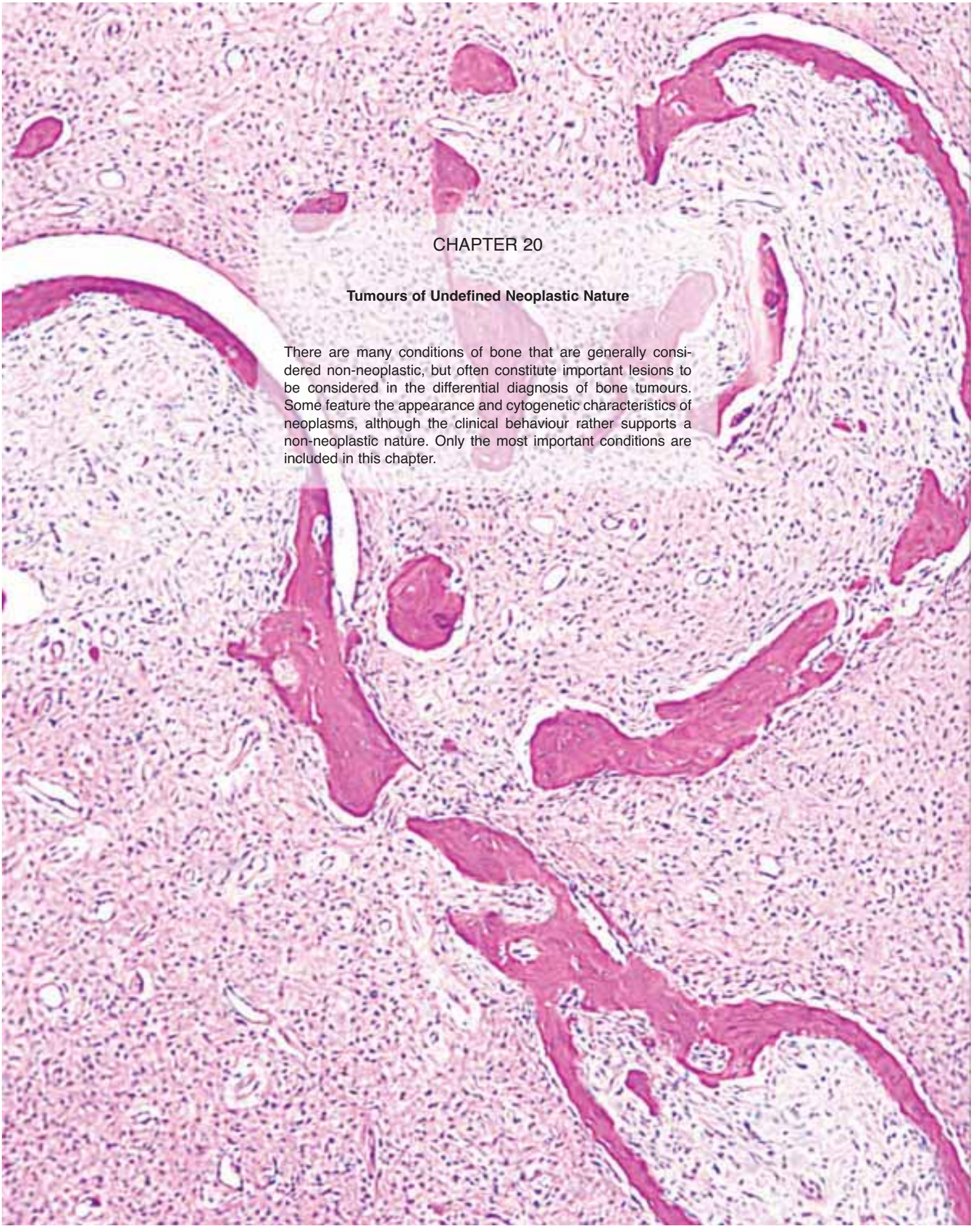

CHAPTER 20

Tumours of Undefined Neoplastic Nature

There are many conditions of bone that are generally considered non-neoplastic, but often constitute important lesions to be considered in the differential diagnosis of bone tumours. Some feature the appearance and cytogenetic characteristics of neoplasms, although the clinical behaviour rather supports a non-neoplastic nature. Only the most important conditions are included in this chapter.

Aneurysmal bone cyst

A.E. Rosenberg
G.P. Nielsen
J.A. Fletcher

Definition
Aneurysmal bone cyst (ABC) is a benign cystic lesion of bone composed of blood filled spaces separated by connective tissue septa containing fibroblasts, osteoclast-type giant cells and reactive woven bone. ABC may arise de novo (primary ABC), or secondarily complicate other benign and malignant bone tumours (secondary ABC) that have undergone haemorrhagic cystic change.

Synonyms
Multilocular haematic cyst, giant cell reparative granuloma.

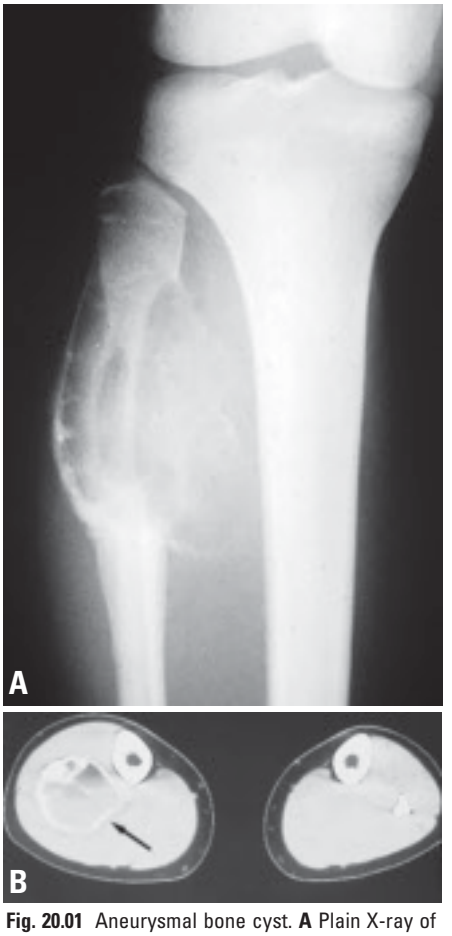

Fig. 20.01 Aneurysmal bone cyst. **A** Plain X-ray of an eccentric lytic mass of the proximal fibula. Note the peripheral shell of reactive bone. **B** CT of the same lesion (arrow).

Epidemiology
ABC affects all age groups, but is most common during the first two decades of life (median age approximately 13 years) and has no sex predilection {1345, 2200}. The estimated annual incidence is 0.15 per million individuals {1239}.

Sites of involvement
ABC can affect any bone but usually arises in the metaphysis of long bones especially the femur, tibia and humerus, and the posterior elements of vertebral bodies. Rare tumours whose morphology is identical to primary ABC of bone have also been described in the soft tissues {53}.

