Skeletal dysplasia

By Assistant Professor

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defintion

Abnormal development of the musculoskeletal system may give rise to a variety of **physical** defects ,These defects categorized to:

skeletal dysplasias (abnormal bone growth and/or modelling),

malformations (e.g. absence or duplication of certain parts)

structural defects of connective tissue;

metabolic abnormality

main causes of the abnormality is a **genetic defect**. and this gene defect broadly divided into three categories:

chromosome disorders,
single gene and polygenic disorders
multifactorial disorders.

A>>Chromosome disorders
Additions, deletions or changes in chromosomal structure e.g. Down's syndrome in which there is one extra chromosome 21 (trisomy 21).
Unlike genetic disorders, these conditions are **not transmitted in future generations**.

B>>gene disorders Gene mutation may occur by insertion, deletion, substitution or fusion in the DNA chain.
result in cartilage growth, collagen structure, matrix patterning and marrow cell metabolism.
The abnormality is then **passed on to future generations** according to **simple mendelian rules**

C>>Embryonal damage result from injury to the developing embryo. Most are of unknown aetiology but some are due to specific viral infections (e.g. rubella), certain drugs (e.g. thalidomide) and ionizing radiation

Patterns of inheritance:

. Autosomal **dominant** transmission
. Autosomal **recessive** transmission
. X-linked **dominant** disorders
. X-linked **recessive** disorders

Diagnosis

Prenatal diagnosis:

High-resolution ultrasound imaging amniocentesis or chorionic villus sampling

Diagnosis in childhood

- family history
- Physical abnormalities may be obvious at birth
- X-ray examination
- Special investigations specific enzyme or metabolic abnormalities.
- Direct testing for gene mutations

Principles of management:

It is best treated in a centre that offers a **'special interest' team** consisting of a

- paediatrician,
- medical geneticist,
- **orthopaedic surgeon,**
- psychologist,
- social worker,
- occupational therapist,
- orthotist prosthetist

Types of mangemnt..

A__Counselling Patients and families may need expert counselling about: (a) the likely outcome of the disorders; (b) what will be required of the family; and (c) the risk of siblings or children being affected.

.B__ Intrauterine surgery The concept of operating on the unborn fetus is already a reality

.C__ Specific medication One example is the treatment of Gaucher's disease by administering the missing enzyme, alglucerase.

D__ Gene therapy is still at the experimental stage

E__Prevention and correction of deformities Realignment of the limb, correction of ligamentous laxity and joint reconstruction can improve joint stability and gait.

Classifcation:-

A practical grouping of generalized developmental disorders

Genetic disorders of cartilage and bone growth (chondro-osteodystrophies)

A_Dysplasias with predominantly epiphyseal changes
Multiple epiphyseal dysplasia
Spondyloepiphyseal dysplasia

B- Dysplasias with predominantly physeal and metaphyseal changes
Hereditary multiple exostosis
Achondroplasia

C -Dysplasias with predominantly diaphyseal changes
Osteopetrosis (marble bones)

2- Collagen disorders

Osteogenesis imperfecta (brittle bones)
Ehlers–Danlos syndrome

3. Enzyme defects and metabolic disorders

Mucopolysaccharidoses
Gaucher's disease

4. Chromosome disorders

Down's syndrome

Dysplasias with predominantly epiphyseal changes

Multiple epiphyseal dysplasia

familial disorder (autosomal dominant in most cases) in which the long-bone epiphyses develop abnormally..

Clinical features

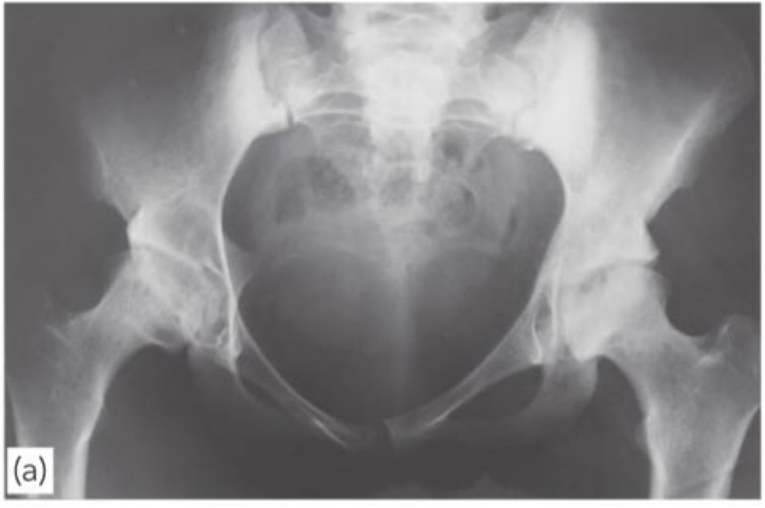
Children may present with **stunted growth** or with **joint pain** and progressive deformity.
The *face, skull and spine* ***are normal***.

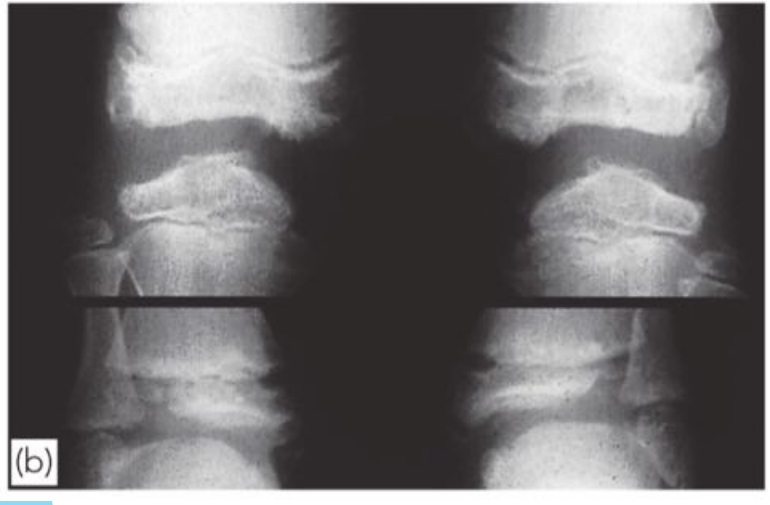
In adult life, residual bone defects may lead to **joint incongruity and secondary osteoarthritis.**

