A CASE OF PSEUDOXANTHOMA ELASTICUM WITH RECURRENT HEMIPARESIA

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SUMMARY

Pseudoxanthoma elasticum is a genetic disorder. It most frequently involves the skin, eyes and arteries. The neurological manifestations of pseudoxanthoma elasticum are not frequent.

In this article a case of pseudoxanthoma elasticum with multiple cerebral infarcts is presented.

Key Words: Pseudoxanthoma elasticum, multiple cerebral infarction, recurrent hemiparesia.

INTRODUCTION

Pseudoxanthoma elasticum (PXE), also known as Groenblad-Strandberg Syndrome, is a rare, autosomal-recessive, heritable disorder of connective tissue characterized by the degeneration of the elastic tissue throughout the body, with secondary calcium deposition (1-4). In the disease there is a derangement in the metabolism and synthesis of the elastic fibres (1,5). The basic abnormality is fragmentation and calcification of elastic fibres in the skin and media of the arteries. Medium-sized arteries are the most severely affected arteries by degenerative atherosclerosis (4). The most prominent signs are chorioretinal lesions with so called angioid streaks, exudative maculer degeneration, yellowish efflorescenses of the skin named "pseudoxanthoma elasticum", cardiovascular and endocrine disorders (6,7).

The neurological complications of PXE is quiet rare. In this article a case with recurrent hemiparesia and multiple cerebral infarcts is presented.

CASE REPORT

A forty-eight-year old male was admitted to Neurology Clinic of Uludağ University Medical Faculty in August 1990. He was complaining of progression on the right-sided hemiparesis and difficulty in speaking. In his history there were attacks of hemiparesia in 1985 and 1988. The cranial computed tomographies revealed areas of cerebral infarction. The case had been considered as a case of cerebrovascular accidents and antiagregan therapy was advised.

In the physical examination: Temperature: 36.5 °C, pulse rate: 80/min, arterial tension: 200/110mmHg. His hair was somewhat blond. There were skin folds in the neck, cervical and inguinal area. The skin appeared lax and redundant. There were yellow - brown maculo - popular lesions in these areas.

In the neurological examination, the patient was alert and orianted. He had an expressive type of dysphasia. He had a right-sided spastic hemiparesis with a motor deficit about 60%. The deep tendon reflexes were brisk and Babinski sign was positive on the right side. Visual acuity and visual fields were normal. The examination of fundus oculi revealed angioid streaks parellel to the blood vessels of the retina (Fig. 1).

Laboratory investigations were normal except prebeta: 32.9 I.U. (8-24). The blood smear and long bone x-rays were normal. In the four vessel angiography of cerebral arteries the vascularity appeared poor. Multiple periventricular lacunar infarcts were noted on the cranial CT (Fig. 2). Dermal biopsy was obtained and at the medium and deep layers of the dermis the elastic fibres appeared edematous and irregular and there was calcium deposition on the elastic fibres (Figs. 3-4).

During the follow-up in the clinic, the patient's serum urea increased and the patient was diagnosed as to have acute renal failure. He had hemodialysis and the renal functions turned to normal gradually. The patient is still under regular clinical controls and has right-sided spastic hemiparesis.

