed that the esophagus was enormously dilated throughout its entire length, and that the distal esophagus was cone-shaped (Fig. 1).

Esophagoscopy revealed that the esophagus was filled with food. No evidence of neoplasm was found when an esophagoscope was passed into the cardia. The patient underwent repeated esophageal dilations with mercury bougies inflated to 15 pounds. During the three months after initial examination she gained 22 pounds in weight.

The patient was next seen in May 1950, approximately three years after the initial examination. She weighed 204 pounds (a weight gain of 86 pounds in the three years). Her only complaint was that food occasionally lodged in the esophagus, particularly when she was not relaxed while eating. Barium swallow showed that the esophagus was less dilated than it had been in 1947. Constriction at the distal end was smooth and tapering, characteristic of cardiospasm. She was given a reducing diet, and during the next three months she lost 18 pounds in weight.

Approximately one year later, in July 1951, the patient was asymptomatic but she weighed 213 pounds. The findings on barium swallow were essentially the same as those in 1950. One esophageal dilation was performed.

When the patient was last seen in October 1954, seven and one-half years after initial examination, she still was asymptomatic. She weighed 206 pounds. Findings on complete physical examination, laboratory studies and roentgenogram of the chest were normal. Barium swallow showed no change in the appearance of the esophagus.

Comment: This patient had a one-year history of dysphagia, retrosternal pain, regurgitation of undigested food, vomiting, dyspnea, and loss in weight. A series of esophageal dilations resulted in regression of the symptoms and gain in weight of 95 pounds. She has remained asymptomatic for seven and one-half years, although the esophagus still

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Fig. 1. (Case 2) Esophagograms. A. Obtained in 1947. Large dilated atonic esophagus tapered at the cardia, consistent with a diagnosis of achalasia. B. Obtained in 1950. The esophagus is less dilated than it was in 1947 (A), and some return of tone to the wall is evident. C. Obtained in 1954. The esophagus still is somewhat dilated, but the dilatation is less than that in 1947 (A).
Fig. 2. (Case 3) A. Esophagogram showing dilated esophagus typical of achalasia. In patients with carcinoma of the esophagus, this extreme degree of dilatation is seldom seen. B. Spot film showing the tapered appearance of the lower end of the esophagus, which is typical of achalasia.
is dilated. When the esophagus becomes dilated to the extent that it was in this patient, medical or surgical treatment seldom can relieve the dilatation. However, this patient's condition responded well to one series of dilations, which indicates that dilatation of the esophagus in itself is not sufficient indication for surgery, since medical treatment may accomplish, as in this patient, everything that surgery would be able to do.

**Case 3.** A 42-year-old man was first examined here on November 15, 1951, because of a three-month history of a tickling cough in his throat, gagging, and vomiting. His symptoms were most distressing at night; he would awaken with a cough, have a small emesis, and then be comfortable for the rest of the night. For the previous ten years he had had some difficulty in swallowing solid food. The patient had no other respiratory symptoms and he had not lost weight.

Findings on physical examination were normal except for a mucopurulent nasal discharge. Laboratory examinations including urinalysis, serologic tests for syphilis, and determinations of blood count and blood sugar level were normal.

On roentgen examination, the chest was normal, but the esophagus was found to be dilated to a diameter of approximately 5 cm., with a smoothly narrowed and tapered distal esophagus (Fig. 2).

Esophagoscopy revealed fluid retained in the esophagus. There was a smooth narrowing of the esophageal hiatus, with no evidence of neoplasm. Esophageal dilation with a French no. 60 dilator was performed, and the patient was given a bland diet, sedation, and vitamin B complex.

The patient was last seen in February 1954, approximately three years after initial examination, at which time a thyroid adenoma was removed. He was asymptomatic in regard to the achalasia, although barium swallow showed no essential change in the appearance of the esophagus. The patient reported by letter in June 1956 that he had remained asymptomatic.

**Comment:** This patient had a ten-year history of mild dysphagia, and a three-month history of coughing, gagging, and vomiting at night. A diagnosis of achalasia and mega-esophagus was made. He has remained asymptomatic after one esophageal dilation.