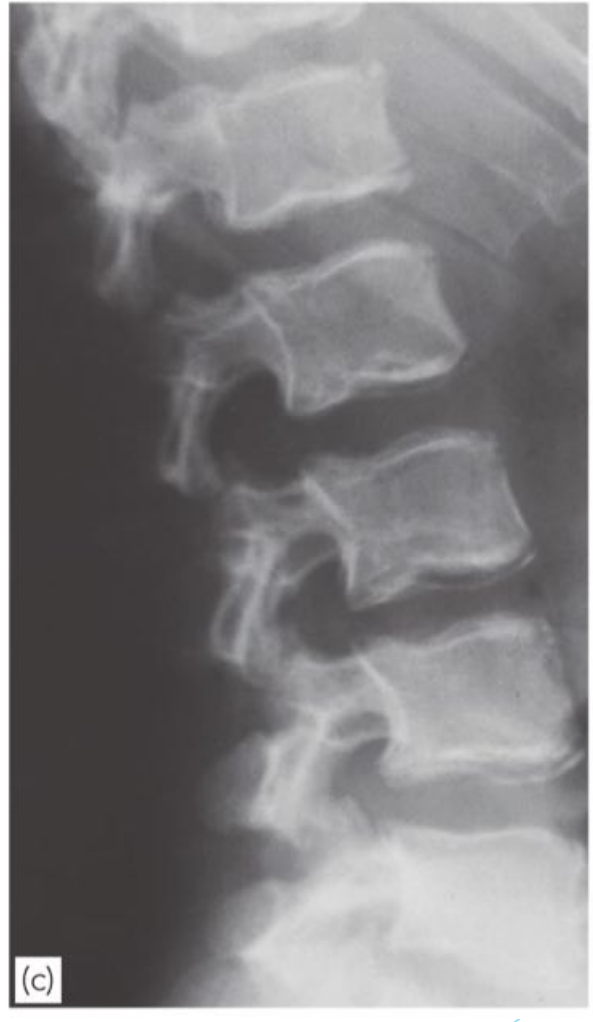
widespread involvement of the epiphyses but the **vertebrae are not at all, or only mildly, affected.**

Management

Children may complain of slight pain and limp, but little can (or need) be done about this.
At maturity, deformities around the hips, knees or ankles sometimes require corrective osteotomy.
In later life, secondary osteoarthritis may call for reconstructive surgery.







8.1 Multiple epiphyseal dysplasia (a,b) X-rays show epiphyseal distortion and flattening at multiple sites, in this case the hips, knees and ankles. (c) The ring epiphyses of the vertebral bodies also may be affected and in spondyloepiphyseal dysplasia this is the dominant feature.

Spondyloepiphyseal dysplasia

Spondyloepiphyseal dyplasia (SED) comprises a heterogeneous group of disorders in which **MED is associated with well-marked vertebral changes**:
delayed ossification,
flattening of the vertebral bodies
irregular ossification of the ring epiphyses and indentations of the end-plates.
▪ Retinal detachment and respiratory problems are common.

Classifiction:

A__SED congenita:

This autosomal dominant disorder can be diagnosed in infancy

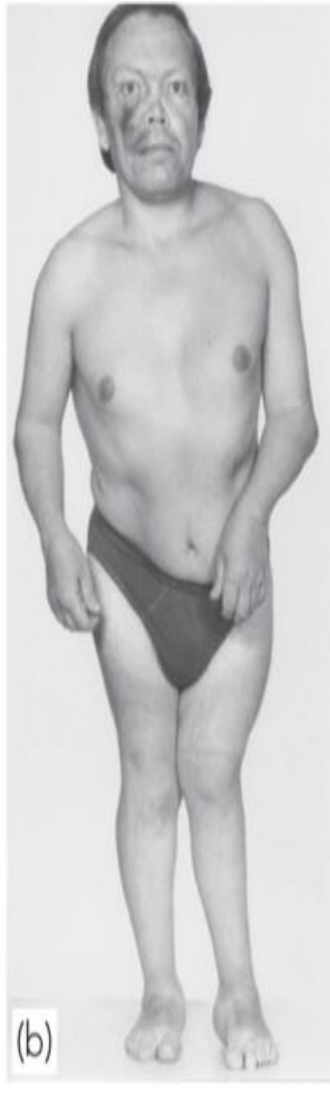
B__SED tarda:

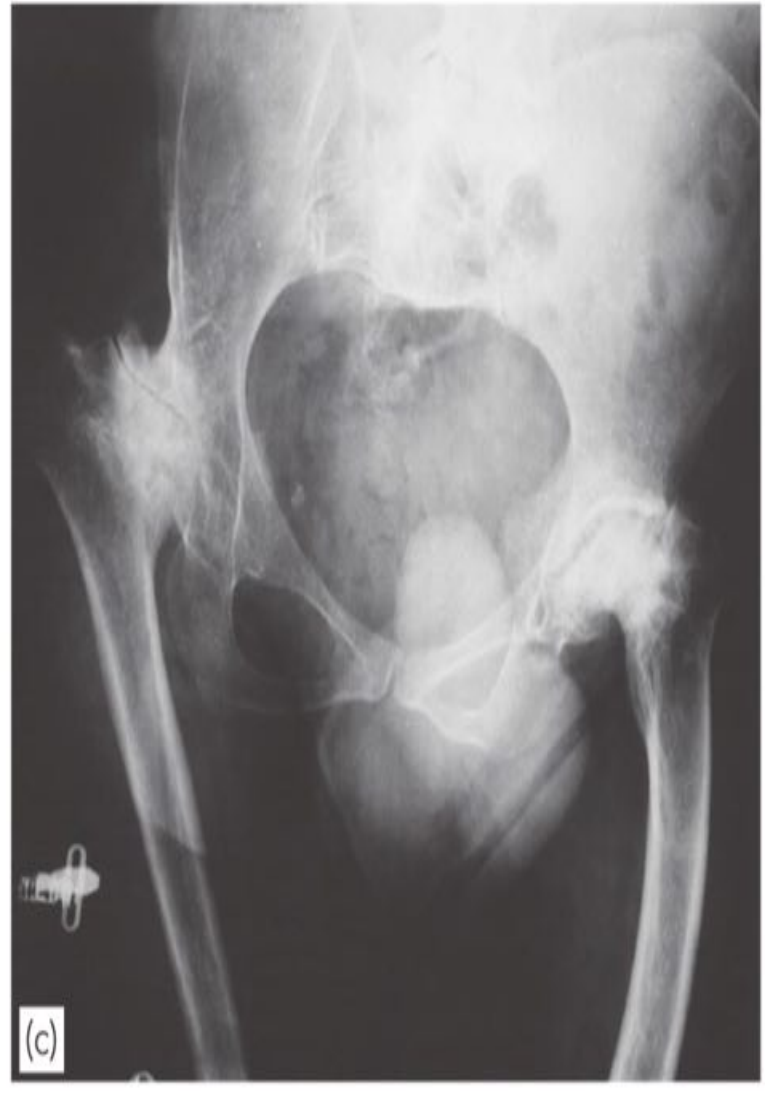
an X-linked recessive disorder,
is less common and less severe than SED congenita,
usully becoming apparent only after the age of 5 years when the child fails to grow normally and develops a **kyphoscoliosis**

Management

may involve corrective osteotomies for severe coxa vara or knee deformities.

