it is not possible to draw any conclusions as to the mode of transmission of this disorder.

Polyorchidism, though rare, is a well recognized finding and triorchidism is the most common (Gould and Pyle, 1956). Familial polyorchidism has been reported, though most instances represent isolated cases.

Since little is known about this syndrome, it is difficult to know its prognosis. There is no adequate treatment for symphalangism, and the presence of an extra testis does not usually require surgical removal. The cause of his short stature is not known.

It is hoped that the recognition of this syndrome by others will clarify the many questions that remain unanswered.

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References

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Gastric adenocarcinoma due to ataxia-telangiectasia (Louis-Bar syndrome)

SUMMARY A 26-year-old male with ataxia-telangiectasia died with a gastric adenocarcinoma. Malignancy is a recognised complication of this condition, the majority of cases being reticuloendothelial. There have been three reports of gastric adenocarcinoma associated with ataxia-telangiectasia; this, however, is the first British published report.

Case report

A 26-year-old male presented with weight loss, anorexia, and dyspepsia. Ataxia-telangiectasia had...
been diagnosed in childhood. The disorder had also been recognised in 2 of 8 sibs, one of whom was being treated for acute leukaemia. His father, who smoked over 50 cigarettes per day, had recently died of carcinoma of the bronchus.

Examination showed telangiectasia of the conjunctivae and internal lining of the pinnae of the ears. Café-au-lait spots were present on the trunk. There was ataxia, myoclonus, and loss of lower limb reflexes with equivocal plantar responses. Central abdominal tenderness and melaena were detected.

Haemoglobin was 10 g/dl, the ESR 43 mm in the first hour. Immunoglobulin assay showed a slightly raised IgA (IgA 224 units/ml, normal 40 to 200 units/ml; IgG 110 units/ml, normal 92 to 200 units/ml; IgM 130 units/ml, normal 69 to 283 units/ml). The alpha-fetoprotein level was 435 µg/l (normal <7 µg/l). Chest x-ray showed small bilateral apical pneumothoraces and a barium meal examination showed a lesser curve gastric ulcer. Dysphagia developed and gastroscopy was prevented by oesophageal spasm. At laparotomy (Prof. Shields) extensive tumour infiltration of the greater omentum was found and histology showed an adenocarcinoma. The patient died five days later. Permission for necropsy examination was refused.

Chromosome studies were not performed on the patient or his family.

Discussion

Ataxia-telangiectasia is characterised by progressive cerebellar ataxia, and degenerative changes occur in various sites in the central and peripheral nervous systems. Telangiectasia develop on the conjunctivae and over areas exposed to friction. Insulin-resistant diabetes and disordered liver function may occur. Cell-mediated and humoral immunity are often impaired, and there is a high incidence of reduced or absent IgA, associated with sino-pulmonary infections (McFarlin et al., 1972).

A raised level of alpha-fetoprotein is an invariable finding (Waldmann and McIntire, 1972). Chromosome fractures have been observed and there are defective DNA repair mechanisms (Paterson et al., 1976).

The alteration in cell-mediated immunity and defective DNA repair mechanisms give rise to an increased incidence of malignant neoplasms and the majority of cases are reticuloendothelial (Peterson et al., 1966). Transmission of the condition is autosomal recessive and there is an increased incidence of neoplasia in heterozygotes (Swift et al., 1976). It is, therefore, significant that the father of the patient described here had recently died of a carcinoma of the bronchus.

Haerer et al. (1969) reported two sibs aged 19 and 21 years who developed gastric adenocarcinoma. They had reduced IgA levels. A further two cases with gastric adenocarcinoma were described by Kondo and Horikawa (1975), and a gastric neoplasm was found in a 14-year-old boy who had normal IgA (Watanabe et al., 1977).

This is a further report of gastric adenocarcinoma in a young person as a consequence of ataxia-telangiectasia, and it is therefore proposed that ataxia-telangiectasia be recognised as predisposing to gastric adenocarcinoma.

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References


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