Localised gigantism of the extremities

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LOCALISED GIGANTISM of the extremities may be caused by a number of distinct clinical entities. The literature abounds with descriptions of various conditions and the nomenclature is confusing. This paper presents some illustrative examples of patients suffering from localised gigantism of the extremities and an attempt is made to propose a practical classification.

Illustrative cases

Elephantiasis Neurofibromatosa

Case No. 1: This is a 14-year-old Chinese girl who presented with a longer and larger left leg since childhood. On examination, she was found to have typical cafe au lait spots and multiple neurofibromatosa over her body. The left leg was larger and longer below the knee. X-rays of this leg show obvious soft tissue enlargement and lengthening and widening of the left tibia (Fig. 1). She elected to have no treatment and was lost to follow up.

Macrodystrophia lipomatosa

Case No. 2: A 84-year-old Chinese female who presented with a larger and longer right leg and a grotesque right foot since childhood. She gave a history of recurrent ulcerations over the dorsum of the left foot. There was no positive family history.

On examination, the right leg was longer and broader both in the thigh and leg. The right foot was grotesquely enlarged and there were two ulcers over the dorsum of this foot (Fig. 2). In addition, there were multiple sessile subcutaneous lipomata scattered especially in the leg with a much larger oblong lump just below the groin.

Fig. 1
Elephantiasis Neurofibromatosa: X-ray showing the left tibia and fibula to be longer and broader. There is also soft tissue enlargement.

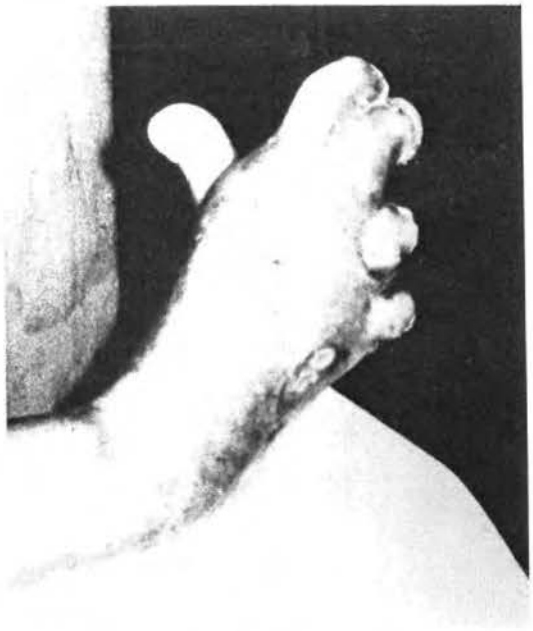

Fig. 2
Macrodystrophia lipomatosa: The grotesque enlargement of right foot and toes.

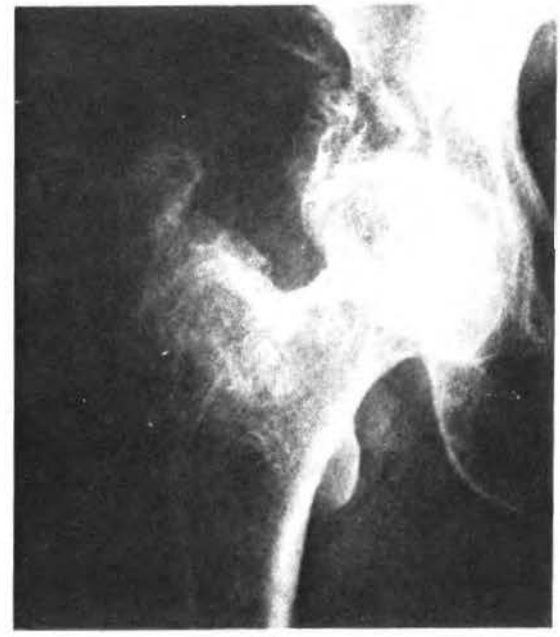

Fig. 3
Macrodystrophia lipomatosa: Exostosis over the right greater trochanter.