**Case 4.** A 34-year-old woman was first examined here on June 30, 1954, with presenting complaints of vomiting, thoracic pain, and a weight loss of 60 pounds during the previous two years. She had frequently regurgitated during sleep, and she had difficulty in swallowing, particularly of solids. Thoracic pain was severe and relatively constant, but it was relieved by vomiting. Two esophageal dilations performed elsewhere, four and seven months previously, had partially relieved the symptoms. For two months preceding our examination the patient had been aspirating herself at bedtime with an Ewald tube with some relief.

Findings on physical examination were essentially normal. Laboratory studies including urinalysis, serologic tests for syphilis, and determinations of blood count, blood sugar level, serum proteins, and prothrombin time were normal.

Barium study of the esophagus revealed a smooth narrowing at the cardia with proximal dilatation to a diameter of 8 cm. (Fig. 3). Food and barium were retained in the esophagus at the end of two hours. Esophageal dilation with a no. 60F Hurst mercury bougie was performed, and the patient was placed on a bland diet with supplemental nourishment, sedation, and an adrenolytic medication; also the head of her bed was elevated eight inches. Initially, moderate improvement in the patient's condition was noted, but there were frequent aspirations at bedtime, and she continued to have pain in the chest, regurgitation during sleep, and some coughing.

The presence of aspiration pneumonitis made surgery advisable. On September 1, 1954, approximately two months after initial examination, a linear myotomy was
performed; the postoperative course was uneventful. The coughing and vomiting subsided and the patient gained 38 pounds in weight. Postoperative barium swallow showed that the diameter of the esophagus had decreased from 8 to 5 cm. (Fig. 4). A year later, in September 1955, she became pregnant. When she was last examined in April 1956, approximately two years after her initial visit, she was asymptomatic concerning the achalasia, and was tolerating her pregnancy satisfactorily.

Comment: This patient had a two-year history of dysphagia, vomiting, regurgitation, cough, thoracic pain, anorexia, and loss in weight. She failed to respond to medical treatment with dilations, continuing to have the thoracic pain, regurgitation during sleep, cough, and evidence of aspiration pneumonitis. After linear myotomy, she became asymptomatic. There was some decrease in the size of the dilated esophagus postoperatively; it is unusual for mega-esophagus to decrease in size after either medical or surgical treatment. Surgery is indicated when, despite medical treatment, thoracic pain and respiratory symptoms, frequently due to aspiration pneumonitis, persist. Surgery should

Fig. 3. (Case 4) Preoperative esophagogram showing moderate mega-esophagus and lack of tone. Surgery was performed because of persistent malnutrition, regurgitation, and aspiration pneumonitis, despite medical treatment including dilations.
be performed before irreversible changes have occurred in the cardiopulmonary systems.

**Case 5.** A 56-year-old man was first examined here on July 19, 1954. His presenting complaint was dyspnea that had its onset four years previously. Four months before initial examination here, the patient noted edema of the ankles, and shortness of breath at rest after prolonged talking. During the month before examination, he had several episodes of paroxysmal nocturnal dyspnea.

The history revealed the following information: at 11 years of age, he received a blow to the epigastrium which caused pain and abdominal distention for a short time; at 18 years of age, he first noted difficulty in swallowing which subsequently progressed until he could eat only soft foods and liquids; at 32 years of age, surgery was advised for achalasia and mega-esophagus but was refused; and at 34 years of age, he had syphilis. At 52 years of age, two episodes of severe upper-gastrointestinal bleeding occurred. Roentgen examination at that time showed mega-esophagus and duodenal ulcer, and surgery again was advised and refused.
Fig. 5. (Case 5) A. Roentgenogram of chest, showing enlargement of the esophagus extending beyond the enlarged border of the right side of the heart. B. Esophagogram showing a greatly enlarged and tortuous esophagus.
On physical examination the blood pressure was 118/80 mm. Hg, and the pulse rate was 96 per minute. The anteroposterior diameter of the chest was found to be increased and there were dullness at the bases of both lungs and many râles and rhonchi throughout both lungs. The heart was questionably enlarged and there was a loud presystolic gallop at the apex. Two-plus pitting edema of the ankles was present.

