

# Epidemiological study of achalasia in children

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**SUMMARY** One hundred and twenty nine children under the age of 15 years were diagnosed as having achalasia in Britain and Ireland between 1976 and 1985 by 175 surgeons with an interest in paediatrics or oesophageal disease. The incidence in Eire was 0.31 cases/10<sup>5</sup> population/year and this was significantly higher than in England and Wales (0.1/10<sup>5</sup>/year), Scotland (0.06/10<sup>5</sup>/year/or Northern Ireland (0.02/10<sup>5</sup>/year). Details of age at diagnosis were only obtained for 36 of the 129 patients. The disease appeared to become commoner with increasing age but there is no evidence of a delay in diagnosis until early adulthood. Such a delay would not account for the apparent regional variations in incidence, as they have also been reported in adults. Southern Irish children are at greater risk of the disease than other children in the British Isles.

To date there have been no specific epidemiological studies of achalasia in childhood, although the clinical impression has been that the condition is particularly rare. There have been a number of small, mainly surgical series reporting the outcome of treatment but no attempt has been made to identify possible aetiological factors in childhood. Achalasia in children has the classical motility abnormalities which are seen in adults. Many paediatricians and paediatric surgeons consider oesophageal motility studies unpleasant, however, and accept a diagnosis based on the characteristic radiographic changes seen on a barium swallow. The vast majority of children with achalasia are treated surgically with a myotomy; endoscopic dilatation has been shown to be of limited benefit in the young and very young.<sup>1-4</sup> It is probable that most children with achalasia undergo a cardiomyotomy, although the diagnosis may not have been established as rigorously as in adults. There have been case reports of family clusters of achalasia, although a detailed study of about 1000 first degree relatives of adults with achalasia failed to identify any genetic factor.<sup>5</sup> In these family clusters the proband has often been a child. In this study we have attempted to measure the incidence of achalasia amongst children and to look at regional variations in frequency. Previous work on adult achalasia<sup>6</sup> has shown that the condition is commoner in Eire than in other parts of the British Isles and in this study of children this difference was re-examined.

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## Methods

### PATIENTS

All ninety six members of the British Association of Paediatric Surgeons (BAPS) were asked to provide details of children who were diagnosed or treated for achalasia during the decade 1976-1985. Children were defined as people under the age of 15 years. Details collected included sex and age at diagnosis. Where such information could not easily be obtained by examination of patients' notes, consultant surgeons were asked to provide details of the total number of cases treated during the decade. Twenty members of the British Oesophageal Group (including 13 thoracic surgeons) and 61 members of the Society of Thoracic and Cardiovascular Surgeons of Great Britain and Ireland were asked to provide similar details of numbers of patients treated between 1976 and 1985. These 81 surgeons were not members of the British Association of Paediatric Surgeons and included all those who were interested in oesophageal and thoracic surgery. Cardiac surgeons were not included in the survey. It is unlikely that major paediatric surgery would be performed by someone who was not a member of one of these professional societies.

Attempts were made to validate the data by: (1) Identifying all cases reported by surgeons from Nottingham County, reviewing their notes and comparing these results with those obtained from an independent computer based register of all hospital admissions in the area (Hospital Activities Analysis). (2) Comparing the observed number of cases in

England and Wales with those expected if the incidence of 0.09/10<sup>5</sup> children under 15 years old/year in Oxford held true throughout the county.<sup>6</sup> A similar comparison was made for Scotland where the incidence was 0.26/10<sup>5</sup>/year.<sup>6</sup> (3) Comparing the observed number of cases in children with the total number of cases reported for each country in a study of prevalence.<sup>6</sup> (4) Comparing the number of surgeons/10<sup>5</sup> population in each country to detect any differences in delivery of health care to the young.

The incidence in various countries was compared by  $\chi^2$  and also by Fleiss's technique<sup>7</sup> using 95% confidence intervals. The rates of the disease in Eire were compared with the rates of disease in England and Wales, Scotland and Northern Ireland separately.