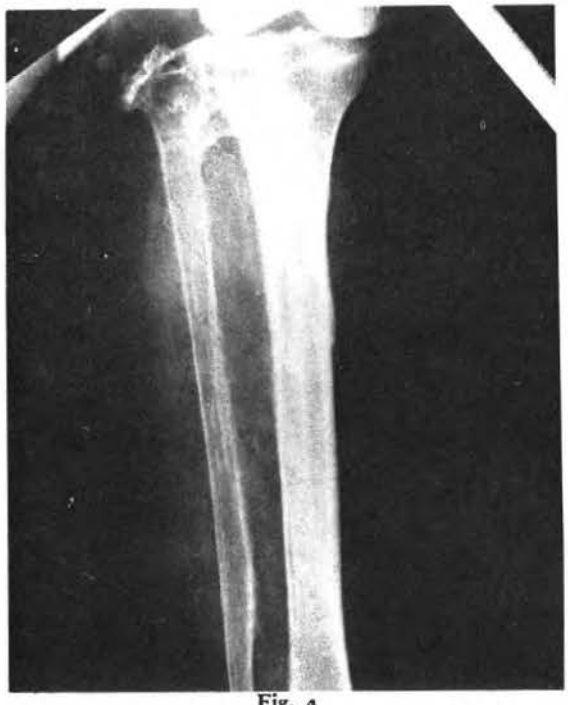

Fig. 4
Macrodystropnia lipomatosa: Exostosis over the head of fibula. Notice periosteal reaction and broadening of the tibia and fibula.

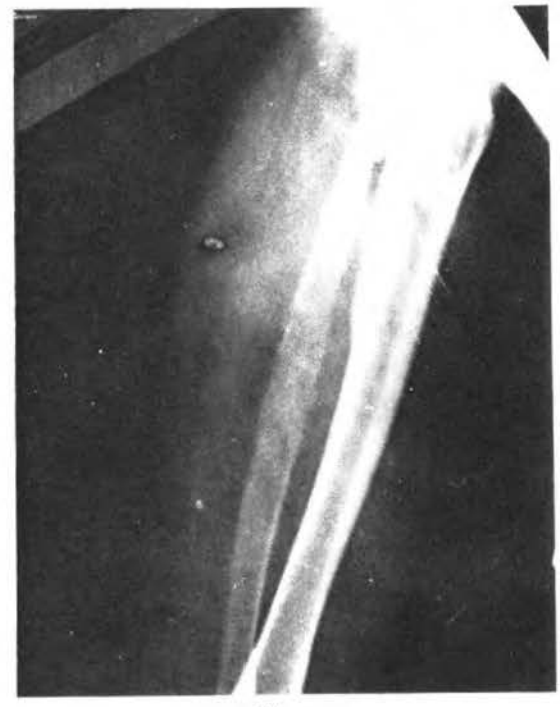

Fig. 5
Macrodystrophia lipomatosa: Broadening of the tibia and fibula.