Odontoid
hypoplasia increases the risks of anaesthesia





8.2 Spondyloepiphyseal dysplasia The x-ray features are similar to those in multiple epiphyseal dysplasia, but are usually more marked and affecting the vertebrae as well. (a,b) As adults these patients may be severely deformed. Note the barrel chest, deformity of all limbs and severe changes in the pelvic x-ray (c).

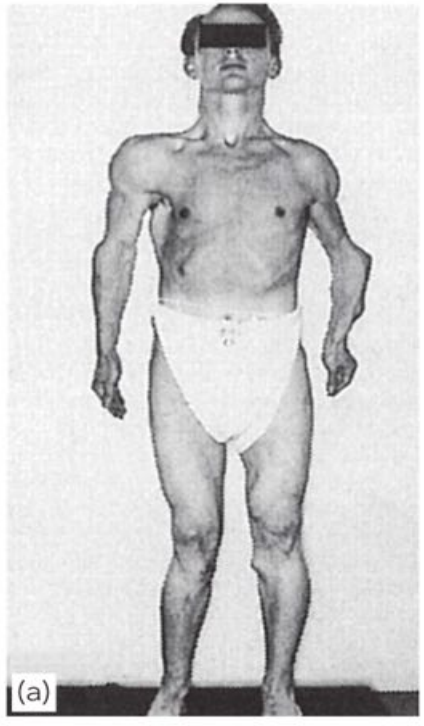
Dysplasias with predominantly physeal and metaphyseal changes

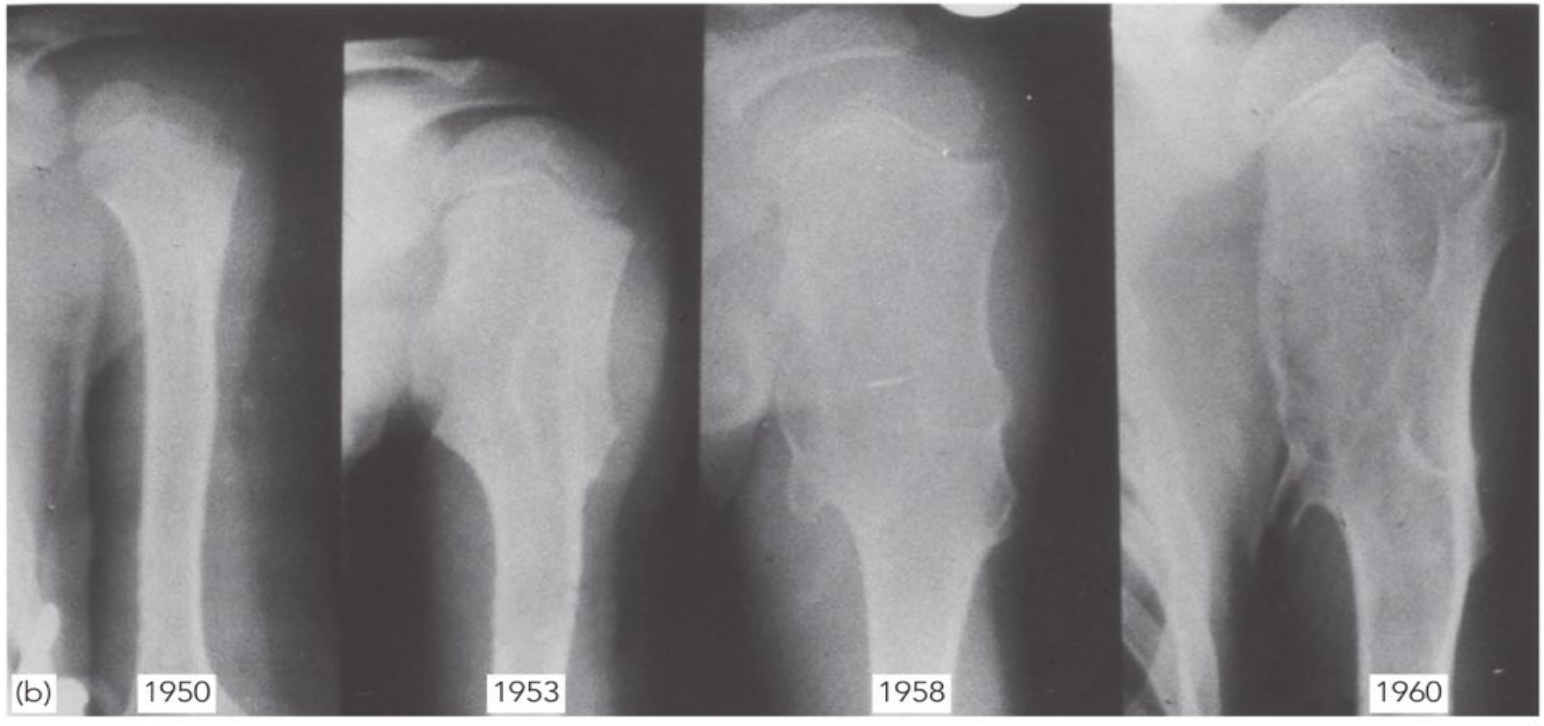
Hereditary multiple exostosis (diaphyseal aclasis)

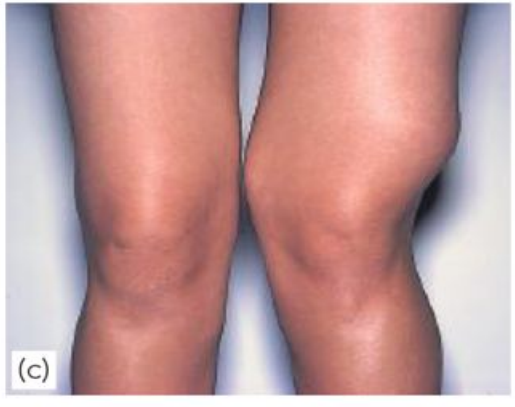
This, the most common of all skeletal dysplasias autosomal dominant congenital disorder in which **multiple exostoses** appear at the long-bone metaphyses and the apophyseal borders of the scapula and pelvis.

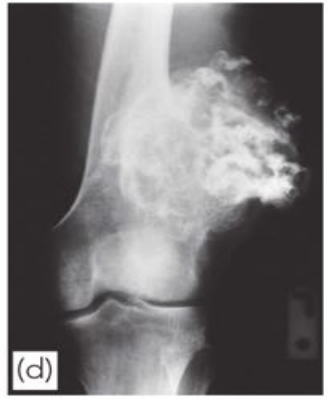
X-rays the pathognomonic exostoses as well as broadening and imperfect modelling of the metaphyses.