## Results

All 96 paediatric surgeons and 19 of 20 oesophageal surgeons completed returns on the number of children they had treated with achalasia. Sixty of the 61 thoracic surgeons also replied (Table 1). The data from these surgeons were used to calculate the minimum likely incidence of achalasia in children under 15 years old. The midterm population data for the decade 1976–1985 were obtained from the Registrar General of the four countries of the British

Table 1 Achalasia in children (1976–1985)

| Association  | Members surveyed (n) | Members who replied (n) | Cases reported (1976–1985) (n) |
|--|----------------------|-------------------------|--------------------------------|
| British Association of Paediatric Surgeons                                   | 96                   | 96                      | 99                             |
| Society of Thoracic and Cardiovascular Surgeons of Great Britain and Ireland | 61                   | 60                      | 24                             |
| British Oesophageal group  | 20                   | 19                      | 6                              |

Members of these three British associations of surgeons were asked to provide details of the numbers of children treated with achalasia between 1976 and 1985.

Table 2 Achalasia in children (1976–1985)

| Country           | Midterm population (0–14 years old) | Cases reported (n) | 95% confidence limits for cases (n) | Incidence (cases/10 <sup>5</sup> pop/year) |   |
|-------------------|-------------------------------------|--------------------|-------------------------------------|--|---|
|                   |                                     |                    |                                     | Actual incidence                           | Range of incidence based on 95% confidence limits |
| Eire              | 484 650                             | 15                 | 9–26                                | 0.31                                       | 0.19–0.54   |
| England and Wales | 10 816 600                          | 106                | 88–129                              | 0.10                                       | 0.08–0.12   |
| Scotland          | 1 203 233                           | 7                  | 3–15                                | 0.06                                       | 0.02–0.12   |
| Northern Ireland  | 443 480                             | 1                  | 0–7                                 | 0.02                                       | 0–0.16  |

Children with achalasia treated between 1976 and 1985 in various parts of the British Isles and Ireland were identified by surgeons working in those countries. The 95% confidence limits for number of cases was calculated by Fleiss's method (1981).

Isles – Eire, Northern Ireland, England and Wales, and Scotland.

One hundred and six cases were reported from 21 centres in England and Wales, seven cases from two centres in Scotland, 15 cases from three centres in Eire and one patient from Northern Ireland. The incidence of achalasia in Eire was 0.31/10<sup>5</sup>/year and was significantly higher than that in England and Wales (corrected  $\chi^2=17.5$ ; one degree of freedom;  $p<0.001$ ), Scotland (corrected  $\chi^2=14.8$ ;  $p<0.001$ ) and Northern Ireland (corrected  $\chi^2=9.5$ ;  $p<0.001$ ). There was no significant difference in incidence between Scotland, Northern Ireland, and England and Wales (Table 2). When total numbers of cases were considered and 95% confidence limits calculated, Fleiss's<sup>7</sup> method of comparison gave similar levels of significance. Eire had a significantly higher incidence of achalasia in children than England and Wales ( $z=4.2$   $p<0.001$ ), Scotland ( $z=3.1$ ;  $p<0.005$ ), and Northern Ireland ( $z=3.1$ ;  $p<0.005$ ).

Using data on total number of cases obtained from the study of prevalence throughout the British Isles,<sup>6</sup> the proportion of young people with the disease was similar in Eire and England and Wales ( $z=1.57$ ; not significant) and Northern Ireland ( $z=1.48$ ; not significant). It was marginally commoner than in Scotland ( $z=2.12$ ;  $p<0.05$ ). This is strong evidence against the suggestion that a delay in diagnosis could be responsible for the regional variations in incidence.

Of the 129 cases details of age at diagnosis and sex were reported for 36 patients of whom 16 were girls and 20 boys. The mean age at diagnosis in the boys was 9.1 ( $\pm 4.1$ ) years and similarly in the girls 10.1 ( $\pm 4.4$ ) years. The disease was more commonly diagnosed in older children (Figure), but in view of the small number of cases in whom age at diagnosis was reported age specific incidences were not calculated. Although not specifically requested three of these 36 children were reported to have congenital malformations such as mental retardation, Down's syndrome and a ganglioneuroblastoma. Three other children from England with achalasia were related.