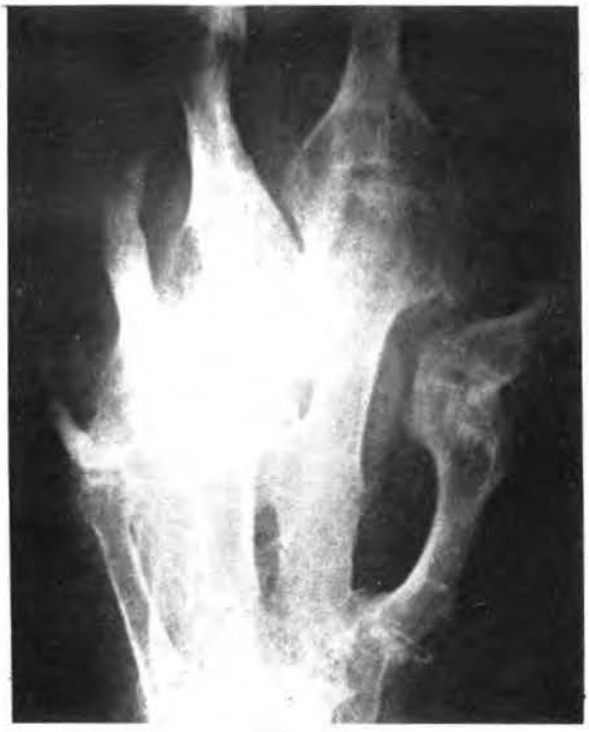

Fig. 6
Macrodystrophia lipomatosa: Antero-posterior X-ray of right foot showing enlargement, ankylosis and arthropathy.

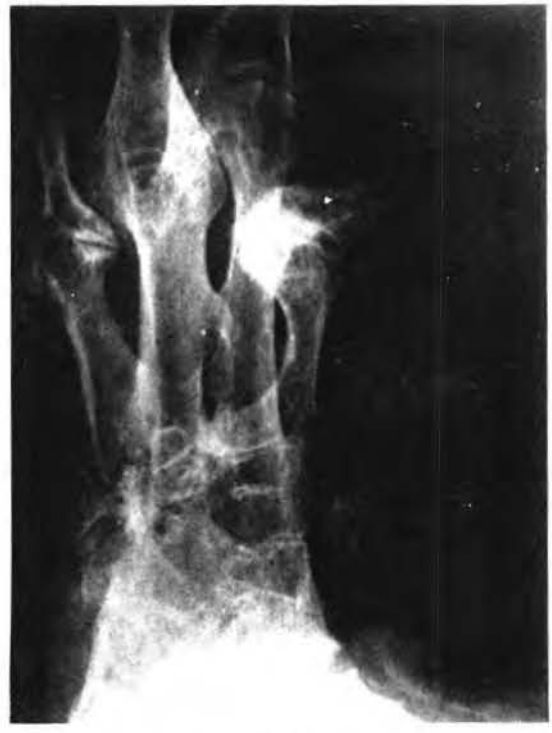

Fig. 7
Macrodystrophia lipomatosa: Lateral X-ray showing some features as Figure 6.

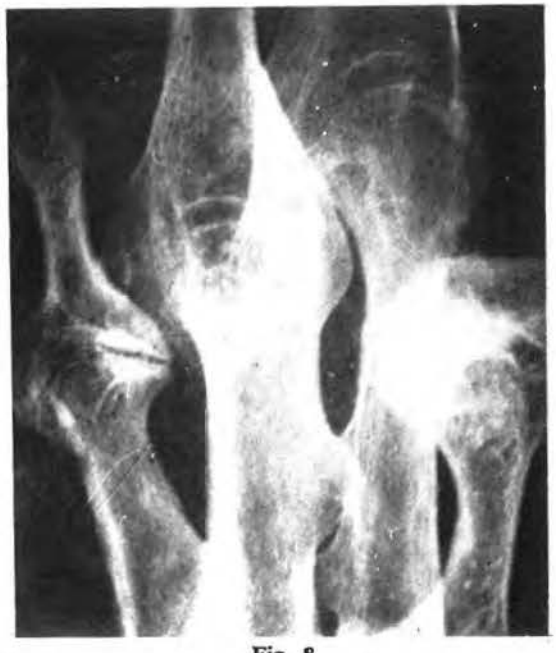

Fig. 8
Macrodystrophia lipomatosa: Magnified view showing the arthropathy.

Skeletal surveys revealed exostosis in the region of the right greater trochanter (Fig. 3) and the right upper fibula (Fig. 4); broadening of the right tibia with periosteal reaction (Figs. 4 & 5); bony ankylosis and arthropathy in the right foot (Figs. 6, 7 & 8). X-rays of the left leg and foot showed no abnormality.

