

# Answers and additional information for Clinical Quiz

## Q 1.

- The pelvis, proximal femur and lumbosacral vertebrae appear dense or osteosclerotic. Both hips show coxa vara deformity with the neck of the left femur having a complete fracture. The greater trochanter apophysis has not fused confirming the age of the patient.
- These are classic features of osteopetrosis or marble bone disease.
- The bones are at risk to develop pathological fracture. The surgeon may face problems of difficult internal fixation, delayed or non union and high risk of osteomyelitis. Systemic complications include anaemia and increase susceptibility to infection. Obliteration of neural foramina of the skull base causes cranial nerves compression leading to deafness and blindness. Foramen magnum narrowing may cause early death.
- Treatment modalities include vitamin D (calcitriol), prednisolone, interferon- $\gamma$  and bone marrow transplantation. All have been used with marginal or non-sustainable success.

### Osteopetrosis

- Also known as Marble bone disease or Albers-Schonberg disease. The severe variants of the disease are inherited as autosomal recessive and its benign counterpart is inherited as autosomal dominant. The disorders are characterized by generalized increase in bone density caused by failure of osteoclasts to resorb bone.
- The severe variants manifest as early as the first 3 years of life in forms of severe anaemia or cranial nerve palsies due to obliteration of skull base neural foramina. Bone marrow obliteration also leads to hepatosplenomegaly due to extra-medullary haemopoiesis.
- The benign or adult variant typically presents with fragility fracture and a tendency to develop osteomyelitis. Dental infection may lead to severe osteomyelitis and osteonecrosis of the jaw.
- In contrast to adult-onset osteopetrosis which requires no treatment except for management of fracture, early-onset osteopetrosis needs treatment to minimize the expected complications. High doses of calcitriol may stimulate bone resorption but clinical improvement is either marginal or non-sustainable. Similarly, prednisolone stimulates bone resorption and haemopoiesis but it has long-term adverse effects. Interferon- $\gamma$ 1B helps to improve WBC function and haemoglobin level, thus reducing susceptibility to osteomyelitis. Transplantation of allogenic hematopoietic stem cells seems to be a promising approach for osteopetrosis subtype OPTB3 with missing CAI1 gene.
- Orthopaedic management includes corrective osteotomy for severe deformity, intramedullary nailing for fractures and total joint replacement for premature hip or knee osteoarthritis.

## Q 2.

- Patches of dense sclerotic areas about the left acetabulum as well as head and neck of femur. On CT-scan, there are sclerotic patches on the surface of the neck and within the bone. These lesions resemble dripping candle wax
- This is typical of melorheostosis
- Mostly seen as incidental finding or asymptomatic
- Most patients need no treatment

### Melorheostosis

- It is a rare form of hyperostosis involving overgrowth of the outer layer of bone in a linear pattern similar to wax dripping down the side of a candle, thus acronyms called as candle wax disease. Usually one or more bones on the same limb are affected.
- It is thought caused by a mutation of the LEMD3 gene.
- Commonly asymptomatic. Occasionally with skin changes, progressive deformity and painful limitation of joint motion attributed to soft tissue and skin contracture.
- Patients with severe deformity or progressive pain may need release of contracture and/or osteotomy. In extreme cases either excision of the affected bone and allograft replacement or amputation may be considered.

## Q 3.

- Degenerative changes in both hips indicated by narrowing of joint space and irregular articular surfaces. The pelvis and proximal femora are dotted with small round sclerotic nodules of uniform size predominantly in the cancellous portion.
- This is the characteristic radiographic appearance of osteopoikilosis
- Enostosis or remnants/residual of previously formed but non-resorbed cortical bone of unknown cause but speculated to have relationship to LEMD3. It is a benign lesion usually without symptoms and often discovered incidentally.

### Osteopoikilosis

- A rare inherited bone disorder transmitted as autosomal-dominant trait and characterized by multiple areas of dense calcification throughout the skeleton, producing a mottled radiographic appearance. It may sometimes be confused with osteoblastic metastatic deposits of prostatic or breast carcinoma. However, metastatic deposits are of unequal sizes and typically large.
- Due to its benign and asymptomatic nature, no treatment is necessary.

## Q 4.

- Increased density straddling the sacro-iliac joint bilaterally typically affecting a triangular area of ilium adjacent to the joint.
- This is typical radiological appearance of 'osteitis condensans ilii'.
- A benign incidental lesion seen most often in women who have borne children than in men or nulliparous women. The lesion is attributed to strain of SIJ during labour. It is a rare cause of low back pain with tendency to spontaneously resolve without treatment.
- 'Condensing osteitis' of the clavicle with focal sclerosis of the sternal end of the clavicle.

### Osteitis condensans ilii

- This lesion occurs predominantly in women who have borne children than in nulliparous women or men. It is speculated to associate with strain of the sacroiliac joint during labour.
- A benign lesion in nature and often discovered incidentally on radiograph for other conditions.
- It may sometimes cause low back pain without sciatica and has natural tendency to resolve even without treatment. This is referred as Barsony-Polgar syndrome.

## Q 5.

- Widespread patchy sclerotic or blastic lesions all over the pelvis, sacrum and proximal femur.
- With history of TURP done previously, the likely diagnosis is secondary metastases from prostatic carcinoma.
- Blood investigations including serum acid and alkaline phosphatases, Prostatic Specific Antigen (PSA). Imaging studies: skeletal survey and radioisotope bone scans. Tissue biopsy is important to establish the primary site.
- The prognosis is best predicted by using Gleason score. Tumor marker prostatic carcinoma antigen 3 (PC3) is recently used to predict the prognosis.

### Bone metastasis in Ca prostate

- Bone metastases are characteristically lytic in 12%, mixed in 4% and blastic in 84% of cases.
- Androgen ablation using either non-steroidal (Flutamide) or steroidal (Cyproterone acetate) anti-androgens medications or injections has minimal adverse effects as compared to conventional chemotherapy
- Parenteral bisphosphonates help to deactivate osteoclast-related lytic process to reduce risk of pathological fractures.
- The risk of pathological fracture is reliably predicted by using Mirel scoring system or Campanacci score.
- Impending fractures are strengthened (prophylactically fixed) wherever possible with intra-medullary devices and internal fixation. In some cases, endoprosthesis replacements may be indicated.
- Spinal metastatic lesions need early decompression and fixation with appropriate instrumentation.

## Q 6.

- Generalised density possessing coarse trabeculations involving the pelvis and proximal femur. The left hip 'protrusio' indicates bone softening.
- This is typical of second stage of Paget disease of bone.
- Secondary osteoarthritis and late-onset secondary osteosarcoma due to sarcomatous transformation.

### Paget disease of bone

- A localized remodeling metabolic bone disorder characterized by a marked increase in bone turnover, in which increased osteoclastic activity is followed by excessive osteoblastic activity. This leads to formation of structurally disorganized bone that is mechanically weak and vulnerable to fracture. Biopsy may be necessary to establish the diagnosis.
- The condition is common amongst Western white races though rare in Asians and Africans.
- The disease may involve one bone (monostotic) or many bones (polyostotic). It has variable clinical features with bone pain is most severe at night. Bone enlargement and deformities typically manifest as gait abnormalities. Skin erythema over the affected bone is attributed to hypervascularity of the underlying bone.
- Complications of the disease include pathological fracture, secondary osteoarthritis and late-onset sarcomatous changes.