Clinical features / Imaging
The most common signs and symptoms are pain and swelling, which are rarely secondary to fracture. In the vertebrae it can compress nerves or the spinal cord and cause neurological symptoms. Radiographically, ABC presents as a lytic, eccentric, expansile mass with well defined margins. Most tumours contain a thin shell of subperiosteal reactive bone. Computed tomography and magnetic resonance imaging studies show internal septa and characteristic fluid-fluid levels created by the different densities of the cyst fluid caused by the settling of red blood cells {1173,2200}. In secondary ABC, CT and MRI may show evidence of an underlying primary lesion.

Macroscopy
ABC is a well defined and multiloculated mass of blood filled cystic spaces separated by tan white gritty septa. More solid areas can be seen which may represent either a solid portion of the ABC or a component of a primary tumour that has undergone secondary ABC-like changes.

Histopathology
ABC may arise de novo (primary ABC), or secondarily complicate other benign and malignant bone tumours (secondary ABC) that have undergone haemorrhagic cystic change {1281,1557,1699,1849, 1926}.

Primary ABC is well circumscribed and composed of blood filled cystic spaces separated by fibrous septa. The fibrous septa are composed of a moderately dense cellular proliferation of bland fibroblasts, with scattered multinucleated osteoclast-type giant cells and reactive woven bone rimmed by osteoblasts. The woven bone frequently follows the contours of the fibrous septa. In approximately 1/3 of cases the bone is basophilic and has been termed "blue bone", however, its presence is not diagnostic as it can be seen in other entities. Mitoses are commonly present and can be numerous, however, atypical forms are absent. Necrosis is rare unless there has been a pathological fracture. The solid variant of ABC has the same components as the septa and is very similar, if not identical, to giant cell reparative granuloma. Primary ABC accounts for approximately 70% of all cases {177,1859}.
The majority of secondary ABC develop in association with benign neoplasms, most commonly giant cell tumour of bone, osteoblastoma, chondroblastoma and fibrous dysplasia {1173,1345, 2200}. However, ABC-like changes may also omplicate sarcomas, especially osteosarcoma.

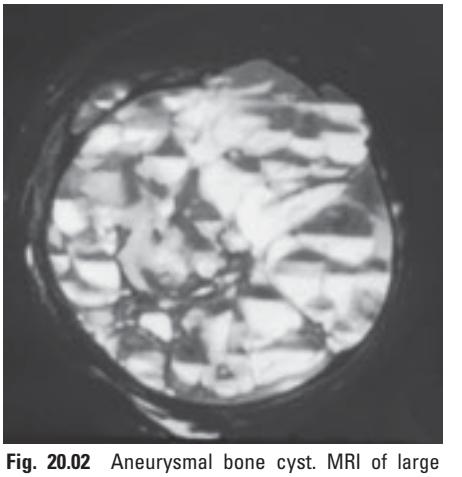

Fig. 20.02 Aneurysmal bone cyst. MRI of large destructive lesion of distal femur. Note numerous fluid-fluid levels.

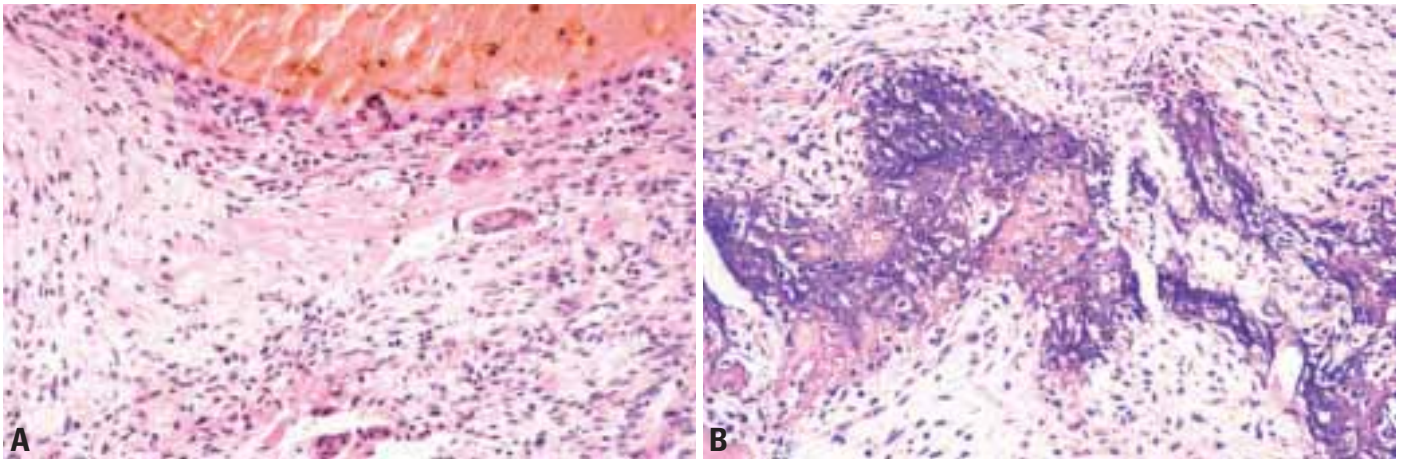

Fig. 20.03 Aneurysmal bone cyst. **A** Septa composed of reactive woven bone, fibroblasts, and scattered osteoclast-like giant cells. **B** So-called 'blue bone" in wall of the lesion.

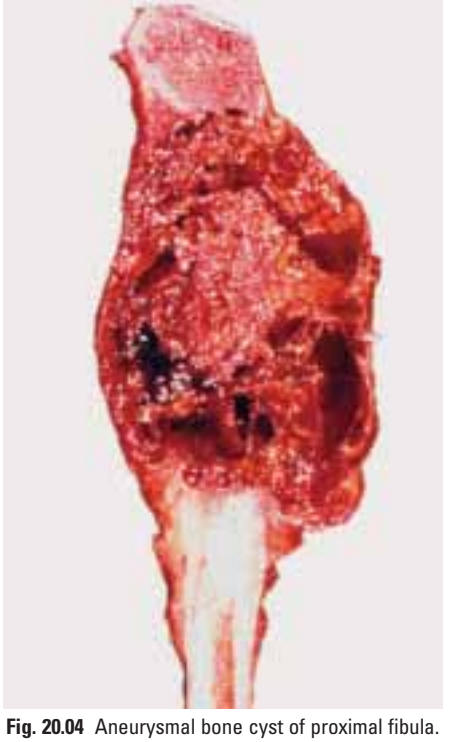

Fig. 20.04 Aneurysmal bone cyst of proximal fibula. The well-defined haemorrhagic multicystic mass has a prominent solid component in the centre.

