



QUIZ 2

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**History:**

Young child presented for skeletal survey with suspicion of congenital anomaly on physical appearance.



Image 1: Lateral Skull xray, Image 2: Chest, Image 3:,Pelvis, Image 4: Wrist, Image 5: Arms, Image 6: Spine lateral, Image 7: Legs

**Questions**

- Q1. What is the abnormality in the skeletal survey?
- Q2. What are the differentials of the overall appearance of skeletal survey?
- Q3. What is the most likely diagnosis and how can we confirm?
- Q4. What are complications of this disease?

## QUIZ 2

### Answers

**Answer 1:** Multiple skeletal abnormalities are visualized:

**SKULL:** Large head and J-shaped sella

**CHEST:** Short thick clavicles, Oar shaped ribs thin centrally and expanded peripherally, expanded proximal end of humerus

**PELVIS:** Wine glass pelvis-constricted iliac blades, Elongated pelvis inlet flared iliac wings, Oblique/flat short acetabular roofs, Lateral subluxation of femoral heads, Wide femoral neck and coxa valga

**FOREARM, WRIST AND HANDS:** Short bones of forearm and expanded proximal end, Proximally pointed short metacarpals, inclination of the distal portions of the radius and ulna toward each other

**SPINE:** Widening of intervertebral disk spaces, Ovoid vertebral bodies with central beaking

Platyspondyly, Gibbus deformity at lumbosacral region

**LOWER LIMBS:** Genu valgus, metaphyseal expansion of long bones.

**Answer 2:** Mucopolysaccharidoses (Hurler, Hunter, Morquio, Sanfilippo), Epiphyseal Dysplasias (Multiple epiphyseal dysplasia, Spondyloepiphyseal dysplasia)

**Answer 3:** Diagnosis based on;

- Typical skeletal findings as seen in this case
- Excessive secretion of keratan sulfate in urine
- ELISA test is available to quantify keratan sulfate levels in the urine and blood
- Direct enzyme activity of N-acetyl-galactosamine-6-sulfate-sulfatase in leucocytes or fibroblasts or skin biopsy (Confirmatory test).

**Answer 4:**

- Atlanto-axial instability
- Reduced mobility and pain
- Cervical myelopathy
- Obstructive sleep apnoea
- Hearing deficits
- Visual impairment: corneal clouding
- Dental caries (enamel abnormalities)

### References

1. Parashar Ui, Khanduri S, Bhadury S, Rawat S Roentgenographic diagnosis of mucopolysaccharidosis with particular reference to Morquio syndrome. South African Journal of Radiology 2012; **16(1)**: 32-4.
2. Marlow T, Han T Morquio's syndrome. Applied Radiology 2003; **32(6)**: 44.