Management If an exostosis is troublesome (and certainly if it starts to 'grow' after the parent bone has stopped) it should be removed.
Long-bone deformities may call for corrective osteotomy









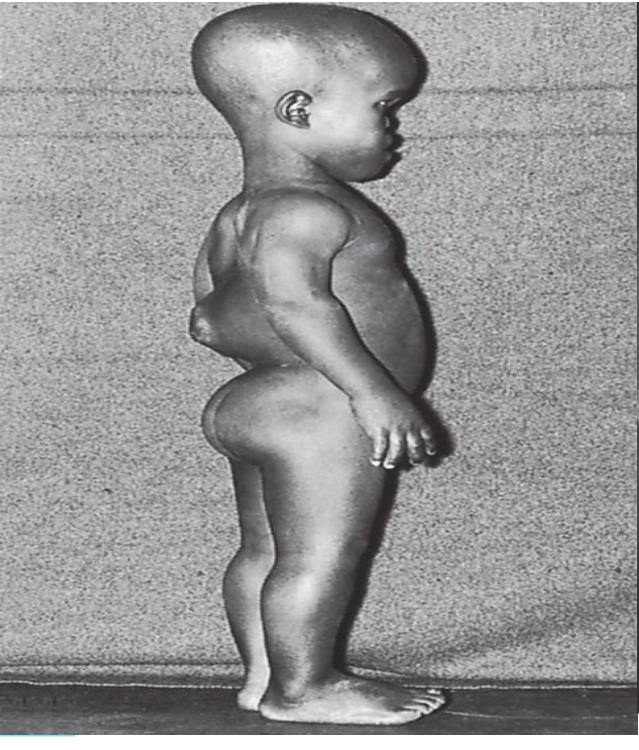
8.3 Hereditary multiple exostosis (a) Adult with multiple exostoses and typical deformities of the arms and legs. Note the numerous small 'bumps' in the limb bones, bowing of the left radius, shortening of the left forearm and valgus deformity of the right knee. (b) Serial x-rays showing the evolution of the shortened left humerus and the wide metaphysis during growth. (c,d) Patient with a large lump above the knee due to a cartilage-capped exostosis.

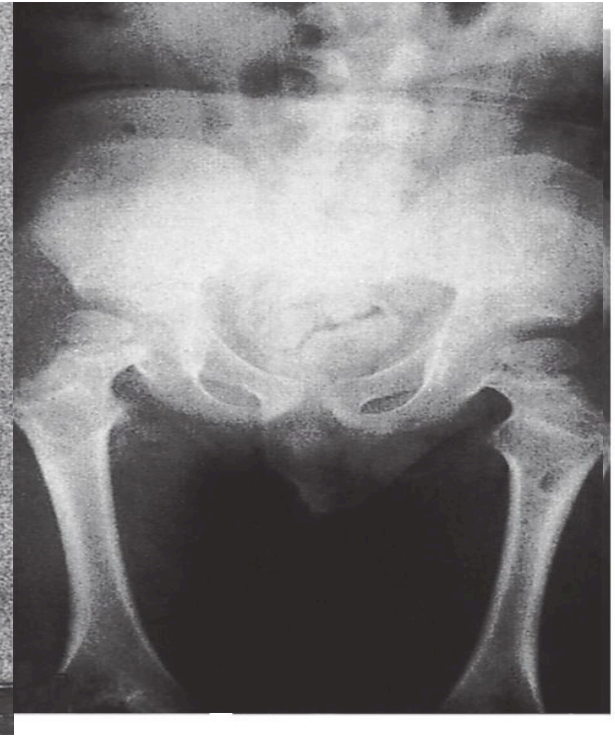
Achondroplasia (Dwarfism)

This is the classic example of chondrodysplasia, commonest form of abnormally short stature with autosomal dominant trait.

Management

During childhood, operative treatment may be needed for lower limb deformities (usually genu varum).





8.4 Achondroplasia
(a) Mother and child with achondroplasia, showing the typical disproportionate shortening of the tubular bones, particularly the proximal segments of the upper and lower limbs. (b) Other features are seen in this child: lumbar lordosis, a prominent thoracolumbar gibbus and bossing of the forehead. (c) X-ray showing the flat pelvis and short femora.

Dysplasias with mainly diaphyseal changes

Most of the '**diaphyseal dysplasias**' appear to be the result of defective bone modelling.
Unlike the physeal and epiphyseal disorders, **dwarfing is not a feature**

Osteopetrosis (marble bones, Albers–Schönberg disease)

one of several conditions that are characterized by sclerosis and thickening of the bones which then appear unusually 'dense' on x-ray.

types

Osteopetrosis congenita

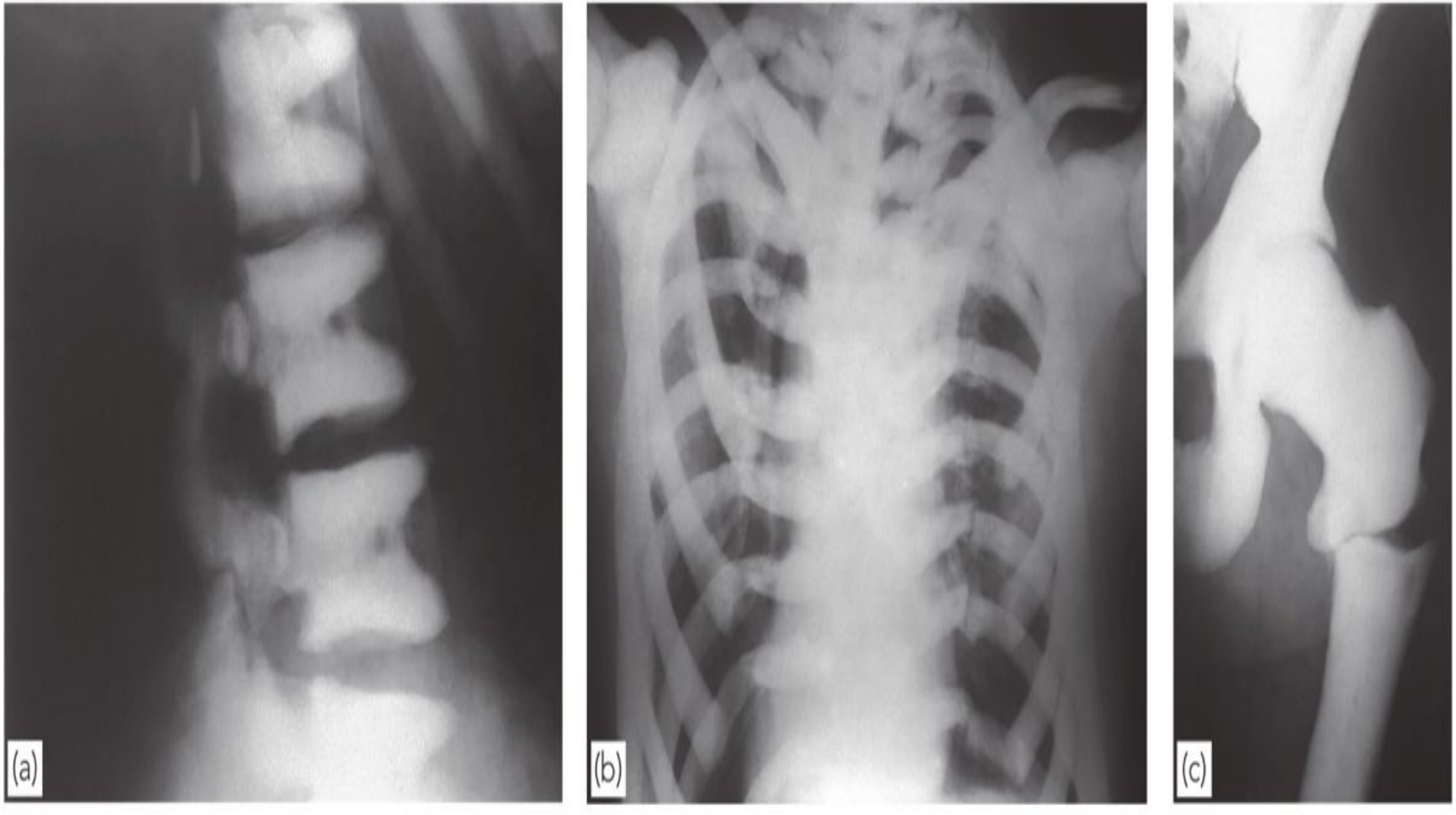
rare, autosomal recessive form of osteopetrosis is present **at birth** and causes severe disability.
Bone encroachment on marrow results in pancytopenia, haemolysis, anaemia and hepatosplenomegaly

