QUIZ 2

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Clinical History:
Six years old boy presented with hard swelling over right cervical region for last 2 years. He has limited movement of jaw, neck and back.

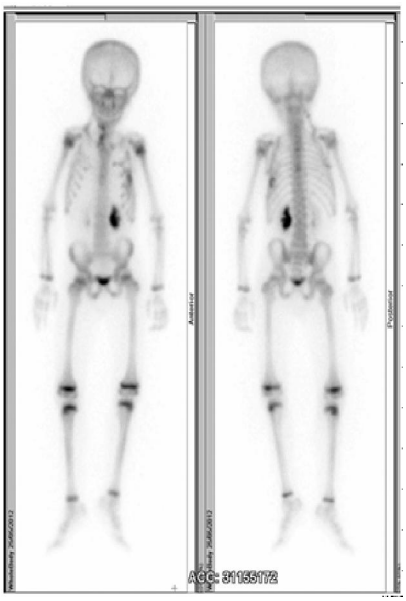

Figure 1

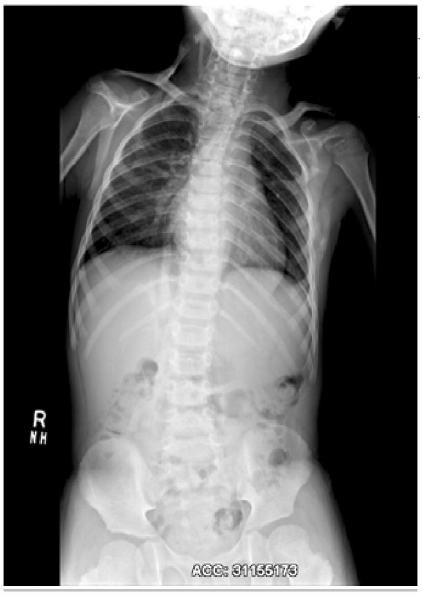

Figure 2

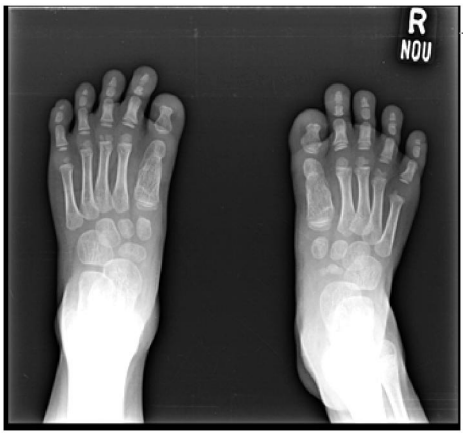

Figure 3

Questions

Q1. What are abnormal findings on whole body bone scan (Figure 1)?

Q2. Define x-ray findings (Figures 2 and 3)?

Q3. What is your diagnosis?

QUIZ 2

Answers

Answer 1: There is evidence of linear shape area of increased uptake over the right cervical region laterally and medially and a well defined area of increased tracer deposit over left chest wall anterolaterally. Hold up of tracer is seen over left renal pelvis (PUJO?).

Answer 2: A well ossified Y shaped abnormal bone is identified in the muscles and soft tissues of the right side neck which is attached proximally to the occipital bone and distally to right clavicle anteriorly. Sheet like extra bony lesions are also identified in the right paravertebral region and soft tissues of the back. Both great toes appear hypoplastic containing single phalanx. Bilateral first metatarsals also appear shorter and broader.

Answer 3: Fibrodysplasia Ossificans Progressiva (FOP).

Discussion

Fibrodysplasia Ossificans Pprogressiva (FOP) is a rare (1 in 2 million people in world), severely disabling, autosomal dominant disease caused by mutations in the ACVR1 gene. FOP is characterized by heterotopic ossification (true bone formation) in the axial musculature, the ligaments, the fascia, the aponeurosis, the tendons, and the joint capsules. One of the signs associated with FOP is malformation of the person's big toes. The abnormality of the person's big toes is a characteristic feature that assists in distinguishing the disorder from other types of muscle and bone problems. Persons affected by FOP may also have short thumbs, or additional skeletal abnormalities.

References

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2. Shore EM, Glaser DL, Gannon FH. Osteogenic induction in hereditary disorders of heterotopic ossification. Clin Orthop Relat Res. 2000; 303-16.

3. Shore EM, Xu M, Feldman GJ, et al. A recurrent mutation in the BMP type I receptor ACVR1 causes inherited and sporadic fibrodysplasia ossificans progressiva. Nat Genet. May 2006;**38(5):** 525-7.