Klippel-Trenaunay Syndrome

Case No. 3: A six-year-old Chinese boy, who presented with overgrowth of the left thumb. According to the parents, this was noticed in infancy and gradually increased in size.

On examination, the left thumb was twice as large as the right thumb. The web between the left thumb and index finger was occupied by a large firm swelling and on auscultation a definite bruit was heard. X-rays are reproduced in figure 9.

The enlargement clearly involves soft tissues as well as bone. A clinical diagnosis of haemangioma was made. Unfortunately, the parents refused surgery and thus no histological confirmation was possible.

Case No. 4: A 14-year-old Chinese girl, who pre-

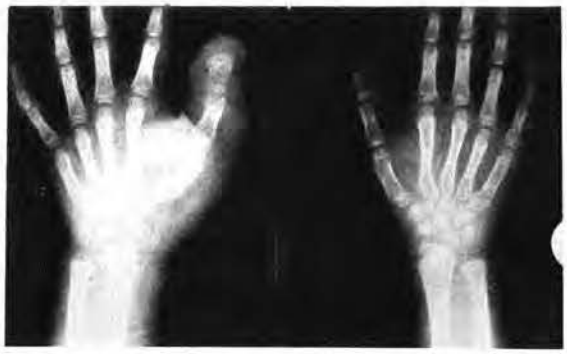

Fig. 9
Klippel–Trenaunay Syndrome: Radiograph showing enlargement of the left thumb — soft tissue and bone.

sented with a larger and longer right leg (figs. 10 & 11). The right leg was noticed to be definitely larger than the left since the age of one month and has been enlarging progressively. There was associated pink discoloration of this leg and over the last years this has assumed a blue black character. In addition, this leg had a tendency to bleed on the slightest trauma but there was no history of a generalised bleeding diasthesis. There was no similar disorder in the parents or her siblings.

On examination, the right leg was lengthened by two inches and the girth was also larger — being three inches bigger in the calf. There was a peculiar blue black discoloration of the leg and nodules of thrombosed vessels could be felt. There was no palpable thrill or audible bruit but there was definite increase of local temperature.

On skeletal survey, a few areas of shallow notchings were noted in the long bones (Fig. 12).

Fig 10
Klippel – Trenaunay Syndrome: Haemangiomatosis. Larger and longer right leg. Notice the discoloration and the irregular nodules.

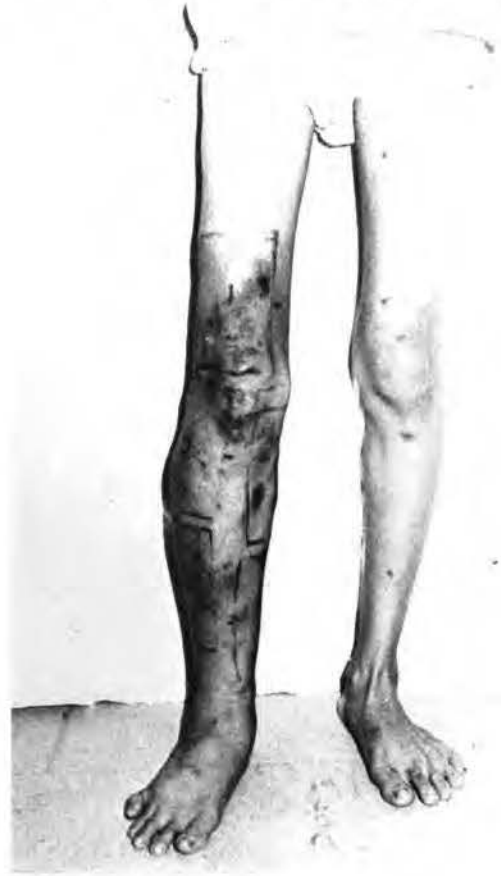

Fig. 11
Klippel – Trenaunay Syndrome: Haemangiomatosis. Close-up view showing the discoloration and nodules.