Genetics

The most notable genetic feature is the characteristic rearrangement of the chromosome 17 short arm {1645}. The chromosome 17 rearrangements are often in the form of balanced translocations, in which material is exchanged with the long arm of chromosome 16. However, there are many variations on this theme, and at least five different chromosomes can serve as translocation partners with chromosome 17 {435,938,1645,1909,2281,2311}. The cytogenetic analyses invariably reveal normal metaphases along with those bearing the translocations. Therefore, the translocations can be assumed to result from acquired aberrations, arising in cytogenetically normal precursor cells. The cytogenetic findings provide compelling evidence that many aneurysmal bone cysts are clonal proliferations, with activation of a 17p oncogene playing a key role in their tumourigenesis. The mechanisms of oncogene activation appear to be heterogeneous, as shown by the different types of 17p rearrangement, and as evidenced by the absence of 17p rearrangement in some cytogenetically abnormal aneursymal bone cysts {135,435,938,1645,1909,2281, 2311}. It is also striking that these varied, but related, cytogenetic abnormalities have been reported across the entire clinicopathological spectrum of aneursymal bone cysts. Chromosome 16 rearrangement was identified in a solid variant aneursymal bone cyst, whereas chromosome 17 rearrangement was found in an extra-osseous case {435}. Hence, it appears that there are generalisable transforming mechanisms, that are utilised irrespective of histological subtype or site of origin.

Prognostic factors

ABC is a benign potentially locally recurrent lesion. The recurrence rate following curettage is variable (20-70%). Spon- taneous regression following incomplete removal is very unusual. Rare cases of apparent malignant transformation of ABC have been reported {1197}.

Simple bone cyst

R.K. Kalil
E.S. Araujo

Definition
An intramedullary, usually unilocular, bone cyst (cavity) filled with serous or sero-sanguineous fluid.

Synonyms
Solitary bone cyst; unicameral bone cyst; juvenile bone cyst; essential bone cyst.

Epidemiology
Males predominate in a ratio of 3:1.
About 85% of patients are in the first two decades of life.

Sites of involvement
There is a predilection for long bones, proximal humerus, proximal femur and proximal tibia accounting for up to 90% of cases. Pelvis and calcaneus are also common locations in older patients.

Clinical features / Imaging
Simple bone cyst can produce pain and swelling but, more frequently, patients present with a pathological fracture.

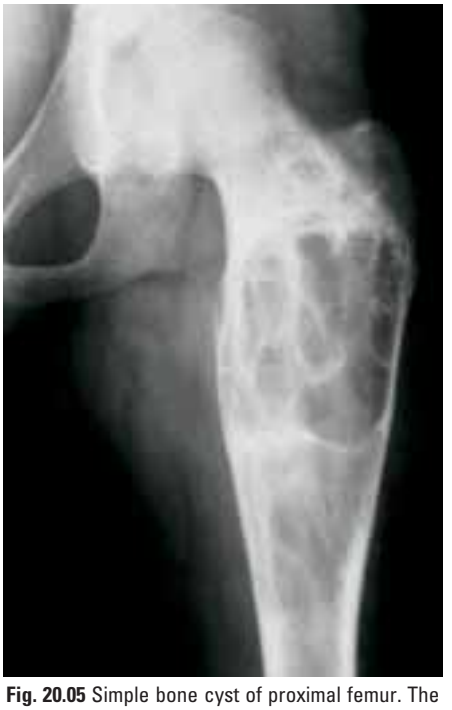

Fig. 20.05 Simple bone cyst of proximal femur. The lesion does not expand the bone.

Roentgenograms show a metaphysio-diaphyseal lucency, extending up to epiphyseal plate, with little or no expansion of bone; marginal sclerosis is absent or very thin. The cortex is usually eroded and thin, but is intact unless pathological fracture has occurred. There can be partial or complete septations of the cavity. MRI usually confirms its fluid content, that can be bloody in fractured lesions {1328}.

Aetiology
Growth defect at the epiphyseal plate has been postulated, or that a venous blood flow obstruction causes the simple cyst {342}.

Macroscopy
The cystic cavity is usually filled with serous or sero-sanguineous fluid. The inner surface of the cyst shows ridges separating depressed zones covered by a layer of thin membrane. Partial septae may be seen.
The occasionally curetted specimen consists of fragments of a usually thin, whitish membrane that may be attached at one surface to bone spicules.

Histopathology
The inner lining and septae of the cyst consist of connective tissue that can, occasionally, contain foci of reactive new bone formation, haemosiderin pigment and scattered giant cells. Fibrinous deposits are often seen. Some of these are mineralized, resembling cementum. Occasionally, histological features of fracture callus may be prominent. Rare "solidified" cases of simple bone cyst have been described in older subjects.

Genetics
A highly complex clonal structural rearrangement involving chromosomes 4, 6, 8, 16, 21 and both chomosomes 12 has been described in a surgically resected solitary bone cyst in an 11-year-old boy {2195}.

Prognostic factors
Recurrence is reported at 10-20% of cases, especially in children. Growth arrest of the affected bone and avascular necrosis of the head of the femur after pathological fracture can occur {2022}. Spontaneous healing after fracture has been described {52}.

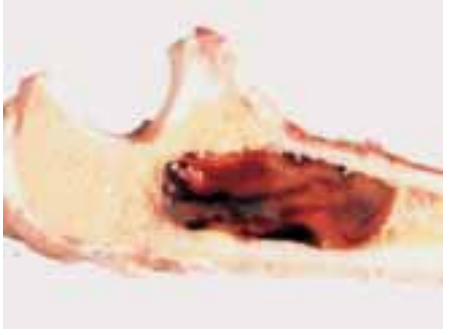

Fig. 20.06 Simple bone cyst of proximal ulna. A unilocular cyst contains fibrin clot.

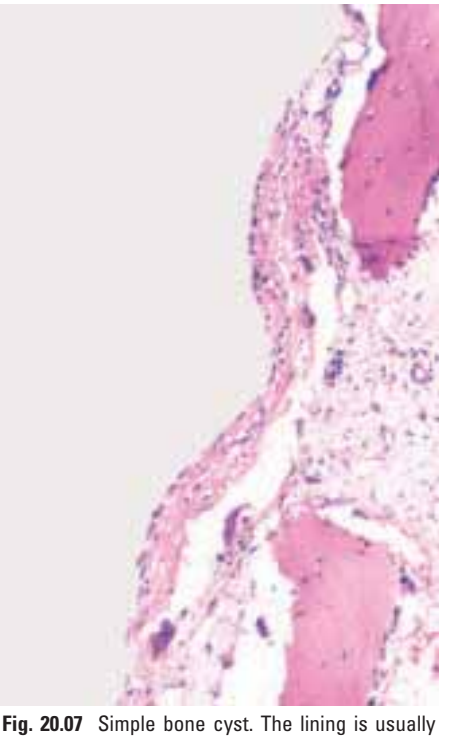

Fig. 20.07 Simple bone cyst. The lining is usually inconspicuous and contains scattered spindle cells and giant cells.

Fibrous dysplasia

G. Siegal
P. Dal Cin
E.S. Araujo

Definition
Fibrous dysplasia (FD) is a benign medullary fibro-osseous lesion which may involve one or more bones.

Synonyms
Fibrocartilagenous dysplasia, generalized fibrocystic disease of bone.

Epidemiology
Fibrous dysplasia occurs in children and adults world-wide and affects all racial groups with an equal sex distribution. The monostotic form is six times more common than polyostotic fibrous dysplasia.

