

Australian Paediatric Surveillance Unit

PROTOCOL SHEET - HIRSCHSPRUNG'S DISEASE (from January 1997)

Background to study

Hirschsprung's disease results from a lack of ganglion cells in the distal colon. Presentation is most often in the neonatal period with delayed passage of meconium, vomiting or abdominal distension. Less severe cases may present later in childhood with constipation, abdominal distension or bowel obstruction. Although the diagnosis may be suspected clinically and from barium enema examination, it can only be confirmed by detection of aganglionosis in the distal colon on submucosal (rectal suction) or seromuscular (surgical) biopsy.

The extent of colonic involvement is variable, ranging from very short segment to total intestinal involvement. The extent of intestinal involvement will affect both presentation and treatment. Definitive surgical treatment includes Soave, Duhamel and Swenson procedures which may be preceded by colostomy.

Despite increased awareness of the condition and improved treatment, Hirschsprung's disease still has significant morbidity and occasional mortality. An incidence of 1:4000-5000 has been reported. In Australia between 50-60 new cases would be expected each year. Previous studies suggest an unexplained male predominance and have identified a number of associated conditions including Down syndrome, Smith-Lemli-Opitz syndrome, neuroblastoma, extra digits, skin depigmentation, isolated cardiac defects, Crohn's disease and thyroid carcinoma in patients or their families.

Four gene defects for Hirschsprung's disease have now been identified but probably only account for about 30% of cases. **Positive family history may be missed unless an extended family history is taken.**

Objectives:

1. To define the epidemiology of Hirschsprung's disease in Australia - its incidence, demographic and clinical features
2. To determine the extent to which a positive family history exists
3. To document associated anomalies

CASE DEFINITION and REPORTING INSTRUCTIONS

Please report any child under 15 years of age newly diagnosed with Hirschsprung's disease in the last month and in whom aganglionosis of the distal bowel has been confirmed by either submucosal or seromuscular biopsy.

Follow up of notifications

Clinicians notifying a case of Hirschsprung's disease will be requested to complete a two page reply-paid questionnaire. **Since information about family history is requested which you may not routinely ask, we have enclosed a copy of the questionnaire for your information.**

Genetic testing available for cases

Testing for the known gene defects associated with Hirschsprung's disease is currently available at the Royal Alexandra Hospital for Children, Westmead and will be performed at no charge on request of the notifying clinician on patients reported to the APSU. The work is mostly research and samples are analysed in batches, priority being given to samples from patients with a positive family history, long segment disease or associated anomalies. Interested clinicians should contact the Investigators Dr Danny Cass on 02 9845 3059, or Mr Lawrence Lam on 02 9845 3055, for further information.

Support group

For the information of clinicians, the Australian Pseudo-obstruction Support Association (APSA) provides support to families and carers of those who suffer from Hirschsprung's Disease, pseudo-obstruction syndrome and gastrointestinal motility disorders. Contact the secretary on PO Box 40, Oakdale NSW. Tel: 046 596186.

Investigators

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References

1. Cass DT, Unravelling the pathogenesis and molecular genetics of Hirschsprung's disease. *Semin Neonatol* 1996;**1**:211-217
2. Cass DT, Hirschsprung's disease: improving care and discovery of gene defect. *Modern Medicine of Australia* 1994;**July**:42-49