Case Report

Iran J Pediatr
Jun 2010; Vol 20 (No 2), Pp:233-236

Familial Achalasia, a Case Report

Farzaneh Motamed, MD; Vajiheh Modaresi*, MD and Kambiz Eftekhari, MD

Department of Pediatrics, Children's Medical Center, Pediatric Center of Excellence, Tehran University of Medical Sciences, Tehran, IR Iran

Received: *May 03, 2009*; Final Revision: *Sep 24, 2009*; Accepted: *Nov 05, 2009*

Abstract

Background: Although achalasia is a relatively rare disease in pediatric age group, it must be considered for differential diagnosis of esophageal disorders in children with positive family history even in the absence of typical clinical manifestations.

Case Presentation: A 5-month old boy was hospitalized for cough and mild respiratory distress. Because of positive history of achalasia in his mother, achalasia was detected in esophgagography. Pneumatic dilation through endoscopy was successful. A 12-month follow-up revealed no problem.

Conclusion: Achalasia must be considered for differential diagnosis in children with positive family history of achalasia even in the absence of typical clinical manifestations. An autosomal recessive mode of inheritance is probable. We suggest further researches and genetic studies to establish the pattern of inheritance.

Iranian Journal of Pediatrics, Volume 20 (Number 2), June 2010, Pages: 233-236

Key Words: Achalasia; Familial Achalasia; Infantile feeding difficulties; Pneumatic dilation; LES

Introduction

One of the causes of dysphagia due to more distal primary esophageal dysmotility is achalasia with unknown etiology characterized by loss of lower esophageal sphincter relaxation and loss of esophageal peristalsis, both contributing to a functional obstruction of the distal esophagus.

It is a rare condition in children with an incidence range of 0.1-0.3 case per 100,000 children per year in UK[2]. Most associations are seen among siblings or in monozygotic twins. Parent-child association is even rarer[1]. Degenerative, autoimmune and infectious factors are possible etiologies. In rare cases, achalasia is familial or part of a syndrome.

Common signs and symptoms are based on duration and age of onset and can be variable from feeding problem to respiratory distress. Early diagnosis and appropriate treatment can prevent failure to thrive and respiratory sequels.

*** Corresponding Author;**

Address: Pediatric Gastroenterology Division, Children's Medical Center, Dr Gharib St., Keshavarz Blvd, Tehran, IR Iran
E-mail: drmodarressi@yahoo.com

Case Presentation

A 5-month old male infant was admitted for mild respiratory distress. He had no feeding problem prior to the onset of respiratory distress. The disease began around two weeks before admission. Parents were consanguineous (cousins, mother side). His mother, one of his cousins and also one of his aunts (mother side) suffered from dysphagia and poor weight gain for a period of time in preschool age. Lower esophageal sphincter (LES) myotomy was performed successfully for all of them with diagnosis of achalasia.

Physical examination revealed appropriate height and weight. Except for mild respiratory distress, no other positive finding was detected. We evaluated the patient by esophagography only because of his family history. Typical feature of achalasia in radiography was confirmed (Fig. 1).

Upper gastrointestinal endoscopy revealed a tight LES that would not open with insufflation of air into distal esophagus.

Through endoscopy Rigiflex balloon (20 mm diameter) was introduced by mouth in an antegrade approach. After a satisfactory position was obtained, the balloon was fully inflated, requiring about 10 ml of water, until the pressure reached 50 PSI and kept inflated for 60 seconds. Then, the balloon was rapidly deflated. Tearing was successfully induced without complication and the procedure was repeated for three consecutive sessions with 4 weeks interval.

A 1-year follow-up revealed no problem. Now he is in good nutritional state with appropriate weight and height.