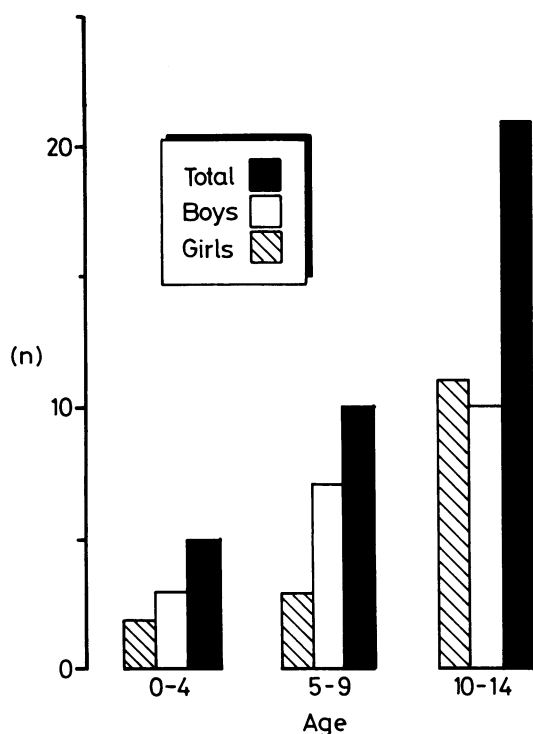


Figure Age at diagnosis of achalasia in children.

#### VALIDITY OF DATA

The surgical care from paediatric, thoracic, and oesophageal surgeons available to children with achalasia was not statistically different in Eire (2.3 surgeons/ $10^5$  population of children), England and Wales (1.2/ $10^5$ ), Scotland (2.4/ $10^5$ ), and Northern Ireland (1.1/ $10^5$ ).

Three cases were reported in Nottingham County between 1976 and 1985; one was treated in Nottingham and two in Sheffield. The diagnosis was confirmed in all three cases. The incidence of 0.15/ $10^5$ /year was similar to that of 0.09/ $10^5$ /year in Oxford and it seems likely that the majority of cases were detected in the survey. Further support for this view comes from the national figures. If the incidence in Oxford was typical of England and Wales 97 cases (95% confidence interval 79–119) would have occurred between 1976 and 1985 compared with the 106 (95% confidence interval 88–129) reported in this study. If 97 cases had been reported instead of 106, the disease would still be significantly less common than in Eire ( $z=4.98$ ;  $p<0.001$ ). The number of cases from Scotland (seven) was, however, less than the 31 estimated from Scottish Record Linkage.<sup>6</sup> Even if 31 cases had been reported, the incidence would still have been significantly less than that observed in Eire ( $z=3.8$ ;  $p<0.001$ ). These checks on the validity of the

data confirm that achalasia is more common in Eire than the rest of the British Isles.

#### Discussion

There have been few epidemiological studies of achalasia<sup>8-11</sup> and none have looked specifically at the disease in childhood. Clinical impressions are that it is an unusual disease in childhood with an incidence reported in this study of less than 0.1/ $10^5$  population/year in mainland Britain. It does, however, occur with significantly greater incidence in Eire where a high prevalence of achalasia has also been reported in adults.<sup>6</sup> In this study we have reported the minimum likely incidence of achalasia in various parts of the British Isles; it is probable that the number of cases were under reported and the true incidence may be higher. Perhaps the main reasons for this are the low incidence of the disease in childhood and the definition of cases. Oesophageal manometry is rarely carried out in young people although the diagnosis can usually be established by barium swallow examination. It is unlikely that the pool of undiagnosed cases is substantial as the condition causes symptoms of dysphagia and recurrent chest infection. In an epidemiological study from Nottingham<sup>11</sup> less than 5% of cases identified during an 18 year period were asymptomatic.

Despite this it seems likely that there is a truly higher incidence of achalasia in Eire. This survey has shown a fairly high detection rate of cases in England and Wales; although less so in Scotland. There was no apparent difference in availability of surgeons between Eire and England.

The increased number of cases in Eire was true at all ages with no disproportionate increase in young people compared with older patients with the disease. Consequently there is no evidence of a more prolonged delay in diagnosis in young people. Indeed evidence from Nottingham has shown a median delay in diagnosis of six months in those aged 11 to 30 compared with two years in those aged 31–50. It is of interest that as many as six cases of achalasia were treated in County Galway, an area also known to have a high prevalence of coeliac disease.

The accuracy of surgeons recall was not assessed. The majority however, (115), reported no cases with an interquartile range of 0 to 1 case. It is unlikely that surgeons would fail to recall such an unusual event as achalasia in childhood, which would have been treated by a distinctive procedure – Heller's myotomy. In Nottingham County all patients identified in earlier epidemiological studies were reported by surgeons in this survey. It seems likely that the data collected by this postal survey of surgeons are an accurate reflection of their clinical experience.

During childhood there have been a number of case reports of familial clusters of cases<sup>12-15</sup> and there is some limited evidence from this study to support this view. In adults there is no evidence to support a familial component to the disease.<sup>5</sup> Further studies of possible aetiological factors among children may clarify our understanding of the cause of this disease.

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