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Familial infantile oesophageal achalasia

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Accepted 16 July 1991

(*Arch Dis Child* 1991;66:1353-4)

Abstract

Oesophageal achalasia is uncommon in children and in its familial form it is a rarity. The presentation and management of two male siblings who presented with oesophageal achalasia as infants are reported. A high degree of consanguinity in the parents of the children existed, suggesting autosomal recessive transmission.

Oesophageal achalasia is a disease of unknown aetiology characterised by a functional obstruction of the lower oesophagus due to failure of relaxation of the lower oesophageal sphincter and altered motility of the body and distal oesophagus. It is an uncommon disease, usually presenting in adult life, and its occurrence in childhood is rare. Occasionally it occurs in a familial form. We report two male siblings who presented with oesophageal achalasia during infancy and who underwent corrective surgical treatment.

Case reports

The parents of the children were first cousins. Clinical examination and laboratory investigations excluded any syndromic type of achalasia in both cases.

CASE 1

A first born boy presented at 5 months of age with recurrent respiratory tract infection, abdominal distension, and failure to thrive. A barium swallow performed showed typical features of achalasia. At operation at the age of 10 months the child underwent a transabdominal modified Heller's anterior oesophagomyotomy of 8 cm length and Nissen fundoplication. The child is now well and thriving after three years of follow up.

CASE 2

The second and only other child in the family was also male and presented at 8 months of age with a history of respiratory tract infections,

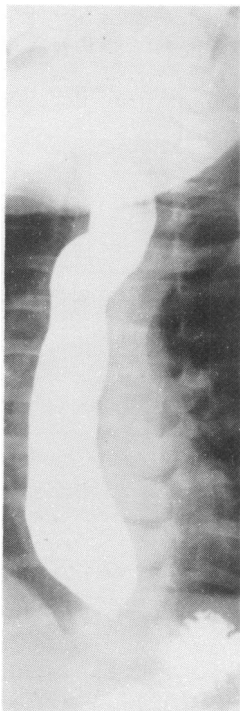
regurgitation and vomiting, and failure to thrive since birth. Barium swallow was performed which again showed the features of well established achalasia (figure). Operation was performed at 10 months of age and again a transabdominal modified Heller's anterior oesophagomyotomy (8.5 cm long) with Nissen fundoplication was performed. The child made a good recovery postoperatively and is now thriving after six months of follow up.

Discussion

Achalasia of the oesophagus is uncommon with an incidence estimated at approximately one per 100 000 population per year.¹ In children achalasia is rare with only 2% of all cases presenting before the age of 6 years.²

A familial form of achalasia presenting in infant siblings was first described by Thibert *et al* in 1965.³ Before this in 1962 Tyce and Brough reported a family with multiple diseases inherited including mental retardation, oesophageal achalasia, speech disorder, and neurological diseases.⁴ Dayalan *et al* in 1971 reported the presentation of three siblings with achalasia during the first year of life.⁵ In the case they reported the parents of the affected children were closely consanguineous, the father being the maternal uncle of the mother; there were two boys and one girl.

Westley *et al* in 1975 proposed that infantile achalasia is inherited as an autosomal recessive disorder when describing its occurrence in an Apache Indian kindred.⁶ He postulated that the high degree of inbreeding allowed a rare recessive gene to be expressed in several members. The existence of consanguinity in the parents of the children in this report lends further weight to this argument. Although vertical transmission of oesophageal achalasia has been described, the lack of consistent vertical transmission is also thought to indicate a probable autosomal recessive gene disorder. Further support for autosomal recessive inheritance is the well documented occurrence of achalasia in association with other conditions having a similar mode of inheritance.



Barium swallow in case 2 showing achalasia.

We suggest that familial achalasia of the oesophagus may be transmitted as an autosomal recessive trait. We further suggest that parents of affected children be counselled regarding the risk of transmission to other offspring. A high index of suspicion is warranted in the follow up of siblings of index cases.

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Neonatal BCG

Randomised controlled trials have estimated the protective efficacy of BCG to be anything from zero to 80%. A recent study has put it at up to 97%. So we seem to be just three points short of a full house. Case-control studies may produce results in a much shorter time than randomised controlled trials, which means that not only do you have to wait a shorter time for the results but they may be more relevant to present circumstances when you get them.

The recent study referred to above is an excellent case-control study from Bangkok (Sayomporn Sirinavin and colleagues, *Pediatric Infectious Disease Journal* 1991;**10**:359-64). Altogether 130 cases of tuberculosis in children aged 3 months to 14 years were diagnosed in the two study hospitals between March 1987 and August 1988. These gave 75 cases and 207 controls suitable for analysis, the controls being matched for year of birth and district of residence. Those who had received neonatal BCG vaccination were compared with those who were unvaccinated against tuberculosis. Children who have been given BCG after the neonatal period were excluded from the analysis.

The overall protective efficacy of neonatal BCG was 83%. When the analysis was confined to laboratory confirmed cases and adjusted for socioeconomic status and household contact, the protective efficacy was 97%.

Neonatal BCG was shown to confer greater protection against more severe disease. In other words children who get tuberculosis despite vaccination are likely to have a less severe form. The protective efficacy against respiratory tuberculosis was only 64% but against superficial glandular tuberculosis it was 81% and against disseminated disease (meningitis, pericarditis, abdominal tuberculosis, and skeletal disease) 100%. Protection decreased from 96% in the first five years to 71% after six to 14 years. The degree of exposure to the disease is also important. With no household contact protection was 90% but when there was household contact it was only 50%.

The protection from neonatal BCG doesn't last forever. The World Health Organisation has apparently recommended revaccination at six years but the Thai workers suggest that 10 years would be more appropriate.

These results seem to provide ample justification for continuing to offer BCG vaccination in the neonatal period to at-risk groups in Britain and confirm its effectiveness for worldwide use.

ARCHIVIST