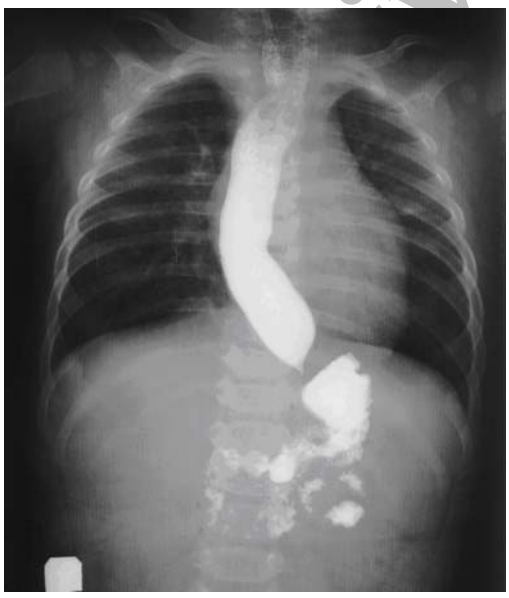

Fig. 1: Barium swallow showing esophageal dilatation and beaking

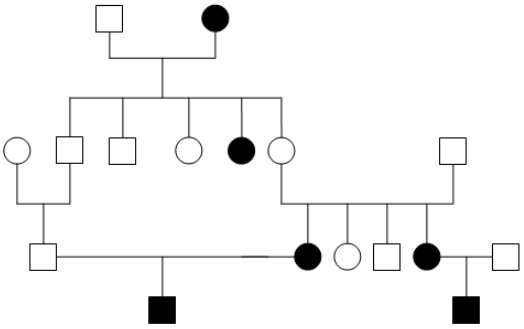

Fig.2: pedigree demonstrating pattern of inheritance.Black,affected patients

Discussion

Achalasia is a primary esophageal disorder involving the body of the esophagus and lower esophageal sphincter affecting all ages and equally both genders[3]. Fewer than 5% of all patients present in pediatric age group.

Incomplete relaxation of LES and loss of esophageal peristalsis are believed to be secondary to absent or abnormal inhibitory innervations in the myentric plexus in majority of cases. Much less frequently, a central nervous system lesion can cause LES achalasia.

Autoimmune, infectious and environmental factors are approved as main causes of these changes in some of the cases but most of time it is idiopathic[2]. The influence of genetic factors as a cause of achalasia is suggested by the clustering in certain syndromes like Algroove, Rozychi, and Pierre-Robin. Most of the cases are

sporadic. Reports of isolated familial achalasia represents less than 1% of all patients with achalasia[1,4,5].

Most cases of familial achalasia are horizontally transmitted as a result of consanguineous relationship, presenting in the pediatric age group and in siblings, suggesting an autosomal recessive inheritance[6]. Thibert, et al described 2 families, each with 2 affected siblings under 16 years of age[7].

Although the role of genetic predisposition to achalasia is argued by some authors, large community based-studies failed to identify familial clustering[8].

Mean age at diagnosis is usually 8.8 years and feeding problems are prominent manifestation, but presentation is depending on duration and age of onset. Sometimes it is accompanied by malnutrition or respiratory symptoms, and complicated by esophagitis because of retained esophageal food[3]. Infants and toddlers present with choking, cough, recurrent chest infections, feeding aversion and failure to thrive[5]. Achalasia can remain undiagnosed for years due to initially oligosymptomatic progression and relative low prevalence of disease[3], Of interest is to note that our patient presented with only mild respiratory problems in early infancy. He had no feeding problems and diagnosis was suspected because of positive familial history. He also did not show any signs of above mentioned syndromes and was in well nutritional condition, probably because of early diagnosis.

In most instances, the diagnosis of achalasia is considered after barium swallow, showing a variable degree of esophageal dilatation with tapering at the gastroesophageal junction[9]. This is the typical radiologic feature that was also obvious in this case (Fig. 1).

For diagnosis, esophageal manometry is the gold standard to confirm achalasia[3]. Compared to esophageal manometry, the positive predictive value of a barium swallow is as high as 96%[10]. Our first option was esophgography because manometry was not accessible in our center.