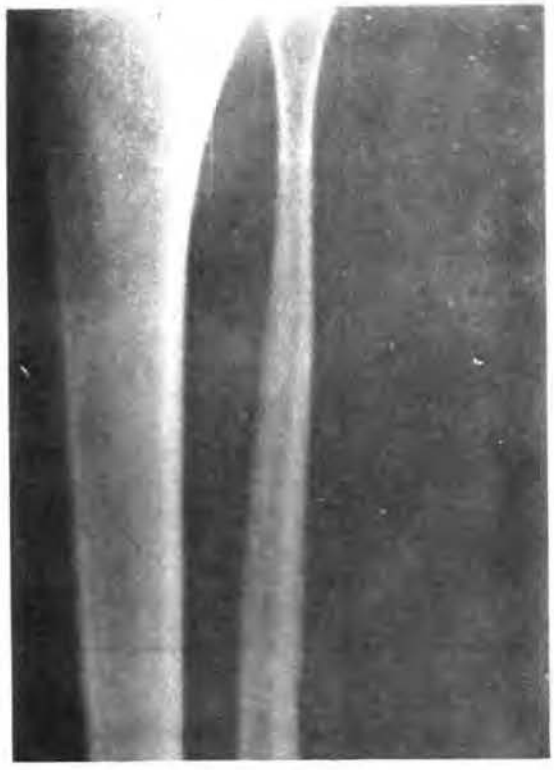

Fig. 12
Klippel – Trenaunay Syndrome: Haemangiomatosis. Radiography showing superficial notches in the fibula.

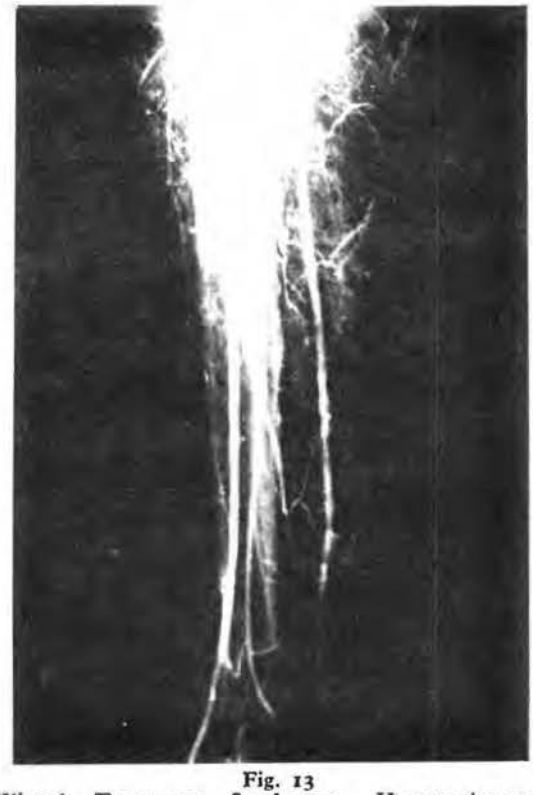

Fig. 13
Klippel – Trenaunay Syndrome: Haemangiomatosis. Arteriogram showing marked vascularity and numerous capillaries coursing around the soft tissues.

Arteriography was carried out and on injecting the femoral artery, there was almost instant filling of the femoral vein. In addition, there was marked vascularity of the leg with numerous capillaries coursing around the soft tissues. No distinct arteriovenous fistulae were demonstrated (Figs. 13 & 14). This picture is consistent with generalised haemangiomatosis. After much debate, she was given a course of radiotherapy with moderately good response.

Case No. 5: A 10-year-old Malay girl, who presented with a longer and larger right leg and was noticed to limp ever since she started walking.

On examination, the whole right leg from the thigh downwards was grossly enlarged. The right hip was obviously dislocated. There was diffuse blue black and pinkish discoloration over almost the whole limb. There were also numerous varicosities and the whole limb felt very much warmer. On auscultation, definite bruit was heard over various parts of the limb.