Sites of involvement
The gnathic (jaw) bones are the most common site of involvement in surgical series (because they are often symptomatic) {1596}. In women, long bones are more often involved, whereas ribs and the skull are favoured sites in men {2154}. In the monostotic form, about 35% of cases involve the head, a second 1/3 occur in the femur and tibia, and an additional 20% in the ribs. In the polyostotic form, the femur, pelvis, and tibia are involved in the majority of cases {890}.

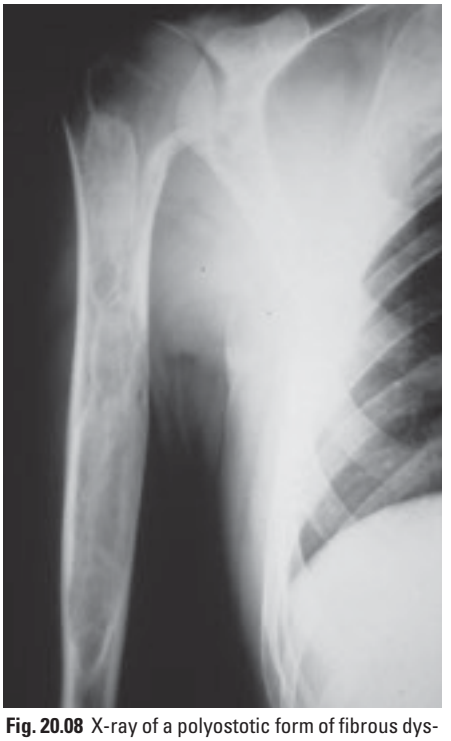

Fig. 20.08 X-ray of a polyostotic form of fibrous dysplasia. There is a well defined lucency with sclerotic margins.

Clinical features / Imaging
Fibrous dysplasia may present in a monostotic or polyostotic form, and in the latter case, can be confined to one extremity or one side of the body or be diffuse. The polyostotic form often manifests earlier in life than the monostotic form {890}. The lesion is often asymptomatic but pain and fractures may be part of the clinical spectrum {333}. Fibrous dysplasia may also be associated with oncogenic osteomalacia {1660}.
The polyostotic form of fibrous dysplasia is intimately associated with McCune-Albright syndrome, in which there are endocrine abnormalities and skin pigmentation. There is also a relationship between fibrous dysplasia and intramuscular myxomas (Mazabraud syndrome) {630}.
Rontgenographic studies often show a non-aggressive geographic lesion with a ground glass matrix. There is generally no soft-tissue extension, and a periosteal reaction is not seen unless there is a complicating fracture. CT scans and MRI further delineate these features and better define the extent {422,1035,2118}.

Aetiology
Activating mutations of the G proteins have been identified in both the monostotic and polyostotic forms and may be aetiologically important.

Macroscopy
The bone is often expanded and the lesional tissue has a tan grey colour with a firm-to-gritty consistency. There may be cysts, which may contain some yellow-tinged fluid {1948}. When cartilage is present, it often stands out as sharply circumscribed of blue-tinged translucent material {2154}.

Histopathology
The lesion is generally well circumscribed and composed of fibrous and osseous components; which are present in varying proportions from lesion to lesion and also within the same lesion. The fibrous component is composed of cytologically bland spindle cells with a low mitotic rate. The osseous component is comprised of irregular curvilinear trabeculae of woven (or rarely lamellar) bone. Occasionally, the osseous component may take the form of rounded psammomatous or cementum-like bone. Secondary changes such as foam cells,

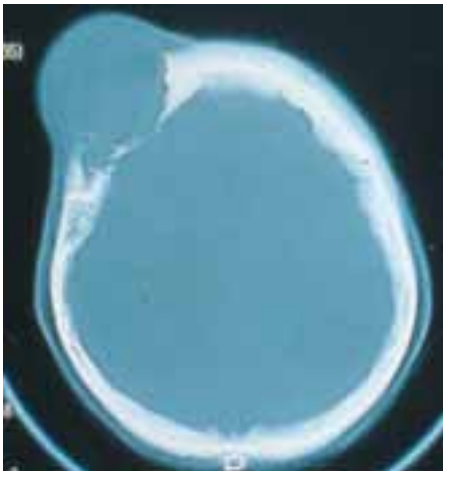

Fig. 20.09 CT of skull with fibrous dysplasia. In flat bones the process is often expansile.

Fig. 20.10 Fibrous dysplasia with gross cartilaginous components.

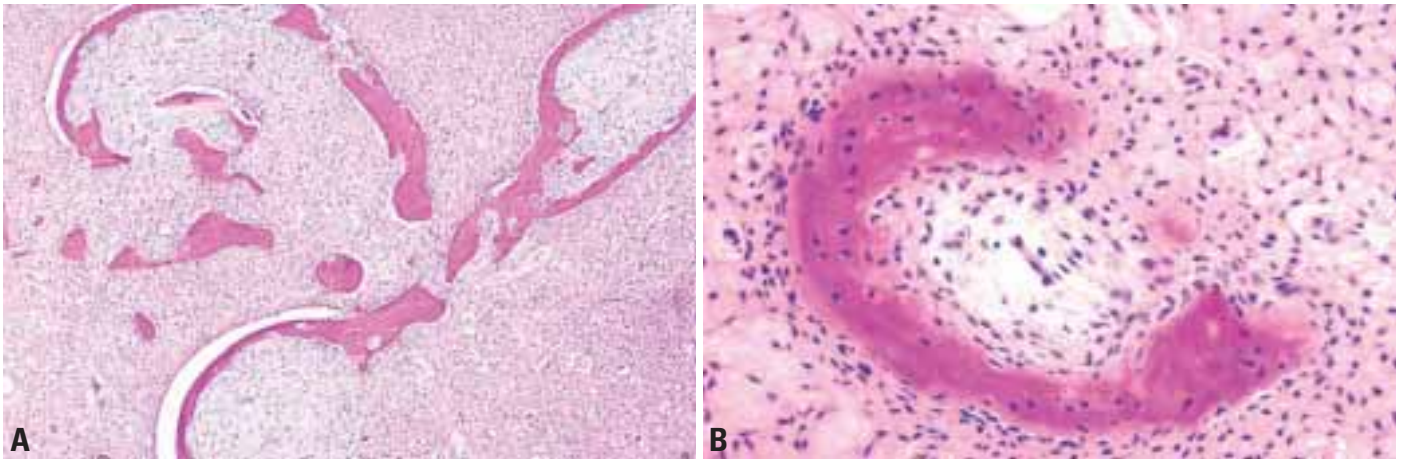

Fig. 20.11 Fibrous dysplasia. **A** Characteristic C shaped bony spicules with hypocellular spindle cell stroma. **B** High power appearance showing the typical appearance of bone which seems to be dissected by spindle cell proliferation. Note that there is no osteoblastic rimming.

multinucleate giant cells, a secondary aneurysmal bone cyst or myxoid change may occur.