Treatment has focused on methods of **enhancing bone resorption and haematopoeisis**, e.g. by transplanting marrow from normal donors and by long-term treatment with gamma-interferon.

Osteopetrosis tarda

may only be discovered in adolescence or adulthood– hence the designation tarda

common form of osteopetrosis is a fairly benign,
autosomal dominant disorder that seldom causes symptoms.
Shape and function are unimpaired



8.6 Osteopetrosis. (a–c) Despite the remarkable density, the bones fracture easily. Typical x-ray features in the spine

Connective-tissue disorders

Heritable defects of collagen synthesis give rise to a number of disorders involving either the ***soft connective tissues or bone, or both.***

Ehlers-Danlos syndrome (EDS)

heterogeneous group of connective tissue disorders characterized by **joint hypermobility**, ***skin laxity*** and (in some cases) blood **vessel fragility**.
autosomal dominant inheritance.

Management

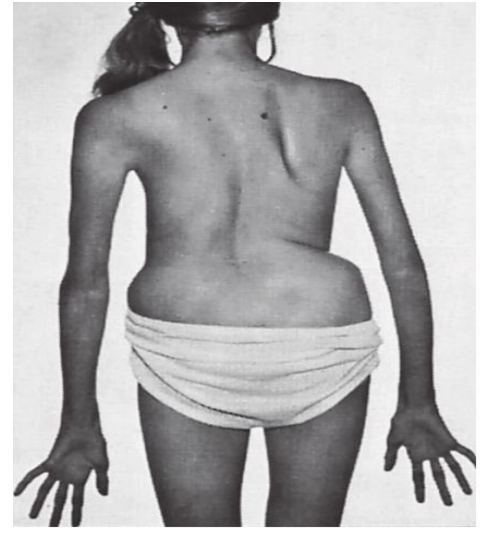
Complications (e.g. recurrent dislocation or scoliosis) may need treatment.
joint laxity is marked,. Joint instability may lead to osteoarthritis, which will require treatment in later life

Marfan's syndrome

autosomal dominant disorder
affecting the bones, joint ligaments, eyes and cardiovascular structures.
It is thought to be due to a cross-linkage defect in collagen and elastin

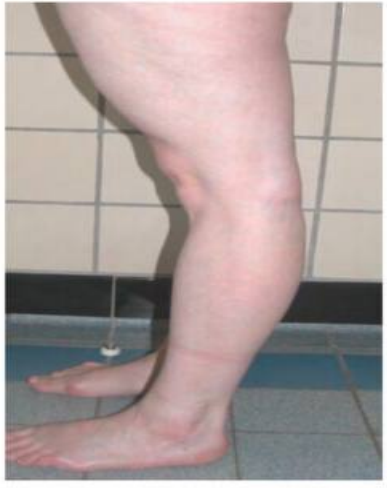
Management Patients

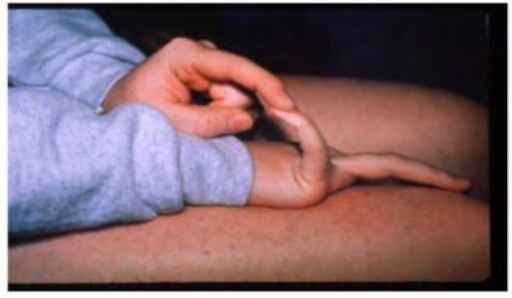
occasionally need treatment for progressive scoliosis or flat-feet.
The heart should be carefully checked before operation

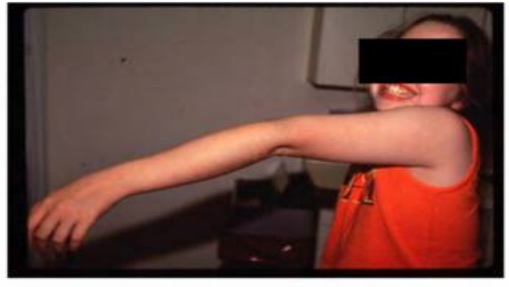


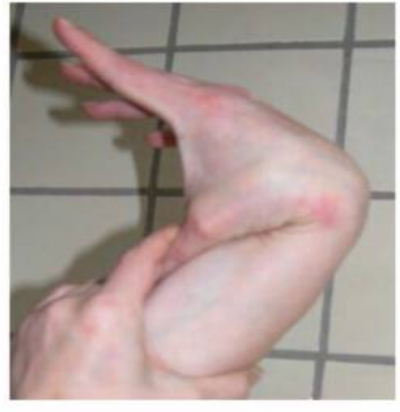
8.8 Marfan's syndrome The combination of disproportionately long arms, 'spider fingers' and scoliosis is characteristic.