Radiological examination confirmed the dislocated right hip (Fig. 15). The femur and tibia and fibula were long and fragile looking (Figs. 16 & 17) and in Figure 17, scattered streaks of opacity could be seen in the region of the tibia with one of these overlying a notch in the tibia. In addition, there was pathological fracture of the lower tibia and fibula.

The clinical diagnosis was unilaternal gigantism due to congenital arterioverous fistulae. Unfortunately, the parents did not agree to limb ablation or further investigation. Hence there is no histological or angiographic confirmation.

Discussion

A practical classification for localised gigantism of the extremities is proposed (Table I). The term dysplasia is used to denote disturbed development and growth. From the pathological point of view hamartomatous malformation could be a more accurate term. However, this is not only clumsy but the word hamartoma invites confused connotations. The classification proposed is not meant to displace all others but we feel that it is practical and also stresses the basic lesion in each group.

Fig. 14
Klippel – Trenaunay Syndrome: Haemangiomatosis. Close-up view of arteriogram.

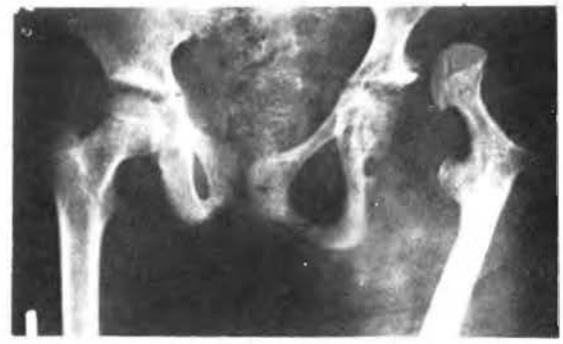

Fig. 15
Klippel-Trenaunay Syndrome: Congenital A.V. fistulae — grossly enlarged right leg with pathological dislocation of right hip.

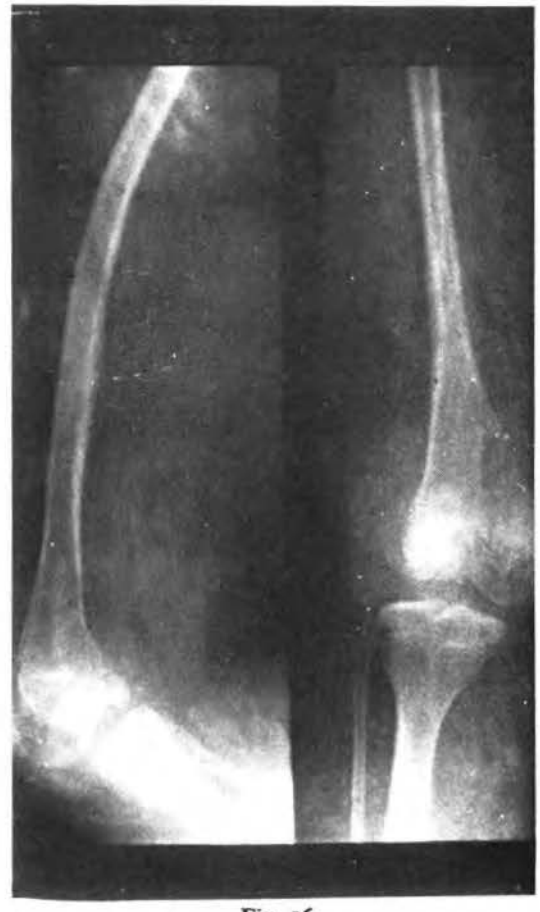

Fig. 16
Klippel-Trenaunay Syndrome: Congenital A.V. fistulae — fragile looking femur with marked soft tissue swelling Streaks of calcification in soft tissues.