On laboratory examination the complete blood count, blood sugar level, serologic tests for syphilis, blood urea, and urinalysis were normal. Roentgen study of the chest showed pronounced enlargement of the esophagus with food and fluid levels extending far into the right pulmonary field. The pulmonary fields showed some fibrosis at both bases. Barium swallow showed gross dilatation of the esophagus to approximately 8 cm. in diameter with associated tortuosity (Fig. 5). After about two hours, two thirds of the barium still remained in the dilated esophagus. An electrocardiogram showed myocardial changes.

The patient refused to be hospitalized, so he was given medical treatment that consisted of a low-sodium diet, digitalis, and frequent mercurial injections.

During the first two weeks of treatment, the patient lost 15 pounds in weight. When the patient was last examined in June 1956, two years after initial examination, he still became short of breath after any exertion and frequently required diuretics. It was believed that this patient had pulmonary fibrosis with cor pulmonale and congestive failure and that the impairment of pulmonary and cardiac reserve contraindicated major operation.

Comment: This patient had a 38-year history of untreated achalasia that resulted in episodes of aspiration pneumonitis, and irreversible pulmonary fibrosis and emphysema, chronic cor pulmonale, and right cardiac failure. His condition illustrates the severe pulmonary and cardiac changes that can result from achalasia. Adequate medical treatment with dilations, diet, and other measures, at the onset of the achalasia might have prevented the pulmonary and cardiac changes. If dilations had been unsuccessful in controlling his symptoms, surgical treatment before the pulmonary and cardiac changes had become irreversible, probably would have been effective. Whether the blow to the abdomen or the syphilitic infection were etiologic factors in the achalasia is not known.

Summary and Conclusions

Achalasia, the failure of relaxation of the esophageal cardia, is associated with varying degrees of mega-esophagus, and should be regarded as a disease of the whole esophagus since primary peristaltic waves usually are absent throughout its entire length. The etiology of achalasia is unknown, but the condition frequently is associated with degeneration of Auerbach’s plexus; it has been experimentally reproduced in cats by high vagotomy, and it has been reported to be clinically associated with deficiency of vitamin B1.

The diagnosis is based upon: (a) history of food lodging in the lower esophagus, retrosternal pain, loss in weight, malnutrition, anemia, and regurgitation; (b) roentgen findings of diffuse dilatation of the esophagus tapering to a smooth constriction at the cardia, with a loss of primary peristaltic activity; and (c) esophagoscopic findings confirming the roentgen findings and excluding other disease.

The treatment of achalasia is primarily medical, chiefly through dilation of
the cardiac sphincter. Supplementary measures include diet, vitamin B, sedation, and other drugs. Surgery is indicated only in patients whose conditions do not respond to medical treatment, and who develop regurgitation and aspiration pneumonitis, persistent anemia and malnutrition, and persistent severe pain. The dilated esophagus rarely returns to normal size after either medical or surgical treatment.

The reports of five cases are presented which demonstrate the diagnostic findings, the treatment, and some of the complications of achalasia. The first patient received only one dilation, and has remained essentially asymptomatic for six years since treatment. The second patient received several dilations, and has remained asymptomatic and has gained 95 pounds in weight during the seven and one-half years since the original dilation. The third patient who had a ten-year history of dysphagia, has remained asymptomatic for five years since one dilation was performed. The results in these three patients demonstrate that the symptoms of achalasia can be alleviated by medical treatment, including esophageal dilation.

Despite medical treatment including dilations, the fourth patient continued to have symptoms (chest pain, regurgitation, coughing, and aspiration pneumonitis), and a linear myotomy was performed. She has remained asymptomatic for one and one-half years after operation. The fifth patient had had achalasia for 38 years, and repeatedly had refused either medical or surgical treatment. He developed some of the complications associated with long-standing untreated achalasia: a large mega-esophagus, regurgitation, aspiration pneumonitis, pulmonary fibrosis, and chronic right cardiac failure thought to be associated with the chronic pneumonitis. It was believed that because of the chronic cardiac failure, he would not survive any major surgical procedure.

References