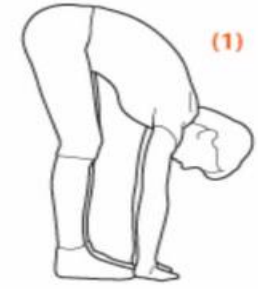
Beighton score

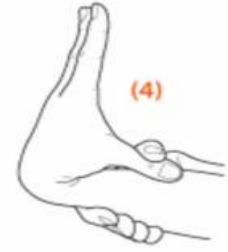






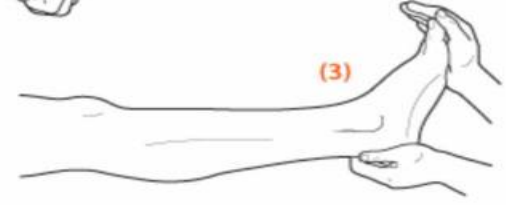


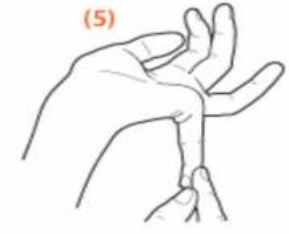




The Beighton score

Beighton's modification of the Carter and Wilkinson scoring system. Give youself 1 point for each of the manoeuvres you can do, up to a maximum of 9 points.





	SCORE Left	Right
1. Can you put your hands flat on the floor with your knees straight?	1	
2. Can you bend your elbow backwards?	1	1
3. Can you bend your knee backwards?	1	1
4. Can you bend your thumb back on to the front of your forearm?	1	1
5. Can you bend your little finger up at 90° (right angles) to the back of your hand?	1	1
		9

Source: Arthritis Research UK

Osteogenesis imperfecta (brittle bones)

one of the commonest of the heritable bone disorders defective synthesis of **type I collagen**
involvement of the **bones, teeth, ligaments, sclerae and skin**.

Clinical feature

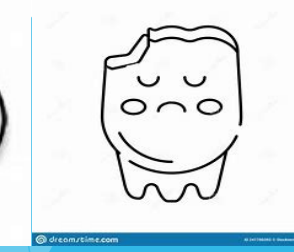
Osteopenia
(2) proneness to fracture;
(3) laxity of ligaments. About two-thirds of patients have
(4) blue sclerae and about one-half have
(5) 'crumbling teeth' (dentinogenesis imperfecta)

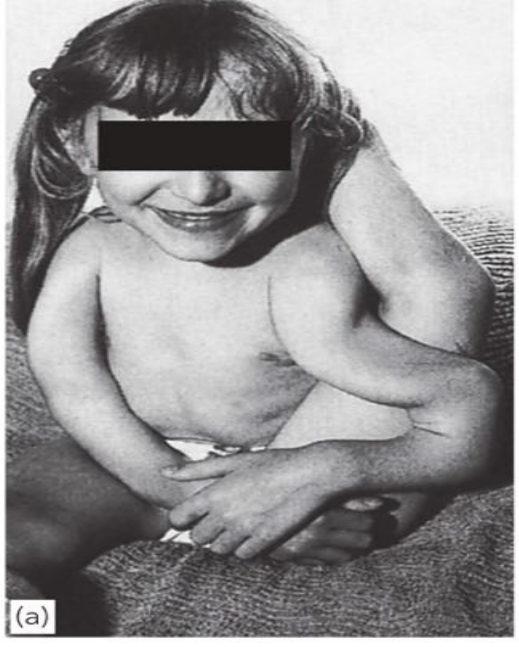
Classification

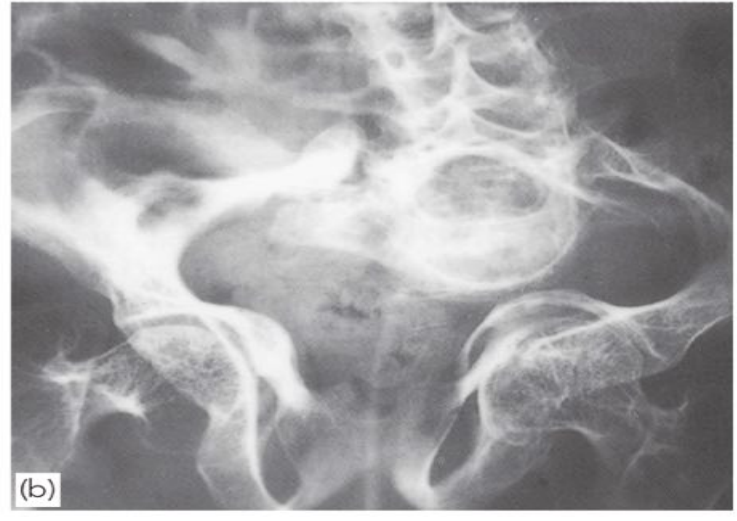
Four clinical sub-groups of OI have been identified: mild, lethal, severe and moderately severe

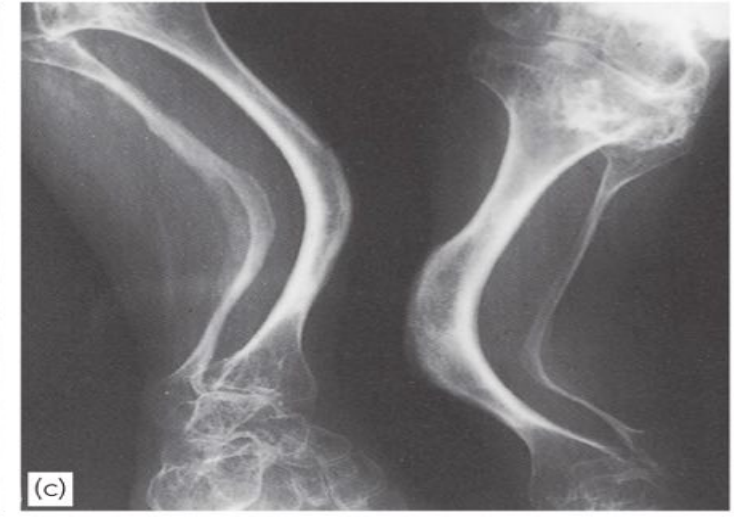
OI Type	Inheritance	Features
I	AD	Osseous fragility (variable) Adulthood hearing loss Blue sclerae
II	AD, AR	Extremely severe osseous fragi Perinatally lethal
III	AR	Moderate to severe osseous fragility Normal sclerae Severe deformity of long bones and spine Variable clinical and radiographic phenotyp
IV	AD, AR	Osseous fragility Generally normal sclerae Severe deformity of long bones and spine