Table I

1. **Congenital Dysplasias**
   - (a) **All elements of connective tissue affected**
     - (i) Congenital Hemihypertrophy
     - (ii) Congenital Macrodactyly
   - (b) **One element of connective tissue predominantly affected**
     - (i) Elephantiasis neurofibromatosa
     - (ii) Macrodystrophia lipomatosa
     - (iii) Klippel-Trenaunay Syndrome
       - — haemangiomatosis
       - — A.V. fistulae
     - (iv) Congenital lymphodema
     - (v) Bone dysplasia
       - — Olliers disease
       - — Melorheostosis
2. **Acquired conditions**
   - Lymphatic Blockage
     - — Filarial elephantiasis
     - — Brawny Arm of Cancer Breast
     - — Others

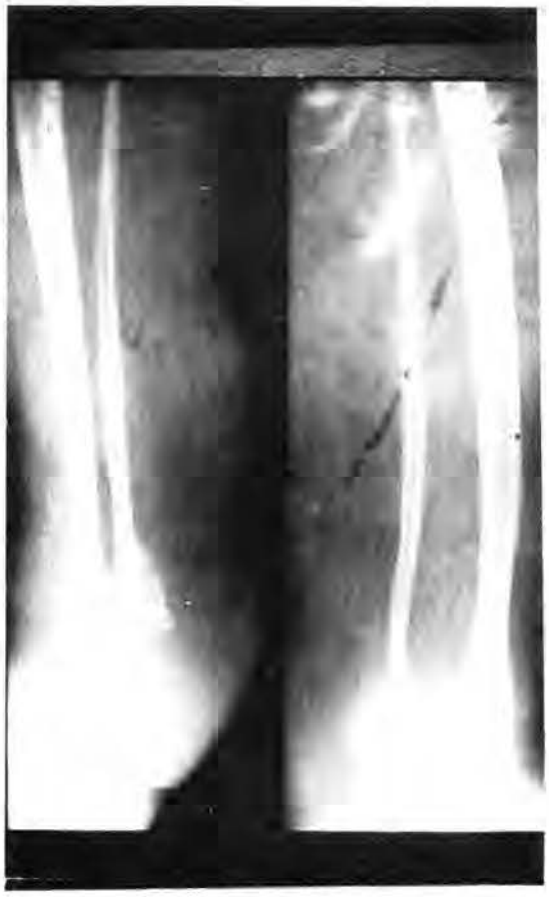

Fig. 17
Klippel–Trenaunay Syndrome: Congenital A.V. fistulae — fragile looking tibia and fibula with pathological fracture dislocation of the ankle. Streaks of calcification in the soft tissue and one of these overlies a notch in the upper tibia.

Congenital hemihypertrophy is a developmental condition where there is hypertrophy of all the elements of the connective tissue. A further distinguishing feature is that the hypertrophy involves the head and trunk in addition to the extremities.

In congenital macrodactyly, there is also hypertrophy of all the elements of connective tisue, but in this condition the enlargement involves only the digits.

Elephantiasis neurofibromatosa is known to be associated with plexiform neuromata related to a particular nerve. The radiology of the condition is well reviewed by Hunt and Pugh 1961 and Edeiken and Hodes 1967. **Case No. 1** shows features typical of the disease but no histological confirmation was available.

Macrodystrophia lipomatosa is a term coined by Werthemann in 1952, although the condition was first described by Oosthuizen et al in 1947 as "lipomatosis involving bone". **Case No. 2** shows all the typical clinical and radiological features of the disease.

The Klippel-Trenaunay Syndrome comprises a group of conditions where local gigantism results from congenital angiomatous malformations. **Case 3 and** 4 represent manifestations of haemangiomatosis. In **Case 5** the most likely basic lesion is congenital arteriovenous fistulae although no pathological or angiographic confirmation was available.

Summary

A simple scheme is proposed to classify local gigantism in extremities based on the predominant basic lesion.

Five personal cases are described to illustrate gigantism associated with neurofibromatosis, lipomatous, haemangiomatosis and congenital arteriovenous fistulae.

Acknowledgement

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