Genetics

Activating mutations in the *GNAS1* gene, encoding the alfa subunit of stimulatory G protein, has been demonstrated in monostotic as well as polyostotic fibrous dysplasia {382} (see also chapter on McCune-Albright syndrome).

Clonal chromosome aberrations have been reported in eight of eleven investigated cases, suggesting that this entity is neoplastic in nature {439}. The only recurrent changes described so far are structural rearrangements involving 12p13 and trisomy 2 (three cases each).

Prognostic factors

The prognosis of patients with FD is good. Malignant transformation occurs, but rarely.

Osteofibrous dysplasia

V.J. Vigorita
B. Ghelman
P.C.W. Hogendoorn

Definition
Osteofibrous dysplasia (OFD) is a self-limited benign fibro-osseous lesion of bone characteristically involving cortical bone of the anterior mid-shaft of the tibia during infancy and childhood.

Synonyms
Kempson-Campanacci lesion, cortical fibrous dysplasia.

Epidemiology
The lesion is more commonly seen in boys during the first two decades of life with a precipitous drop-off thereafter, OFD has been reported in neonates, but is extremely rare after skeletal maturation.

Sites of involvement
The proximal or middle-third of the tibia is the most frequent site of involvement {301}. Lesions can be bilateral with ipsilateral or contralateral involvement of the fibula. Other sites include the ulna and radius {1055}. Multifocal or large confluent lesions oriented longitudinally along the cortical axis are not unusual.

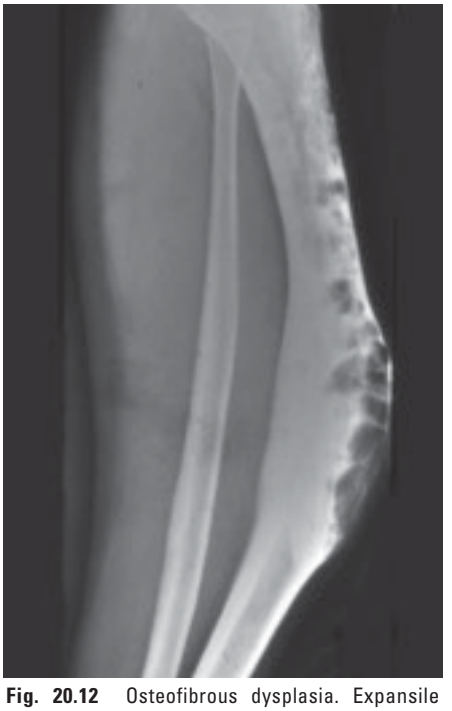

Fig. 20.12 Osteofibrous dysplasia. Expansile lucent, longitudinally-oriented tibial lesion surrounded by sclerosis and thinning of the anterior cortex of the diaphysis of the tibia. Note the anterior bowing of the tibia.

Clinical features / Imaging
The lesion is rare after the age of 15. The most common presenting symptoms are swelling or a painless deforming bowing of the involved segment of the limb. OFD is typically epicentered in the cortical bone but may involve the medullary cavity by extension. Although slow growth is characteristic of OFD, some lesions are aggressive and may involve the entire bone with significant bowing deformity. Often well demarcated, it is associated with a thinning, expanding or even missing cortex. The expanding cortex is often sclerotically rimmed near the medullary bone. Separate or confluent oval-shaped, scalloped, saw-toothed or bubbly multiloculated lytic lesions are often noted. Perilesional sclerosis may be considerable. The radiodensity of the interior of the lytic foci are typically more radiodense than soft tissue. Periosteal reactions and soft tissue extensions are unusual. Bone scans are typically hot. CT scans classically delineate a cortical epicentre to the lesion not breaking through into the soft tissue and demarcated from medullary bone by sclerosis. MRI findings show high intensity lesions on T2 weighted images and mixed signals on T1 and fat suppressed images.

Aetiology
The occurrence of so-called OFD-like adamantinoma, to be distinguished from classic epithelium-rich adamantinoma but differentiated from OFD with difficulty, raises the possibility of an association between OFD and adamantinoma {112, 918,1188}. Some cases of OFD may arise de novo and are not related to adamantinoma.

Macroscopy
OFD is solid with a whitish, yellowish or reddish colour and soft or gritty texture blending into the surrounding host bone. The periosteum often appears intact but the cortex is thin or absent. The medullary extension is usually demarcated by a sclerotic rim.

Histopathology
The histopathologic findings in OFD are irregular fragments of woven bone often rimmed by lamellar layers of bone laid down by well defined osteoblasts. Osteoclasts may be present. The fibrous component consists of bland spindle cells with collagen production and a matrix that varies from a myxoid component to one that is moderately fibrous. Mitoses are extremely rare. A zonal architecture has been delineated with thin spicules and woven bone or even fibrous tissue predominating in the centre of the lesion with more abundant anastomosing and lamellar bone peripherally, the latter often blending

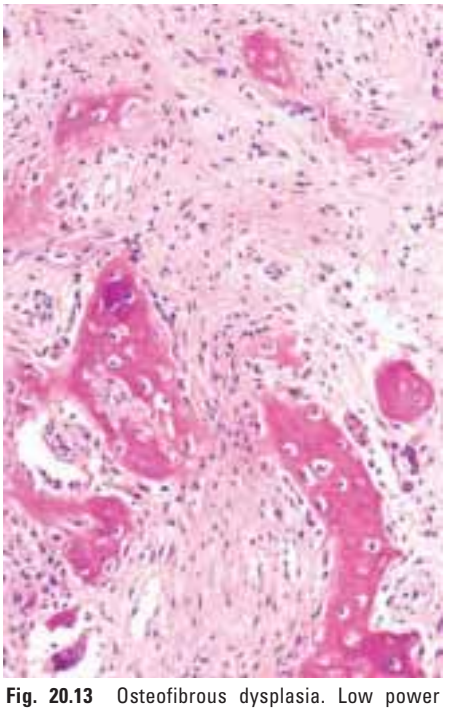

Fig. 20.13 Osteofibrous dysplasia. Low power magnification of the lesion featuring hypocellular spindle cell proliferation and spicules of bone. The bony spicules display prominent osteoblastic rimming.

Table 20.01
Chromosomal abnormalities in osteofibrous dysplasia.

No./Author	Age/sex	Tumour (type)	Karyotype abnormality
1 Bridge {256}	11,M	OFD (R)	47,XY,+12 (FISH: also +8,+20)
2 Bridge {256}	19,M	OFD (R)	49,XY,+7,+8,+22
3 Bridge {257}	18,F	OFD (P/R)	52,XX,+5,+7,+7,+8,+21,+21

P, primary tumour; R, recurrence, FISH: fluorescence in situ hybridization. Cases 1/2:keratin-negative OFDs.

into the surrounding host bone {298}. Secondary changes of hyalinization, haemorrhage, xanthomatous change, cyst formation and foci of giant cells are rare. Cartilage or clusters of epithelial cells are absent.

Immunophenotype

Osteofibrous dysplasia is positive for vimentin and occasionally so for S100 and Leu7. Isolated cytokeratin positive mast cells have been mentioned.