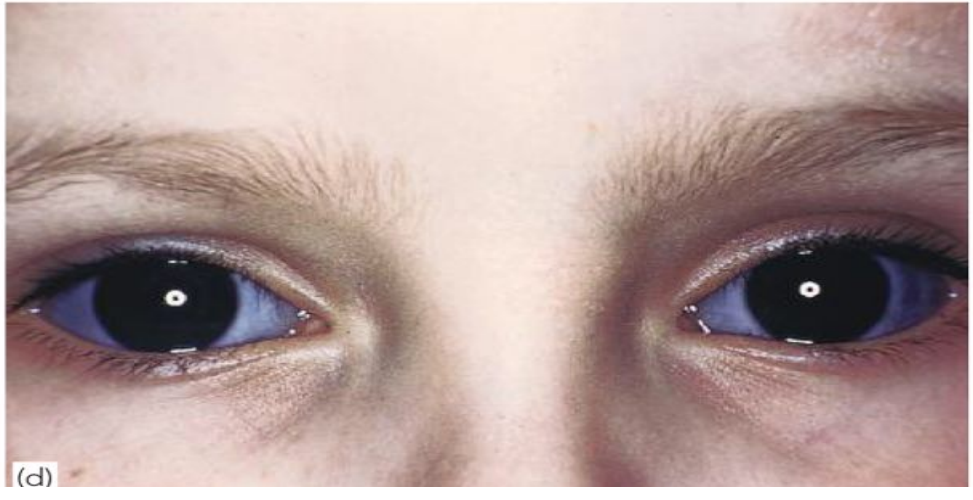
AD = autosomal dominant; AR = autosomal recessive; OI = osteogenesis imperfecta











8.9 Osteogenesis imperfecta (a) This young girl had severe deformities of all her limbs, the result of multiple mini-fractures of the long bones over time. This is the classic (Type III) form of osteogenisis imperfecta (OI). (b,c) X-ray features in a slightly older patient with the same condition. (d) Blue sclerae usually occur in the milder, Type I OI.

Management

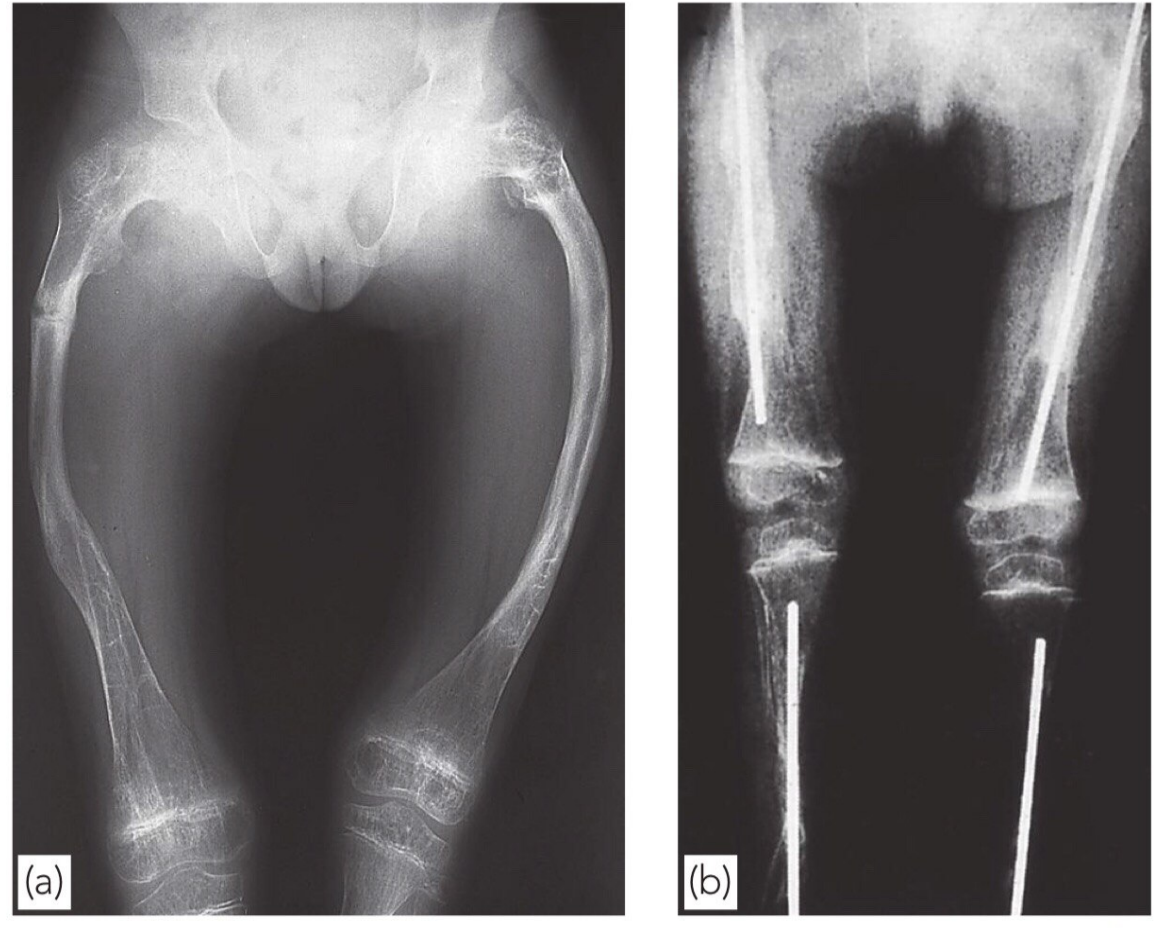
AIM to:

1--gentle nursing of infants to prevent fractures as far as possible;

(2) --prompt splinting when fractures do occur, to prevent unnecessary deformity;

(3) -mobilization to prevent further osteoporosis; and

(4) -correction of deformities, if necessary by multiple osteotomies, bone realignment and intramedullary fixation(sofiled osteotomy or called shish kebab muitiple osteotomy)



8.10 Osteogenesis imperfecta (a) Moderately severe (type IV) disease. These deformities can be corrected by multiple osteotomies and 'rodding' (b). Shish Kebab osteotomy

Neurofibromatosis

one of the commonest single gene disorders affecting the skeleton.

