Subarachnoid haemorrhage in pseudoxanthoma elasticum

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Summary

A case of pseudoxanthoma elasticum presenting with the rare complication of subarachnoid haemorrhage is described.

Introduction

Pseudoxanthoma elasticum (PXE) is a rare heritable disorder of the connective tissue. Up to 1964, approximately 400 cases had been reported (Suerig and Siefert, 1964). The lesion involves mainly the skin, the eyes and the cardiovascular system, giving rise to diverse clinical manifestations. Most of the reports have highlighted the association of cutaneous changes with angioid streaks in the retina, gastro-intestinal haemorrhage and cardiovascular involvement. The neurological manifestations of PXE have not been reported as commonly. We report a patient with PXE who presented with subarachnoid haemorrhage.

Case report

A 35-year-old, single, Hindu male was admitted to the Goa Medical College Hospital, Panjim, on 29 April 1971, with the history of paralysis of the left side of the body of one day's duration. It was sudden in onset and was preceded by giddiness and intense headache. There was no history of unconsciousness, convulsions or fever. He did not complain of any skin lesions. There was no history of consanguinity of his parents. No other member of the family suffered from any obviously relevant illness.

On physical examination, the patient was moderately well nourished and ill-looking. The skin in the neck was lax, inelastic and thrown into folds (Fig. 1). There were small, soft, yellow papules distributed more or less parallel to the skin folds. Similar skin changes were also seen in the axillae, periumbilical area and groins (Fig. 2). Both the radial pulses and all the lower limb pulses were absent. The blood

pressure in both arms was 200/120 mmHg and was not recordable in the lower limbs.

On examination of the central nervous system, the patient was conscious. There was marked neck rigidity. The Kernig's sign was positive. There were signs of left hemiplegia. Fundus examination revealed bilateral macular degeneration, pigmentation around the right disc and chorio-retinitis on the left. There were no angioid streaks. Other systems were normal.

The routine urine examination was normal. Stools were positive for occult blood. The haemoglobin was 13 g/100 ml, WBC 14,000/mm³. Bleeding and clotting times were within normal limits. The serum cholesterol was 160 mg/100 ml, blood urea 24 mg/100 ml, and glucose 168 mg/100 ml. The blood VDRL test was negative. Lumbar puncture revealed a uniformly haemorrhagic cerebrospinal fluid and, on centrifuging, the supernatant fluid was xanthochromic. The cerebrospinal fluid biochemistry was within normal limits except for a slight increase in the protein content.

FIG. 1. The skin of the neck appears lax, inelastic and is thrown into folds.

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FIG. 2. The skin of the groin and peri-umbilical region shows lax inelastic folds which can easily be pinched.

FIG. 3. Lateral picture of right carotid angiogram. The tip of the needle is seen just inside the origin of the internal carotid artery. The artery is not visualized.

The electrocardiogram showed evidence of left ventricular hypertrophy. X-ray of the chest revealed calcification of the skin of the neck and axillae, and a normal heart and lungs. X-rays of the extremities did not show calcification of arteries. Bilateral

FIG. 4. Skin biopsy shows fragmentation of tissue in the middle dermis (haematoxylin and eosin $\times 100$).

carotid angiograms showed obliteration of the right carotid artery (Fig. 3) and generalized diminution in calibre of the left internal carotid artery. No aneurysms were seen.

When stained with haematoxylin and eosin, the skin biopsy specimen from the neck and axillae showed calcified fragments of tissue in the middle and lower dermis, a diagnostic feature of PXE (Fig. 4).

The patient made a good recovery, the cerebrospinal fluid returned to normal and he left the hospital, with residual hemiparesis.

Comment

PXE has a world-wide distribution. The disease is usually inherited as an autosomal dominant trait (Berlyne, Bulmer and Platt, 1961). There has been considerable controversy as to whether the basic disease process involves the elastic fibres or the collagen fibres. Recent reports have shown that the primary site of abnormality is the elastic tissue (Loria *et al.*, 1957; Smith, Sams and Davidson, 1962; Huang *et al.*, 1967).

In PXE, virtually every organ in the body can be affected (Loria *et al.*, 1957). Our patient had changes

mainly in the skin, cardiovascular and cerebrovascular systems. The cutaneous manifestations are small, soft and yellow papules arranged parallel to the skin lines and folds. In advanced cases the skin is thickened and hangs in loose, inelastic, redundant folds in the flexural areas, and has been described as 'plucked chicken-skin', 'crepe-like' skin, etc. Our patient had these advanced skin changes.

Cardiovascular abnormalities in PXE may be extensive and varied. Premature atherosclerosis in this disorder is due to elastic tissue degeneration and fragmentation in the media of the arteries leading to secondary deposition of calcium (Chong, 1970). These changes may result in weak or absent peripheral pulses and arterial insufficiency at various sites. Our patient had absent peripheral pulses, hypertension and evidence of left ventricular hypertrophy on the electrocardiogram. Hypertension is reported to be about three times more common in PXE than in the general population and is thought to be secondary to the involvement of renal glomeruli (Connor *et al.*, 1961).

Internal haemorrhage is a common manifestation of PXE and is attributed to vascular degeneration of the affected viscera. The commonest site of bleeding is the upper gastro-intestinal tract, and the rare sites of haemorrhage are the nose, the gums, the respiratory tract, the genito-urinary tract and the sub-arachnoid space. Eddy and Farber (1962) reported gastro-intestinal haemorrhage in 13% and bleeding from various other organs in only about 3% of the cases reviewed by them. In our patient, the stools were positive for occult blood; but the interesting feature was that he presented with subarachnoid

haemorrhage which is a rare manifestation in this disease. Bilateral carotid angiogram revealed generalized arterial involvement of the internal carotid system with complete obliteration of the right internal carotid artery but no obvious site of haemorrhage.

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