A tumour should be defined as OFD-like adamantinoma when keratin-positive epithelial cells are found {918,1534}.

Genetics

Numerical chromosomal abberations, especially trisomy 7 and 8 have been demonstrated {256, 267}, as well as FOS and JUN proto-oncogene products.

Mutations of the alpha-subunit of signal transducing G-proteins with an increase in cyclic AMP formation are specifically absent {1845}.

Prognostic factors

The natural history of osteofibrous dysplasia is that of gradual growth during the first decade of life with stabilization at about 15 years of age followed by healing or spontaneous resolution. The progression of OFD-like adamantinoma (or 'OFD with keratin positive cells') to classic adamantinoma has been shown in a few patients {562,918, 1041,2016}. In many others, there is at least strong suggestion of a progression {381,2157, 2235}.

OFD-like adam- antinoma seldom progresses to classic adamantinoma.

Langerhans cell histiocytosis

B.R. De Young
K.K. Unni

Definition

Langerhans cell histiocytosis is a neoplastic proliferation of Langerhans cells.

ICD-O codes

Langerhans cell histiocytosis, NOS	9751/1
Langerhans cell histiocytosis, unifocal	9752/1
Langerhans cell histiocytosis, multifocal	9753/1
Langerhans cell histiocytosis, disseminated	9754/3

Synonyms

Eosinophilic granuloma, Langerhans cell granulomatosis, histiocytosis X. Clinical variants have been referred to as Hand-Schuller-Christian disease and Letterer-Siwe disease.

Incidence

Langerhans cell histiocytosis (LCH) is a relatively rare disorder, accounting for less than 1% of all osseous lesions. LCH involving bone has been reported in a wide age distribution ranging from the first months to the 8th decade of life with 80-85% of cases seen in patients under the age of 30, and 60% under the age of 10. Males are affected twice as often as females {1026,1253,1259,2253}.

Sites of involvement

Although any bone may be involved, there is a predilection for LCH to involve the bones of the skull, notably the calvarium. Other frequently involved sites include the femur, the bones of the pelvis, and the mandible {1259,2253}. In adults, the rib is the most frequent site of involvement {2253}. Monostotic disease is much more common than polyostotic.

Clinical features / Imaging

Pain and swelling of the affected area occur most commonly. Other findings are related to the bone involved. In cases of temporal bone involvement, the presenting features can show significant clinical overlap with otitis media or mastoiditis. With mandibular involvement, loosening or loss of teeth can be encountered. Vertebral body disease may result in compression fracture and possible neurological impairment. In adults, the lesion can present as an incidental finding on imaging studies.

Early lesions may appear very aggressive radiographically. Roentgenograms generally show a purely lytic, well demarcated lesion, usually associated with thick periosteal new bone formation. Skull lesions are sometimes described as representing a "hole in a hole" due to uneven involvement of the two osseous tables. In the vertebrae, the body is involved producing collapse giving rise to vertebra-plana.

Macroscopy

The involved tissue is soft and is red in colour.

Histopathology

The diagnosis depends on the recognition of Langerhans cells, which are intermediate size with indistinct cytoplasmic

Fig. 20.14 Langerhans cell histiocytosis. Plain X-ray showing lucency in the shaft of the femur associated with thick periosteal new bone formation.

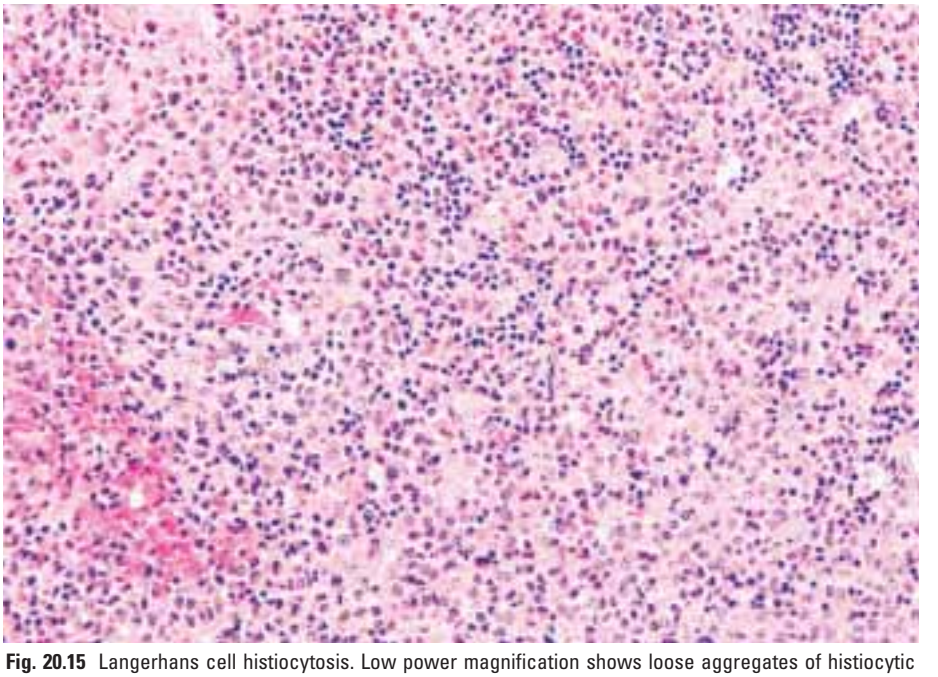

Fig. 20.15 Langerhans cell histiocytosis. Low power magnification shows loose aggregates of histiocytic appearing cells in a mixed inflammatory background with prominent eosinophilia and evidence of recent haemorrhage.

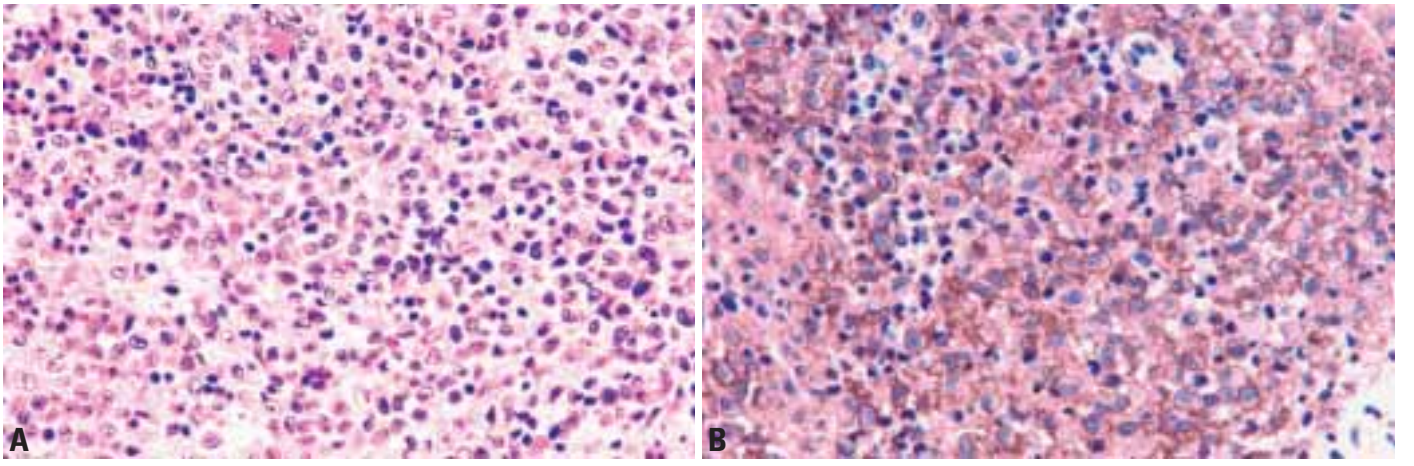

Fig. 20.16 Langerhans cell histiocytosis. **A** High power photomicrograph depicting Langerhans cells with ovoid to reniform nuclei with irregular notches and grooves. **B** Langerhans cells show distinct membrane based immunoreactivity for CD1a.

