Case reports

There have been 2 reports of sibs, one of whom had a defect in the diaphragm and the other a diaphragmatic hernia with a sac (Scott and Paterson, 1966; Thomas et al., 1976).

Diaphragmatic hernia has been reported in first cousins (Mosavy and Ashrafi, 1974) and in distant cousins (Turpin et al., 1959).

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Apical systolic click and murmur associated with neurofibromatosis

SUMMARY In this report we describe a child who had an apical systolic click and murmur, as well as widespread cutaneous neurofibromatosis. We were not able to show an anatomical basis for the click and murmur.

References


Fig. 1 Patient showing cutaneous lesions of neurofibromatosis.
Elliott et al. (1976) have reported a case of idiopathic hypertrophic subaortic stenosis associated with cutaneous neurofibromatosis. They suggest that there may be an hereditary defect of neuroectodermal development in the pathogenesis of idiopathic hypertrophic subaortic stenosis, lentiginosis, phaeochromocytoma, tuberous sclerosis, and neurofibromatosis.

Case report

A symptomless 11-year-old girl with no relevant family history was noted to have a cardiac murmur on routine school medical examination. Physical examination showed widespread cutaneous neurofibromatosis (Fig. 1). There was an apical mid to late systolic murmur grade 1 to 2/6 which varied with posture and Valsalva, preceded by 1 or sometimes 2 variable clicks (Fig. 2). The rest of the physical examination, chest x-ray, electrocardiogram, vectorcardiogram, and echocardiogram were all normal.

Discussion

Apical systolic clicks and murmurs have been described in association with a wide variety of conditions (Ehlers et al., 1970; Pickering, 1972; Aranda et al., 1975; Barlow and Pocock, 1975). These include localised cardiac lesions, both congenital (secundum atrial septal defect, ostium primum defect, persistent duc
tus arteriosus, and the Eisenmenger syndrome), and acquired (obstructive or congestive cardiomyopathies, coronary artery disease, trauma, myocarditis, postcardiac surgery, subvalvar left ventricular aneurysm, and atrial myxoma), and generalised diseases (Marfan syndrome, Turner's syndrome, rheumatic disease, and collagen disease). The associations between neuroectodermatoses and myocardial abnormalities are already known in tuberous sclerosis (Reed et al., 1963), the 'little leopard' syndrome (Gorlin et al., 1969; Pickering et al., 1971), and neurofibromatosis in which pulmonary valvar stenosis (Kaufman et al., 1972) and idiopathic hypertrophic subaortic stenosis (Elliott et al., 1976) have been described. It is postulated that a myocardial lesion may produce a contraction abnormality causing the apical systolic click and murmur. However, we did not feel that it was ethically justifiable to submit a symptom-free child to cardiac catheterisation and biopsy in an attempt to prove this hypothesis.

References


Fig. 2 Phonocardiogram showing systolic click and murmur.
Case reports


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Announcement

**NUTRITION AND GASTROENTEROLOGY SYMPOSIUM IN NEW YORK CITY**

The Institute of Human Nutrition at Columbia University is sponsoring a 2-day Symposium, involving experts from all over the world, on the subject of nutrition and gastroenterology at the Statler Hilton Hotel, Seventh Avenue, 32nd to 33rd Street, New York City, 30 November to 1 December 1978.

Dr Myron Winick, Director of the Institute of Human Nutrition, will serve as Chairman of the Symposium.

The purpose of the meeting is to examine the interrelation of nutrition and gastrointestinal function. The Symposium will cover basic research in gastrointestinal physiology under different nutritional conditions. In addition, it will explore the relation of nutrition to gastrointestinal diseases, and the effect of diseases of the gastrointestinal tract on nutritional status. Finally, nutritional therapy of patients with specific gastrointestinal diseases will be covered.

For further information write to the Director, Institute of Human Nutrition, Columbia University, 701 West 168th Street, New York, New York 10032, USA.