dystrophy, usually carried as a simple recessive character and exhibiting a slower clinical course starting in adolescence or early adult life. Some cases are apparently sporadic and pseudohypertrophy has been described. (3) Facioscapulohumeral dystrophy, a relatively non-disabling disease which is generally inherited in a simple dominant character. Facial involvement is fundamental. The disease is rarely carried as a recessive character. (4) Distal myopathy, involving the hands and the lower limbs below the knees. Middle-aged patients of either sex are found. The condition is often relatively benign and is commoner in Sweden than elsewhere. (5) Ocular myopathy, in which external ocular muscles and the eyelids are slowly affected. Some patients also develop dysphagia and, rarely, late limb involvement.

The congenital muscular dystrophies characteristically produce the 'floppy baby' syndrome. Myotonic dystrophy presents its own distinct clinical picture of myotonia, testicular atrophy, frontal balding and lens opacity in the adult. It has a dominant inheritance.

Most examples of all these groups may show progression but the degree of progression of dystrophic change seems less when onset is in the older age groups. The outlook is so varied that diagnosis within the spectrum is very important. Distinction between myopathies, polymyositides and the pseudomyopathies of a spinal muscular dystrophy rests on clinical and pathological findings. Often the clinical aspects are the most important. Endocrinological and metabolic disturbances have also to be considered. Muscle biopsy is often disappointing and has to be repeated before positive evidence is obtained. Electromyographic recordings may help to differentiate primary muscle disorders from muscle wasting secondary to nervous disease and to establish a diagnosis. Enzyme changes are not always reliable. Serum levels of phosphocreatine kinase, alanine transaminases, aspartate transaminase and aldolase, if raised, certainly confirm a clinical suspicion. Absence of raised serum levels of these enzymes does not in any way rule out the conditions. Cardiac involvement is found with frequency in the progressive forms of the dystrophies and ECG changes have included prolonged PR intervals, slurring of the QRS complexes and ST wave depression.

The case presented has some characteristics of the limb girdle disease and is very slowly progressive. In the absence of a definite family history one must assume that the causative gene is mutant. It is most unusual in showing complete heart block and it may be that this is a limb girdle dystrophy associated with a congenital heart block.

REFERENCE
Walton J N & Gardner-Medwin D
(1968) In: IV Symposium on Research on Muscular Dystrophy. Ed. J N Walton London; p 45

Postscript: Since the presentation of this report the patient has attended St George's Hospital, London, where a cardiac pacemaker was successfully fitted. His pulse rate is now 66/min.

Ehlers-Danlos Syndrome with Vertebral Artery Aneurysm

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Mr P D S, aged 28. Engineer from Goa

History: Bruised easily and wounds healed poorly as long as he could remember. Although there was no similar family history all five of his father's siblings died before middle age and his baby daughter, who was premature, exhibits hyperextensibility of the fingers. 1962: large spontaneous painful hæmatoma of his left thigh. March 1968: appendicectomy, complicated by pneumonitis, paralytic ileus and Gram-negative septicæmia. Ten days post-operatively 1,500 ml of infected hæmatoma was removed from his peritoneal cavity at laparotomy. Subsequently he complained of pain in his neck, which on two occasions was infiltrated, in the region of the occipital attachment of the left trapezius muscle, with hydrocortisone and lignocaine. Two days later a pulsatile swelling appeared in his neck, associated with tinnitus and headache.

He was transferred to the care of Professor G W Taylor when he was found to have a 6 cm tender left vertebral false aneurysm, confirmed by angiography, at the level of C2. He was also noted to have numerous thin hyperpigmented scars on his limbs and forehead, faintly blue sclera, and hyperextensibility of the joints of his hands. His skin was smooth, with little subcutaneous fat, and with very slight increase in elasticity.

Operation (8.7.68)*:* Ligation of the aneurysm was performed. The internal jugular vein was found to be extremely friable, 'like wet blotting paper' (McKusick 1966), failed to sustain suture, and had to be controlled by plugging the jugular foramen of the skull with oxidized cellulose. The vertebral artery was occluded with tantalum clips as ligature was too dangerous. He was transfused with 5 litres of blood.

Post-operatively he did well except for some palsy of the last three cranial nerves. Biopsy of forearm skin showed gross abnormality of elastic tissue, and the collagen appeared after heat denaturation to be more than normally resistant to peptic digestion. No defect in clotting factors was observed and X-rays of the skull, cervical and lumbar spines were normal.