borders, eosinophilic to clear cytoplasm with oval nuclei which frequently are indented, irregular in outline, and typically possess nuclear grooves. Chromatin is either diffusely dispersed or condensed along the nuclear membrane. In osseous LCH, the Langerhans cells are found in nests or clusters. Diffuse sheet-like architecture is rare, and, if present, should raise the suspicion of haematolymphoid malignancy. The Langerhans cells are frequently admixed with inflammatory cells including large numbers of eosinophils, as well as lymphocytes, neutrophils and plasma cells. Necrosis is common and does not portend an aggressive clinical course. Multinucleated osteoclast-like giant cells and occassionally lipid laden histiocytes may be present. The cells of LCH can exhibit a relatively brisk mitotic rate, with up to 5-6 mitoses per 10 high power fields.

Immunohistochemistry

Langerhans cells have a characteristic immunophenotype which includes expression of membrane based CD1a {584} and S100 protein in both a nuclear and cytoplasmic pattern {1530}. These cells typically fail to express CD45.

Ultrastructure

Langerhans cells contain unique intracytoplasmic "tennis racket" shaped inclusions known as Birbeck granules which are thought to arise from the cell membrane.

Genetics

Studies of X-chromosome inactivation demonstrated that LCH is clonal {2275}.

Prognostic factors

The prognosis for patients with either monostotic or limited polyostotic disease is good. Death can result from LCH, but this is a rare event and is associated only with the disseminated forms of the disease and usually occurs in younger individuals less than three years at diagnosis and with visceral involvement.

Erdheim-Chester disease

T.N. Vinh
D.E. Sweet

Definition
Erdheim-Chester disease (ECD) is a rare histiocytosis characterized by infiltration of skeleton and viscera by lipid laden histiocytes leading to fibrosis and osteosclerosis.

Synonyms
Lipogranulomatosis, lipoidgranulomatosis, lipid (cholesterol) granulomatosis, polyostotic sclerosing histiocytosis.

Epidemiology
The disease demonstrates a slight male predominance with a peak incidence in the 5th through the 7th decades (age range is 7 to 84 years; mean age 53) {2203}.

Sites of involvement
ECD predominantly affects the major long bones of the extremities; but flat bones can also be involved {306,664, 1138}. Extraskeletal manifestations occur in more than 50% of cases, e.g. kidney/retroperitoneum, heart/pericardium, and lung.

Clinical features / Imaging
General symptoms consist of mild bone pain, occasionally associated with soft tissue swelling, fever, weight loss, and weakness. Other manifestations include exophthalmos, diabetes insipidus, kidney failure, cardiac, pulmonary, or neurological symptoms, eyelid xanthomas, and hepatosplenomegaly {627,1091, 1218,2045,2203}. Despite the impressive lipid laden histiocytic infiltration, the serum lipid profile is relatively normal.
The radiographic picture of ECD is unique and includes bilateral, symmetric, patchy or diffuse sclerosis of the medullary cavity of major long bones, with relative epiphyseal sparing {1785}. One third of cases have a mixed osteolytic and sclerotic pattern {276,1463, 2045}. The sclerotic lesions show increased uptake on bone scan. CT scan serves to detect orbital, dural, and retroperitoneal lesions. On MRI the lesion is of low signal intensity on T1-weighted sequences, enhances intensely after gadolinium injection {2299}, and gives mixed signal intensity on T2-weighted sequences {118,2045}.

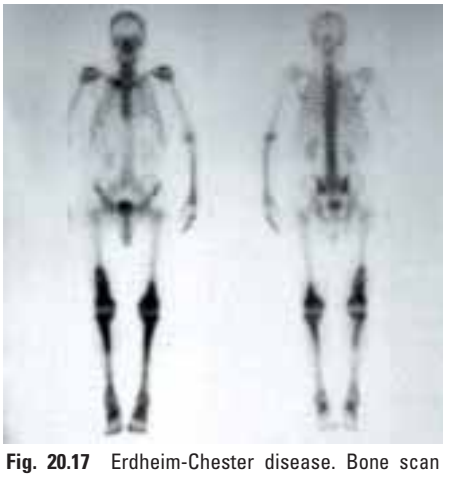

Fig. 20.17 Erdheim-Chester disease. Bone scan highlights the increased uptake throughout the entire length of the bones involved.

Macroscopy
On gross examination, the lesions appear as sulphur-yellow and variably firm.

Histopathology
The histology consists of a diffuse infiltration of marrow by foamy histiocytes associated with dense fibrosis, lymphocytes, plasma cells and Touton giant cells. There is massive reactive sclerosis of cortical and cancellous bone with irregular cement lines.

Immunophenotype
Immunohistochemistry confirms the monocyte/macrophage lineage of the lipid laden foamy histiocytes and giant cells by their expression for lysozyme, Mac387, CD68 (Kp-1), CD4 {2168}, alpha-1-antichymotrypsin, alpha-1-antitrypsin and S100 protein (variable) {1615}. They are negative for CD1a.

Ultrastructure
Electron microscopy shows a predominance of histiocytes with indented nuclei, abundant intracytoplasmic lipid vacuoles and sparse mitochondria, lysosomes, and endoplasmic reticulum. Birbeck granules are absent {664}.

Prognostic factors
The majority of patients eventually die within 3 years of renal, cardiovascular, pulmonary, or CNS complications {2203}.

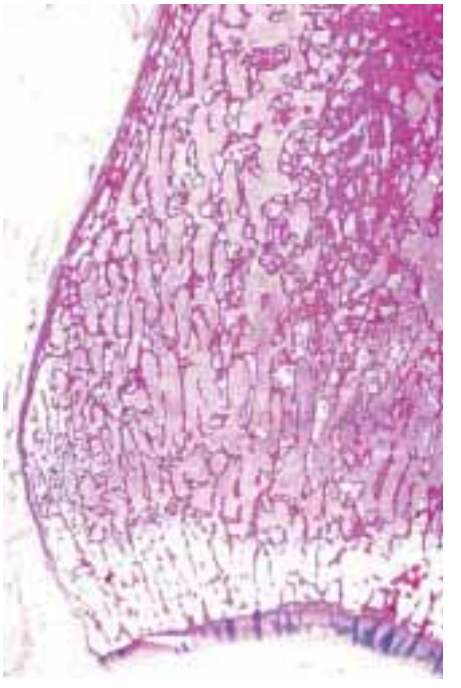

Fig. 20.18 Erdheim-Chester disease. Macrosection of tibia showing medullary sclerosis, which abruptly ends at the physis.

