QUIZ

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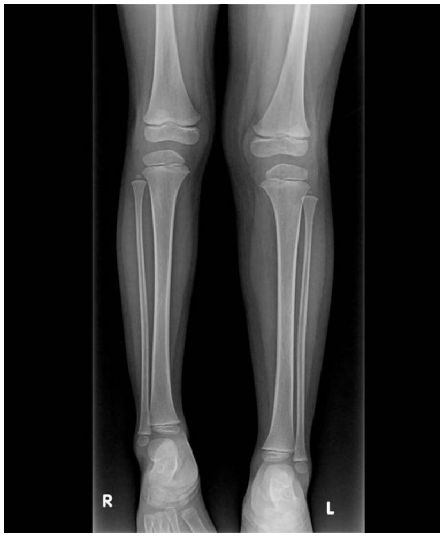
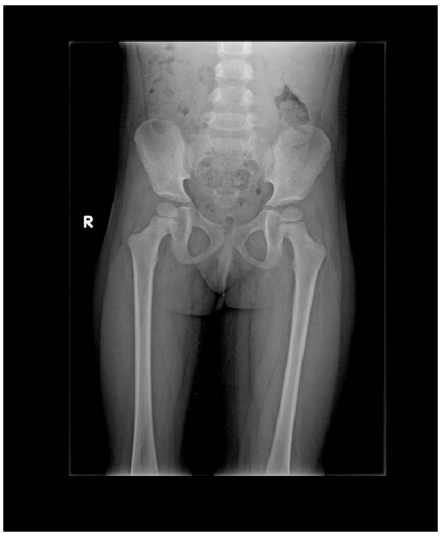
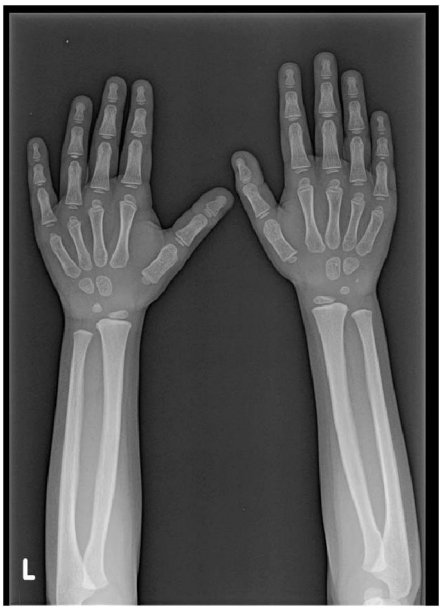
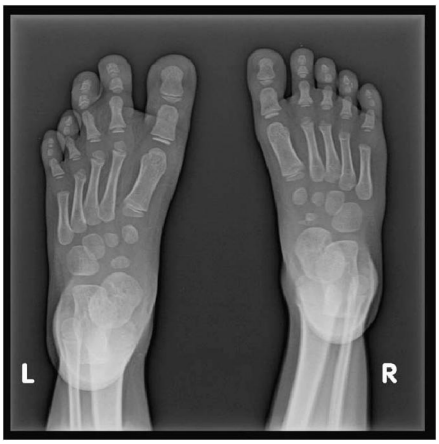
CLINICAL HISTORY : Painless enlargement of left lower limb









Questions

Q1. What are the x-ray findings?

Q2. What is your diagnosis?

Q3. What are the differential diagnoses?

QUIZ

Answers

ANSWER1: There is increased bulk of soft tissue around left knee joint and in the left lower leg. There is increased soft tissue density at the left ankle and in the left foot. There is hypertrophy of the soft tissues in the big toe, 2nd toe and 3rd toe of the left foot. There is evidence of small lucent areas in the soft tissues of the big toe, 2nd toe and 3rd toe of the left foot representing fat. The metatarsals and phalanges of the left big toe, 2nd toe and 3rd toe are broader and longer than those in the right foot. No definite evidence of cortical thickening noted. Long bones of upper limb appear normal. The tibiae and fibulae are normal and symmetrical on both sides.

ANSWER 2: Macrodystrophialipomatosa (Local Gigantism)

ANSWER 3:
i. Neurofibromatosis Type I
ii. Klippel – Trenaunay - weber syndrome.
iii. Hemangiomatosis / Lymphangiomatosis.
iv. Proteus Syndrome.

DISCUSSION : Macrodystrophialipomatosa, hamartomatous enlargement of the soft tissue components leading to localized or generalized gigantism of a limb, is a rare congenital disorder which can present anywhere from infancy to late adulthood. This congenital abnormality occurs most frequently in the distribution of the median nerve in the upper extremity and in the distribution of the plantar nerves in the lower extremity. It is usually recognized at birth or in the neonatal period. The lower limb is more often involved than the upper limb and the 2nd and 3rd digits are more commonly affected.There is a high incidence of associated local anomalies, including syndactyly and polydactyly.As the patient grows, the deformity begins to mechanically interfere with joint function, vascular supply, and innervation. Mechanical problems are not encountered until later in childhood or adolescence, when secondary degenerative changes reduce joint function and compression of the neurovascular structures occurs.
The affected part is increased both in length and width and the skin is thickened, pale, and glossy. Growth of the digit ceases at puberty.The most striking pathologic finding is the increase in adipose tissue interspersed in a fine mesh of fibrous tissue, which involves the bone marrow, periosteum, muscles, nerve sheaths, and subcutaneous tissues. Radiological investigations include plain film radiographs, US and MRI, where findings may be typical. Plain films show lucent soft-tissue overgrowth as well as hypertrophy of osseous structures.
The radiographic differential diagnosis of localized gigantism can be difficult to approach. On the basis of the history, acquired causes of macrodactyly can be eliminated from consideration. The majority of congenital etiologies can also be excluded. Hyperemia secondary to overgrowth of the hemangiomatous and lymphangiomatous elements produces only soft tissue hypertrophy. Neither lymphangiomatosis nor hemangiomatosis show osseous growth. The Klippel-Trenaunay syndrome has obvious cutaneous vascular abnormalities along with macrodactyly. It is characterized by the presence of cutaneous hemangiomas and varicose veins. Patients of Proteus syndrome typically present with skull abnormalities, pigmented naevi, lung cysts, dermatologic changes like palmar and plantar cerebroid thickening and intra-abdominal lipomas.
The most difficult differential diagnosis, both radiographically and pathologically, is neurofibro-matosis. Macrodactyly in patients with Reck-linghausen's diseaseis the result of plexiform-neurofibromas-combined with a mesodermal dysplasia. Several radiographic findings can be helpful in differentiating this condition from macrodys-trophialipomatosa. First, in neurofibromatosis the enlarged digits may be bilateral and the distal phalanges are not always the most severely effected. Second, the hemangiomatous elements of the plexiform-neurofibroma can produce premature fusionof the growth plates . Growth in a digit involved by macro-dystrophialipomatosa ceases with puberty. Third, the enlarged osseous structures in neurofibromatosis may have a wavy cortex and elongated sinuous appearance. Finally, in Neurofibromatosis, besides positive family history, there are caf'e-au-lait spots on the skin and soft tissue nodules.

References

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