

## Answers and additional information for Clinical Quiz in the previous page

### Q 1.

1. Vertebra plana of L1 vertebral body.
2. The differential diagnoses in an otherwise healthy child include eosinophilic granuloma, haemangioma or a benign cyst.
3. Usually radiographs are characteristic but not diagnostic. Bone scans is useful to exclude other lesions. MRI best defines soft tissue and bone components and help in the diagnosis. Biopsy is seldom indicated as it is a benign lesion.
4. Management of a solitary foci is close monitoring until the child becomes asymptomatic or healing is completed (usually in more than 5 months).

### Eosinophilic Granuloma (EG)

- EG is usually solitary (unifocal), though about 10% may develop multiple lesions. Lesions involving lungs, skin or mucous membrane may be present.
- Histological similarity has been noted between bony lesions and disseminated lesions of bone marrow and visceral organs (Hand-Schuller-Christian disease, Letterer-Siwe syndrome): all characterized by the presence of histiocytes and accumulation of lipid material.
- The whole spectrum of illness is referred as Langerhans cell histiocytosis ('histiocytosis X').

### Q 2.

1. Excessive bone density of spine and ribs (AP view), and typical 'sandwich' vertebrae (lateral view).
2. Osteopetrosis or Albers-Schönberg disease, 'marble bone disease', 'ivory bone disease'
3. Blood count may show profound anaemia and family members need to be screened.
4. Fracture treatment is the main concern. In young patients, trial of conservative treatment (traction for six weeks) is preferred because bone healing is poor and risk of postoperative osteomyelitis is high. Plating or intramedullary nailing are difficult as the bone may be too dense for the placement of screws and the absence of a medullary canal makes it impossible to insert intra-medullary device.

### Osteopetrosis

- The condition was described by Albers-Schönberg in 1904. It is characterized by a systemic increase in bone mass, obliteration of marrow cavities and loss of trabecular architecture. Bones though dense are abnormally fragile.
- Accompanying myelopathic anaemia is the rule. Three variants: a 'malignant' infantile type with autosomal recessive trait, an 'intermediate' type with autosomal recessive pattern, and a 'benign' adult type with autosomal dominant trait, have been recognised.
- The clinical expression of the disease varies from incidentally recognised radiodense bones in asymptomatic adults or symptomatic patients with bone pain and low-energy fractures, to the fulminant infantile form with morbidly abnormal bones. The infantile form has a progressive course and anaemia, haemorrhage and infection may lead to early death.

### Q 3.

1. All the lower thoracic and lumbar vertebral bodies are of decreased height. The lateral view shows the typical central anterior tongue projection between the upper and lower end-plates.
2. These features are typical of mucopolysaccharide (MPS) storage diseases.

### Mucopolysaccharidoses (MPS)

- MPS storage diseases result from deficiency specific lysosomal enzymes necessary for the degradation of glucosaminoglycans and are characterized by the intra-cellular accumulation of partially degraded molecules which result in cell dysfunction or death.
- MPS are classified according to the specific enzyme deficiency and numbered from I to VII. Morquio disease is MPS-IV.

### Q 4.

1. The L2-3 disc space is narrowed with evidence of bony fusion (ankylosis) anteriorly. The normal lumbar lordosis is lost.
2. Pott disease of the spine
3. Erythrocyte sedimentation rate, Mantoux test and chest radiograph.
4. Considering the likelihood of past history of PTB, further investigations (tissue diagnosis using PCR or MRI) are not indicated. A 9-month course of anti-tuberculous drugs was started.

### Spinal Tuberculosis

- This is the most common orthopaedic presentation of tuberculosis.
- Confirmatory tissue biopsy and PCR are not necessary if substantial evidences and past history of pulmonary tuberculosis, are strong.
- Unless neurological deficit is present, anti-TB drugs should be continued and monitored with ESR and clinical improvement.

### Q 5.

1. Spine radiograph shows a straight spine with squaring of anterior border of vertebral bodies and bone formation anterior to the disc. Pelvic radiograph shows absent sacro-iliac joint spaces and hip joint spaces narrowing associated with rough articular surfaces.
2. Ankylosing spondylitis. Other features include a strong family history and associated extra-articular symptoms of iritis or uveitis, inflammatory bowel disease and pulmonary fibrosis
3. HLA-B27 and Rheumatoid factor, ANA and bone scintigraphy.
4. Treatment consists mostly of physiotherapy and NSAIDs or DMARDs or DC-ART. Long-term corticosteroid is associated with osteoporosis and risk of low-energy fracture. Radiotherapy is considered for refractory cases.

### Ankylosing Spondylitis (AS)

- Young adult males are more commonly affected than females in a 4:1 ratio. The incidence of HLA-B27 positive is over 90% of AS patients but only 10% of population with HLA-B27 positive develop AS.
- The principal sites of initial change are bony insertions of ligaments and capsules (entheses). In the spine changes begin at the peripheral parts of annulus fibrosus of the disc and adjacent bones of the thoracolumbar junction
- Spontaneous arrest occurs in many patients but the disease may progress to cause crippling disability.

### Q 6.

1. Dystrophic scoliosis secondary to NF-1.
2. Development of sharp scoliosis and the presence of 2 or more signs of NF-1: at least 5 café-au-lait spots of 5 mm and one plexiform neurofibroma
3. According to the Consensus Development Conference of the National Institute of Health (1987) criteria, other signs include:
  - more than two neurofibromas
  - axillary or groin or base of neck freckles, optic glioma
  - two or more Lisch nodules
  - distinctive bone lesions: long bone cortex thinning with or without pseudoarthrosis, sphenoid wing dysplasia or distortion
  - a first-degree relative with NF-1 by the above criteria

### NF-1 and Spinal Deformity

- A dystrophic curve has high risk to develop neurological complication. It tends to progress even after skeletal maturity and bracing is no longer effective to control progression. Early surgical correction is indicated when dystrophic features are evolving as these indicate modulation from a non-dystrophic to dystrophic spinal deformity, to prevent catastrophic neurological complications.
- Pre-operative MRI is mandatory to exclude other potential causes of neurological complication such as dural ectasia, intraspinal canal tumour and soft tissue mass.