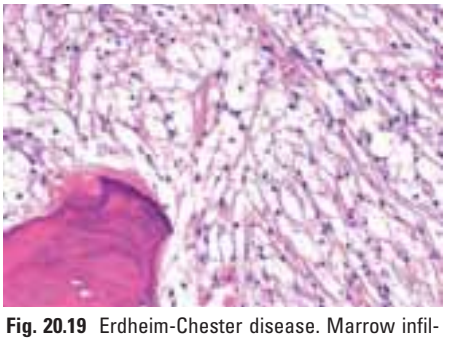

Fig. 20.19 Erdheim-Chester disease. Marrow infiltration by numerous foamy histiocytes associated with dense fibrosis.

Chest wall hamartoma

E.F. McCarthy
H. Dorfman

Definition
Chest wall hamartoma is a non-neoplastic proliferation of mesenchymal tissue, predominantly cartilage, admixed with aneurysmal bone cyst elements. The lesion develops during fetal life and presents at or shortly after birth with an extrapleural mass arising from the rib cage.

Synonyms
Vascular hamartoma of infancy, mesenchymal hamartoma of the chest wall, mesenchymoma.

Epidemiology
The lesion is rare. To date only 59 cases have been documented. In approximately 40% of cases, the mass is apparent at birth. However, most cases present between ages one month to one year {97}. Less frequently, lesions may present in children up to age eight. One adult aged 26 was diagnosed with a chest wall hamartoma {531}. The lesion has also been diagnosed in utero with CT scans or ultrasound {1351,1807}.

Sites of involvement
The lesion is an intrathoracic and extraplural mass and arises from one or more ribs. Almost always, the posterior or lateral portions of the rib are affected. Rarely, the lesion may be multifocal or bilateral in the chest cavity {2132}.

Clinical features
Chest wall hamartoma presents as a mass or fullness of the rib cage. Most often, the bulk of the mass is intrathoracic. As a result, infants frequently develop respiratory distress.
Radiographically, chest wall hamartoma is a partially mineralized mass arising from the inside of the rib cage and extending into the chest cavity. The involved rib is partially destroyed, and adjacent ribs are deformed. CT images show an expansile mass and partial rib destruction. Magnetic resonance images shows alternating areas of high and low signal on T1 and T2 sequences, reflecting both solid and cystic components {1886}.

Macroscopy
Lesions range from 3 to 7 cm in maximum dimension. Cut surface reveals grey to white solid areas adjacent to cystic cavities filled with blood.

Histopathology
Solid areas consist primarily of mature hyaline cartilage, although areas resembling chondroblastoma may be present. The cartilage often shows enchondral ossification. Areas with fibroblast-like cells are also present. Cystic areas show features typical of aneurysmal bone cyst: blood-filled lakes are bounded by fibrous septae which contain reactive bone and osteoclast-like giant cells.

Prognostic factors
Complete surgical removal of the affected ribs results in cure. Scoliosis is an occasional complication of surgery. Rarely untreated patients may die of respiratory insufficiency {1379}. However, most unoperated lesions remain stable. Spontaneous regression has also been reported {721}.

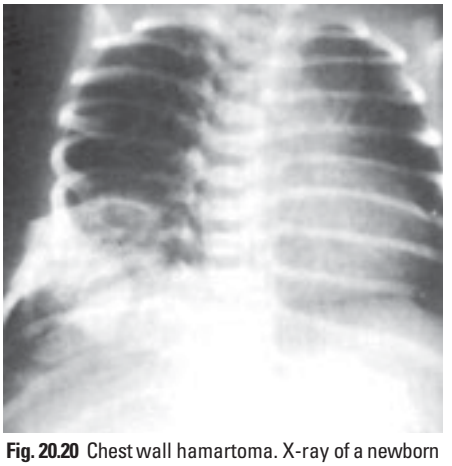

Fig. 20.20 Chest wall hamartoma. X-ray of a newborn showing a lesion in the right lower rib cage, involving several ribs and projecting into the chest cavity.

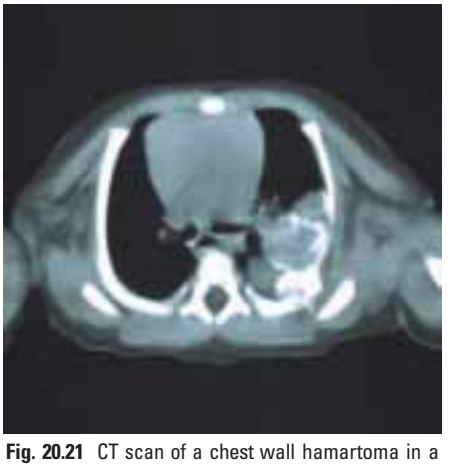

Fig. 20.21 CT scan of a chest wall hamartoma in a three-day-old infant involving the inner aspect of a rib. The lesion has a radiodense component.

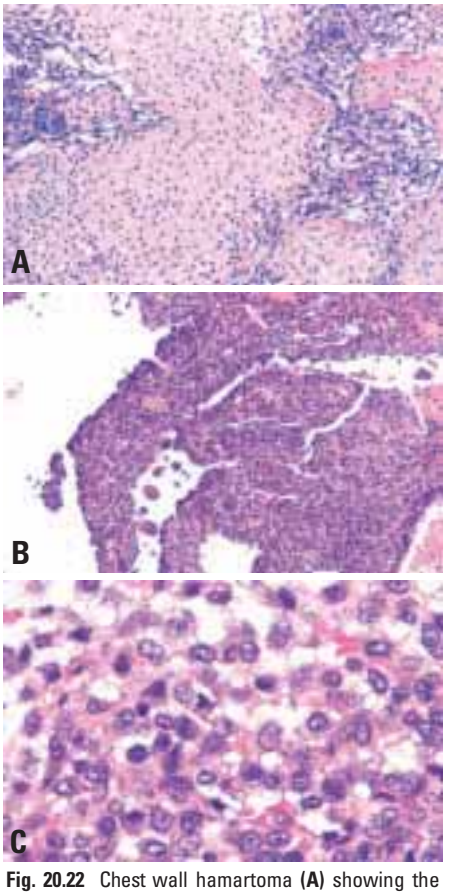

Fig. 20.22 Chest wall hamartoma **(A)** showing the typical chondroid matrix. **B** Histology similar to that of a conventional aneurysmal bone cyst with blood-filled lakes separated by septae composed of stromal cells and multinucleated giant cells. **C** Immature chondroblastoma-like cells.