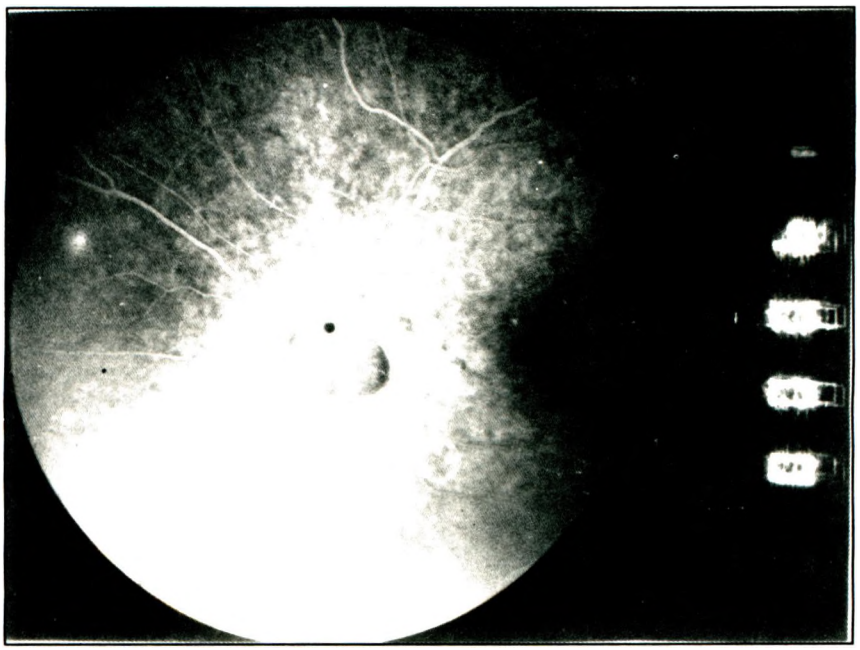

Fig 1. Angioid streaks of retina

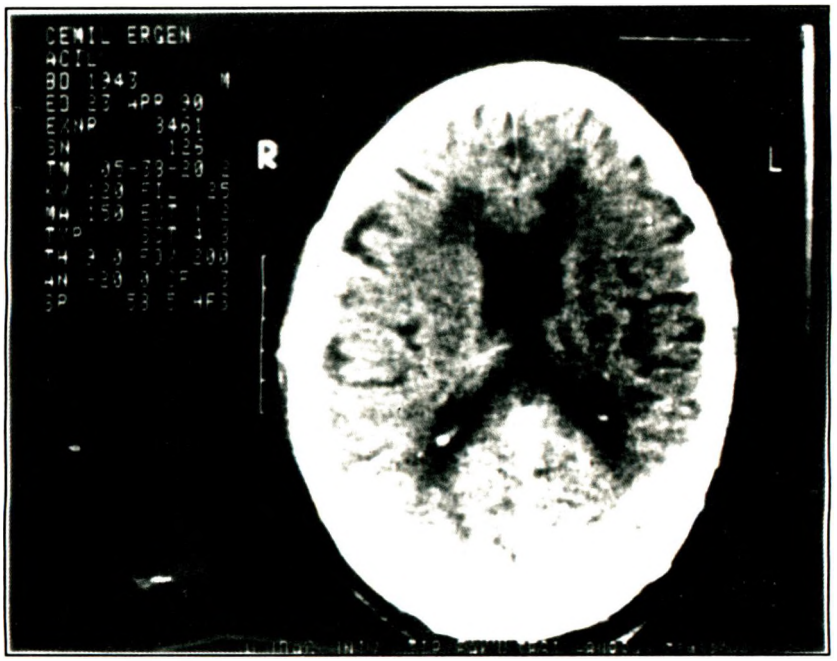

Fig 2. Cranial CT: Multiple infarcts

Fig 3. Note irregularity and swelling of the elastic fibers in medial and deeper layers of the dermis. Verhoeff's stain for elastic tissue.
10 x 25 / 0.65

Fig 4. Calcium deposition in elastic fibers in medial and deeper layers of the dermis. Van Kossa
10 x 40 / 0.70.

DISCUSSION

PXE is a rare disorder usually diagnosed in the second or third decade. The prevalence has been estimated as 1/70 000 and 1/160 000 in adults (4). PXE is a multisystem disease. Skin, mucous membranes, eyes, heart, peripheral blood vessels and gastrointestinal tract may be involved in this disease. But the basic alterations are in the skin, eyes and cardiovascular system (8,9). Dermal biopsy reveals fragmentation and degeneration of elastic fibres in the lower dermis associated with calcium deposition (3,7). Angioid streaks are characteristic and found in 85/% of the patients (2,7). In our case the diagnosis of PXE was first suspected when the angioid streaks were noticed during ophthalmological consultation .The angioid streaks can be seen in some cases of Paget's disease and sickle-cell anemia. In this case these two possibilities were eliminated through the blood smear, direct graphies and long bone x-rays.

Hypertension is three times more frequent in PXE comparing to normal population. Our patient has been receiving treatment for hypertension for five years (5).

When the literature about the PXE cases with neurological involvement is analysed only few cases are noticed. The most important vascular complication of PXE for central nervous system is cerebral aneursym with or without subarachnoid hemorrhage. Stenosis or occlusion of internal caroid and vertebral artery are other vascular complications. Kito et al reported a case of PXE with a ruptured anterior spinal artery aneursym (10).

The presented case was an original case of PXE with hypertension and recurrent attacks of hemiparesia and was found worth reporting.

REFERENCES

1. *Nishida H, Endo M, Koyanagi H, Ichihara T. Coronary artery bypass in a 15 - year - old girl with pseudoxanthoma elasticum. Ann Thorac Surg 1990;49:483-485.*
2. *Messis CP, Budzilovich GN. Pseudoxanthoma elasticum. Report of an autopsied case with cerebral involvement. Neurology 1990;20:703-708.*
3. *Rook A, Wilkinson DS, Ebling FJG, Champion RH. Pseudoxanthoma elasticum. Textbook of Dermatology. Oxford: Blackwell, 1986;1841-1844.*
4. *Belli A, Cawthorne S. Visceral angiographic findings in pseudoxanthoma elasticum. Br J Radiol 1988;61:768-771.*
5. *Mendelsonn G, Bulkley BH, Hutchins GM. Cardiovascular manifestations of pseudoxanthoma elasticum. Arch Pathol Lab Med 1978;102:298-302.*
6. *Fasshauer K, Reimers CD, Gnau HJ, Strempel I. Neurological complications of Grönblad-Strandberg syndrome. J Neurol 1984;231:250-252.*
7. *Schachner L, Young D, Bronx NY. Pseudoxanthoma elasticum with severe cardiovascular disease in a child. Am J Dis Child 1974;127:571-575.*
8. *Sharma NGK, Beohar PC, Ghosh SK, Gupta PS. Subarachnoid haemorrhage in pseudoxanthoma elasticum. Postgrad Med J. 1974;50:774-776.*
9. *Bete JM, Banas JS, Moran FJ, Pinn V, Levine HJ. Coronary artery disease in an 18 year old girl with pseudoxanthoma elasticum: Successful surgical therapy. Am J Cardiol 1975;36:515-520.*
10. *Kito K, Kobayashi N, Mori N, Kohno H. Ruptured aneurysm of the anterior spinal artery associated with pseudoxanthoma elasticum. Case report. J Neurosurgery 1983;58(1): 126-128.*