Final report of completed studies

Hirschsprung Disease

Background

Hirschsprung disease (HD) is a disorder characterised by aganglionosis of the distal bowel. The disease usually presents in the newborn period as a large bowel obstruction. Babies born with HD often have delayed passage of meconium (>24 hours after birth). Abdominal distension and vomiting may occur after the first feed. However, HD may have a more insidious course, which includes constipation, diarrhoea, anaemia, growth delay or enterocolitis. Although untreated enterocolitis is often fatal, increased awareness and modern methods of treatment have improved survival.

Objectives

- To estimate the incidence of HD in Australia and describe its demographic and clinical features
- To gain a more complete picture of the family history of HD
- To document anomalies associated with HD

Case definition

Any child under 15 years of age with newly diagnosed Hirschsprung disease seen in the last month. Aganglionosis of the distal bowel must be confirmed by either submucosal or seromuscular biopsy.

Results and discussion

Cases in 2000

In 2000, 66 cases of HD were notified, for which, 49 (74%) questionnaires were returned. There were twelve duplicates and one error. Thirty six cases of Hirschprung's disease were confirmed. Of these, 28 (78%) were males. In 30 (83%) infants the diagnosis was made in the neonatal period.

The most common presenting features included abdominal distension (30/36 or 83%), delayed passage of meconium (25/36 or 69%) and vomiting (12/36 or 33%). Constipation was present in five of the 36 (14%) children and enterocolitis in seven (19%). Three children had Down syndrome.

Suction biopsy was performed in 33 (92%) patients. A contrast study was performed in eleven (31%) patients and was consistent with the diagnosis of HD. Contrast study and rectal suction biopsy was the most commonly used combination of investigations and was used in eleven (31%) patients. Two patients underwent contrast study, suction rectal biopsy and laparotomy to have the diagnosis confirmed.

Nineteen children had a primary repair and the remaining 17 had a staged repair. Of these 17 infants, eight are still to have their final procedure.

Cases in 1997-2000

A total of 124 children with HD have been identified for the four years, 1997-2000, giving a reported incidence of 0.8/100 000 (95%CI 0.7-0.9) children aged <15 years. A total of 108 children were diagnosed in the neonatal period giving a reported incidence of 10.8 /100 000 (95%CI 0.9-13.0) live births.

The site or length of aganglionosis for children born in 1997-2000 is shown in Table 10.

Table 10 Length & site of aganglionosis of children with Hirschsprung's disease, 1997 - 2000

Length/Site	1997	1998	1999	2000	Total
Ultra short	1	1	1	1	4
Rectal	4	4	6	7	21
Sigmoid	7	11	18	19	55
Descending colon	1	4	-	1	6
Splenic flexure	1	0	1	1	3
Transverse colon	1	4	2	2	9
Caecal	1	1	-	-	2
Ileal	1	2	-	-	3
Proximal small bowel	1	1	1	-	3
Unknown	10	3	-	5	18
Total	**28**	**31**	**29**	**36**	**124**

Of reported cases, there have been a total of 13 children with Down syndrome and Hirschprung's disease. Other associated anomalies include cardiac (2), bilateral congenital talipes equinovarus (2), developmental delay (2) and congenital dislocation of the hip (1).

Surgical procedures used for HD in children born in 1997-2000 are shown in Table 11. Information on the procedure used was unavailable in 18 cases.

Table 11 Surgical procedures used for Hirschsprung's disease in children, 1997 - 2000

	1997	1998	1999	2000	Tota
Primary Procedure					
Soave	2	3	7	17	29
Duhamel	3	2	1	-	6
Swenson	2	0	2	-	4
Other			1	2	3
Secondary Procedure					
Soave	10	7	3	4	24
Duhamel	5	1	1	2	9
Swenson	-	-	1	1	2
Other	3	1	-	1	5
Not yet done	1	2	13	8	24
No information	2	15	-	1	18
Total	**28**	**31**	**29**	**36**	**124**

Of the 106 children for which all information was available, staged surgical repair (the favoured treatment) was performed in 62 (58%) children. However Primary repair was increasingly used over the study period.

Conclusion

The estimated incidence of Hirschprung's Disease cited in the literature is approximately 20/100 000 live births. This study identified 108 infants with Hirschsprung's Disease who were diagnosed in the neonatal period over four years. This is a rate of 11/100 000 live births. This discrepancy may be due to incomplete notification and/or incomplete return rate of questionnaires after notification. However, this is the first attempt to estimate the incidence of HD in Australia, where the rate may in fact be lower than in other countries. The majority (87%) of children with Hirschprung's disease were diagnosed in the neonatal period. This is an improvement compared with past series in the literature, which have shown that less than 15% children had been diagnosed with HD within the first month of life[1].

Interestingly only 69% of children overall were reported to have delayed passage of meconium. Abdominal distension and vomiting were reported more commonly. The absence of delayed passage of meconium should not delay investigation, such as suction rectal biopsy, if Hirshsprung's Disease is suspected. In this study, rectal suction biopsy was the most commonly performed investigation to diagnose Hirschsprung's disease.

The most serious complication of Hirshsprung's Disease is enterocolitis, which can occur either pre or post-operatively. In the past this has been a major cause of death in these children but this series has not reported a single death. This decrease in mortality may be due to increased awareness, prompt diagnosis and appropriate treatment with rectal decompression, intravenous fluid and antibiotics.

Although a staged surgical procedure is still the most common management, the use of a primary pull-though procedure increased during the study period. The Soave technique was the most often used primary procedure.

This study has provided valuable information on the clinical features of Hirschsprung's disease in Australia and its current management. It has also demonstrated some directions for future research. An example would be assessment of the diagnostic accuracy of the suction rectal biopsy.

We would like to acknowledge the dedication of Australian paediatric surgeons and paediatricians for their efforts in notifying cases of HSCR, completing questionnaires and clarifying aspects of the data.

1. Kleinhaus S, Boley SJ, Sheran M, Sieber WK. *J Pediatr Surg*. 1979;14:588-97.

Investigators

Professor Daniel Cass, (Principal Investigator)
Ms Patricia Manglick, Scientific Officer
Dr Shalinder Singh, Consultant Surgeon
Department of Surgery, Children's Hospital at Westmead, Locked bag 4001, Westmead NSW 2145
Tel: 02 9845 3059 Fax: 02 9845 3082
Dr Edi Shi, Department of Surgery, Sydney Children's Hospital, High Street, Randwick NSW 2031
Dr Rosslyn Walker, Paediatric Surgeon, Taylor Medical Centre, 40 Annerley Road, Woolloongabba QLD 4102
Dr Robert Yardley, Department of Paediatrics, Monash Medical Centre, 246 Clayton Road Clayton VIC 3168