Endoscopy is both diagnostic and therapeutic in which dilatation can be made. The LES does not open with insufflation of air into distal esophagus[5]. The same problem was seen in our patient.

The two most effective treatment options are pneumatic dilatation and surgical (Heller) myotomy. Pneumatic dilation is the most common first-line therapy for the treatment of achalasia[11,12]. Nontheless, some studies showed that younger age may predict non-response to balloon dilation [13,14], but our case responded.

Pharmacologic treatment options are various. It has been shown that using nitrates, isosorbide dinitrate, and calcium channel blockers, with different ways of effect cause partially and transient improvement in some patients.

Botulism toxin injection into the LES is an effective, safe, and relatively simple method for the short-term treatment of achalasia, but the duration of its action is temporary and using it in pediatric group is not certain[2,9,15]. We did not need to try any other options because our patient responded effectively to endoscopic pneumatic dilatation.

Conclusion

Although achalasia is a very rare condition and seen in late childhood, it must be considered for differential diagnosis in children with positive family history even in absence of typical clinical manifestations especially if they are products of consanguineous parents. An autosomal recessive mode of inheritance is possible. We suggest further researches and genetic studies to demonstrate the pattern of inheritance.

References

1. Sachdev A, Sandhu BS, D'cruz S, et al. Achalasia cardia in mother and son. Indian J Gastroenterol. 2004;23(3):109.
2. Orenstein S, Peters J, Khan S, et al. Dysmotility disorders: Kligman, Behrman, Jenson, Stanton, Nelson Text Book of Pediatrics, 18th ed, Saunders Elsevier. 2007; Pp:1545-6.

3. Pohl D, Tutuian R. Achalasia: an overview of diagnosis and treatment. J Gastrointestin Liver Dis. 2007;16(3):297-303.
4. Mudawi HM, Fedail SS. Familial achalasia in Sudan. Trop Gastroenterol. 2004;(1):25-27.
5. Boeckxstaens GF, Jonge WD, van den Wijngaard RM, et al. Achalasia: from new insights in pathophysiology to treatment. J Pediatric Gastroenterol Nutr. 2005: (41):S36-7.
6. Hernandez A, Reynoso MC, Soto F, et al. Achalasia microcephaly syndrome in a patient with consanguineous parents: support for AM being a distinct autosomal recessive condition. Clin Genet. 1989; (36):456-8.
7. Mayberry JF, Atkinson M. Family occurrence of achalasia. (Letter) Gut. 1989;(30):559.
8. Thibert F, Chicoine R, Chartier-Ratelle G. Forme familiale de l'achalasie de l'oesophage chez l'enfant. Un Med Canada. 1965; (94):1293-300.
9. Andersson M, Lundell L, Kostic S, et al. Evaluation of the response to treatment in patients with idiopathic achalasia by the timed barium esophagogram: results from a randomized clinical trial. Dis Esophagus. 2009; 22(3):264-73.
10. Vaesi MF, Baker ME, Achkar E, et al. Timed barium oesophagogram: Beter predictor of long term success after pneumatic dilation in achalasia than symptom assessment. Gut. 2002; (50):765-70.
11. Bittinger M, Wienbeck M. Pneumatic dilation in achalasia. Can J Gastro-enterol. 2001;15(3):195-9.
12. Chuah SK, Hu TH, Wu KL, et al. Clinical remission in endoscope-guided pneumatic dilation for the treatment of esophageal achalasia: 7-year follow-up results of a prospective investigation. J Gastrointest Surg. 2009;13(5):862-7.
13. Mehta R, John A, Sadasivan S, et al. Factors determining successful outcome following pneumatic balloon dilation in achalasia cardia. Indian J Gastroenterol. 2005;24(6):243-5.
14. Zhang Y, Xu CD, Zaouche A, Cai W. Diagnosis and management of esophageal achalasia in children: analysis of 13 cases. World J Pediatr. 2009;5(1):56-9.
15. Ahmed A. Achalasia: what is the best treatment? Ann Afr Med. 2008;7(3):141-8.