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Total intestinal aganglionosis
An autosomal recessive condition?

The risk of long-segment Hirschsprung’s disease recurring in further sibs of a patient is well known. Ehrenpreis (1970) estimated the incidence to be 12\%.

The extreme variety of this condition in which the entire large and small bowel is aganglionic seems to constitute a separate group. This is a fatal condition and fortunately is rare. The patient described here is a further affected sib in the family reported by Ahmed et al. in 1971. The second child born in the family was a normal healthy female.

Case report
The male third child of healthy unrelated white British parents was born at 38 weeks’ gestation by normal delivery at home. When feeding was begun he vomited bile-stained fluid. He was admitted as an emergency to hospital late on the third day of life, by which time he had not passed meconium; he was not dehydrated and weighed 3·02 kg. The abdomen was distended but soft. Rectal examination suggested the presence of a microcolon. The full details of the family history were not then available and the possibility of total intestinal aganglionosis was not considered. It was only later that the case notes of the eldest sib were made available, showing that she had died at the age of 20 days from total intestinal aganglionosis.

Hb was 16·6 g/dl, WBC 5·7 x 10^9/l. Serum electrolytes were normal. X-ray of abdomen showed several distended intestinal loops with fluid levels, while in the right lower quadrant there was the characteristic stippled appearance of meconium. A diagnosis was made of small bowel obstruction possibly due to a complicated meconium ileus. At laparotomy the proximal third of the small bowel was grossly distended with a serosal tear already present. The terminal ileum was full of putty-like meconium and was of narrow calibre, as was the colon. The contents of the proximal small bowel were more fluid than commonly found in meconium ileus. The jejunum was deflated by aspiration and the serosal tear repaired, and a cecostomy was established through which the distal ileum was emptied.

The following day, after reviewing the case notes of the eldest sib, the diagnosis of total intestinal aganglionosis was considered. This was confirmed firstly by rectal biopsy, then at a further laparotomy the extent of the aganglionic segment was shown to include the entire small bowel. The child died 14 days after admission and post-mortem studies showed that there were ganglion cells present only as far as the pylorus.

Discussion

The aganglionic segment of Hirschsprung’s disease is limited to the rectosigmoid colon in 70\% of cases. More extensive involvement is described as ‘long-segment disease’, but total colonic aganglionosis and

![Family trees in total intestinal aganglionosis.](http://adc.bmj.com/ViewFullImage.aspx?id=283252)
small bowel aganglionosis occur in only 1–10% of various series. Total intestinal involvement is the rarest form of the disease.

Hirschsprung's disease is probably multifactorial in its causation (Bodian and Carter, 1963). However, in one strain of mice a similar condition appears to be inherited as an autosomal recessive (Bielschowsky and Schofield, 1962). If the family reported here and the 10 previously reported cases of total intestinal aganglionosis are combined (Lee, 1955; Boggs and Kidd, 1958; Bodian and Carter, 1963; Walker et al., 1966; Ahmed et al., 1971; Talwalker, 1976) 50% of sibs are affected, and none with a lesser degree of aganglionosis. In those reports which include a description of other sibs (Fig.), and allowing for failure to ascertain those families at risk in which no children have been affected, the pattern suggests that total intestinal aganglionosis may be a distinct entity, inherited as an autosomal recessive.

Summary

A case of total intestinal aganglionosis in a sib of a previously recorded patient is presented. The number of cases now reported is 9 in six families. The possibility that this condition is a distinct entity inherited in an autosomal recessive manner is discussed.

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References


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Idiopathic late-onset immunoglobulin deficiency with associated defect in cell-mediated immunity

Idiopathic late-onset immunoglobulin deficiency is a disease which is highly variable in severity, age of onset, and pattern of immunoglobulin deficiency. It is regarded as a heterogeneous group of syndromes (Geha et al., 1974) and is designated 'common variable immunodeficiency' in the WHO classification. We describe a Southern Chinese boy who presented with recurrent respiratory infections and malabsorption and who has an associated defect in cell-mediated immunity.

Case report

The patient presented in January 1975 at the age of 8 years, after developing sudden weakness of the legs, preceded by a 2-month history of steatorrhoeic diarrhoea and fever for one week. Hypokalaemia was detected (serum K+ 2.2 mmol/l) and he was treated with Slow-K and referred to our hospital. His perinatal history was normal; he was breast fed and had full courses of BCG, DTP, and polio immunizations. Since early childhood he had had frequent upper and lower respiratory tract infections and episodes of loose stools. He developed uncomplicated measles when aged 7, after which he had persistent diarrhoea and productive cough. There was no family history of susceptibility to infection.

He was a small, thin boy (height 110-5 cm, weight 16.8 kg) with dry skin, clubbing of all digits, and small, discrete cervical and inguinal lymph nodes. Tonsils were of normal size. The chest was of normal shape with satisfactory expansion and occasional basal crepitations. The liver was palpable 1 cm and the spleen 2-3 cm below the costal margins (both were firm). Rectal examination was normal. The cardiovascular and nervous systems were normal.

Investigations. Hb 11.8 g/dl. White cell count 9850/mm³ (9.9 × 10⁹/l); differential count polymorphs 73%, lymphocytes 22%, monocytes 5%. Platelet count normal. Serum Na⁺ 139 mmol/l, K⁺ 2.9 mmol/l, alkaline phosphatase 160 μmol/min per l, bilirubin 10.3 μmol/l (0-6 mg/100 ml), SGOT 15 μmol/min per l, SGPT 13 μmol/min per l, albumin 40 g/l, globulin 17 g/l.

Serum immunoglobulin levels: IgG 2.78 g/l, IgM 0.13 g/l, IgA 0.37 g/l (all subnormal for age). Chest x-ray showed diffuse pulmonary mottling and peribronchial thickening. Bone age was 5-6 years.
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