The interest of this case is that he is thought to represent the so-called 'arterial' type of Ehlers-Danlos variant described by Barabas (1967). Recognition of this entity is extremely important in that, unlike in the classical form, surgery is extremely hazardous because of vessel fragility.

REFERENCES
Barabas A P (1967) *Brit. med. J.* ii, 612
McKusick V A (1966) Heritable Disorders of Connective Tissue. 3rd edition, St Louis; p 90

Mr A P Barabas (*Royal Postgraduate Medical School, London*) said that in 1967 he described 2 patients with clinical features similar to those of the case presented by Mr Edwards. At that time he regarded the condition as a subgroup of the Ehlers-Danlos syndrome (EDS) and termed it 'arterial' type. However, he now believed that this syndrome might be a distinct entity. In 1936 Sack reported the first possible example, and therefore his name could serve as an eponym. Mories (1960) published the first case report in this country, and Beighton (1968) found 4 cases amongst his 100 patients with EDS (he called them the 'ecchymotic' type).

Table 1
Clinical features in Sack's and in Ehlers-Danlos syndromes

	Sack's syndrome (*4 patients*)	*Ehlers-Danlos syndrome* (*27 patients*)
Skin	Thin, transparent, not hyperextensible	Hyperextensible
Bruising	Gross	Present
Joints	Minimal hypermobility	Usually gross hypermobility
Vascular complications	Ruptures of large arteries; traumatic a-v. fistulæ; false aneurysms	Nil
Abdominal complications	Attacks of severe pain; perforation of large bowel	Nil
Surgery	Extremely hazardous	Little difficulty
Affected relatives	All died of arterial or bowel wall ruptures	No serious complications; life span not affected

To date Mr Barabas had seen 4 unrelated patients with 'Sack's syndrome' and 27 patients with EDS. The clinical features in these two groups were summarized in Table 1.

REFERENCES
Beighton P (1968) *Brit. med. J.* iii, 656
Mories A (1960) *Scot. med. J.* 5, 269
Sack G (1936) *Dtsch. Arch. klin. Med.* 178, 663

Osteomalacia

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(for J R Nassim BM FRCP)
(*Royal National Orthopædic Hospital, Stanmore, Middlesex*)

Mrs V K, born 1910, of Russian Jewish origin
Presented in 1955 with pain in left foot and both legs. Found to have a fracture of the 3rd left metatarsal and pseudo-fractures of the left ilium (Fig 1), both femoral shafts and the 8th rib.

On examination: height 152 cm with normal proportions. Tenderness of the left foot and left ilium, otherwise normal.

Investigations: Serum calcium 9·1 mg/100 ml. Alkaline phosphatase 9 K-A units. Phosphate 1·9 mg/100 ml. Urine and amino acids normal. Control urinary calcium averaged 93 mg/24 hours over eighteen days, but balance was not satisfactory due to irregular fæcal collections. She was started on calciferol 10 mg daily and the urinary calcium rose to 120 and then 200 mg/24 hours in successive six-day periods. After her discharge she was maintained on 2·5 mg calciferol and 2 g sodium phosphate daily. There was slow healing of the pseudo-fractures and reduction in pain over the next two to three years. Serum inorganic phosphate rose to 3·5 mg/100 ml and alkaline phosphatase remained at or slightly above normal, maximum 18 units.

Later the serum phosphate fluctuated, often to below 2 mg/100 ml. It was felt that she was taking

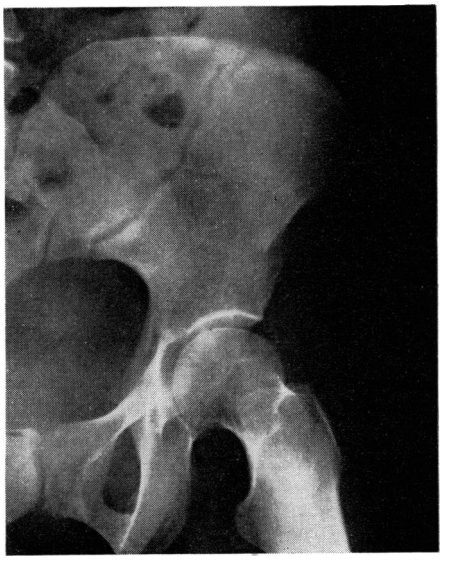

Fig 1 *13.12.55: Left ilium showing extensive pseudo-fracture*