Two types are recognized:

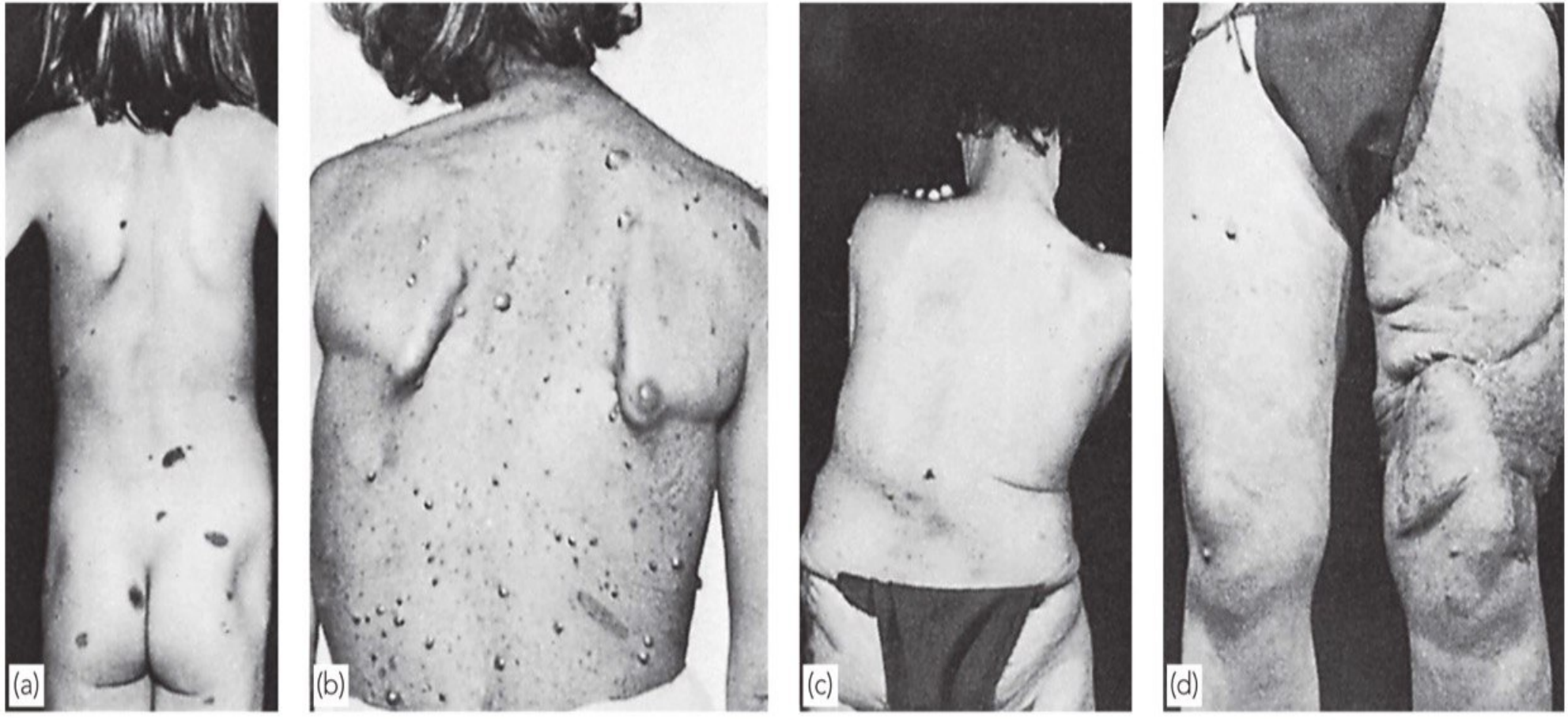
Type 1 (NF-1) – also known as **von Recklinghausen's disease** –

- most characteristic lesions **are neurofibromata (Schwann cell tumours)**
- patches of skin pigmentation **(café-au-lait spots),**
- but other features are remarkably **musculoskeletal abnormalities** are seen in almost one-half of those affected.
- **scoliosis**; the most suggestive deformity
- Congenital tibial dysplasia and **pseudarthrosis**

Type 2 (NF-2) is very rare and is seldom associated with skeletal defects

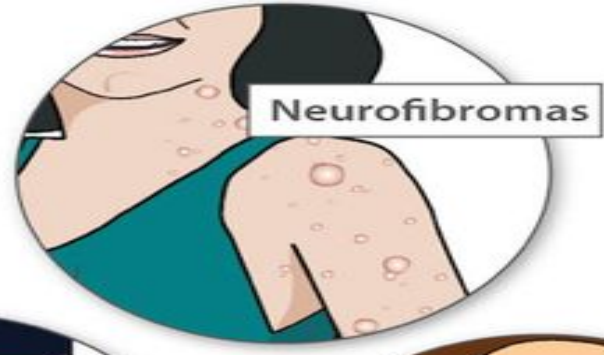
Treatment

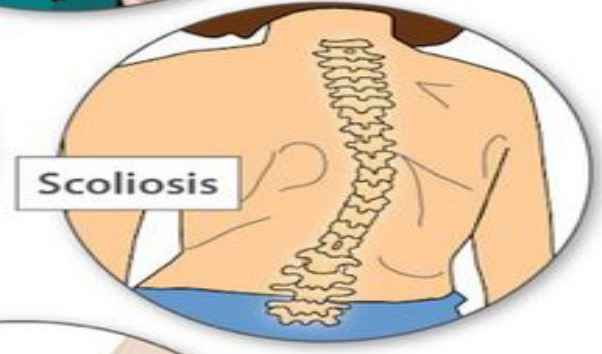
A local tumour causing nerve compression should be removed.
associated conditions such as scoliosis or tibial pseudarthrosis also must be treated

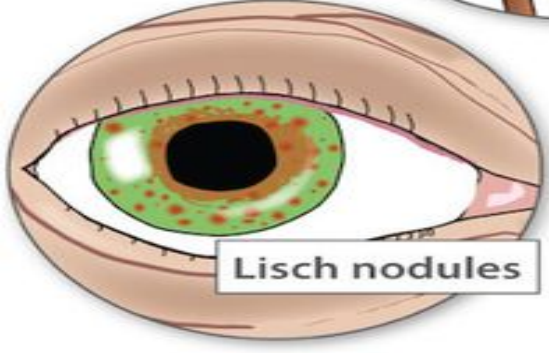


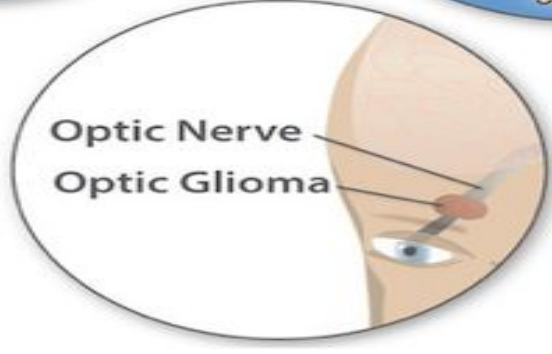
8.11 Neurofibromatosis (a) Café-au-lait spots. (b) Multiple neurofibromata and slight scoliosis. (c,d) A patient with scoliosis and soft-tissue over-growth ('elephantiasis').

Neurofibromatosis Type 1









MNEMONIC:
CAFE SPOT
Cafe-au-lait spots
Axillary, inguinal freckling
Fibroma
Eye: lisch nodules
Skeletal: bowing leg, etc.
Pedigree / **P**ositive family history
Optic Tumor (glioma)

A good surgeon should have a lion's heart, an eagle's eye, & a lady's hand...

(American proverb).


Thank
you