QUIZ 1

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History:

Child presented with reduced physical growth. Due to suspicious characteristics physician sent patient for skeletal survey.

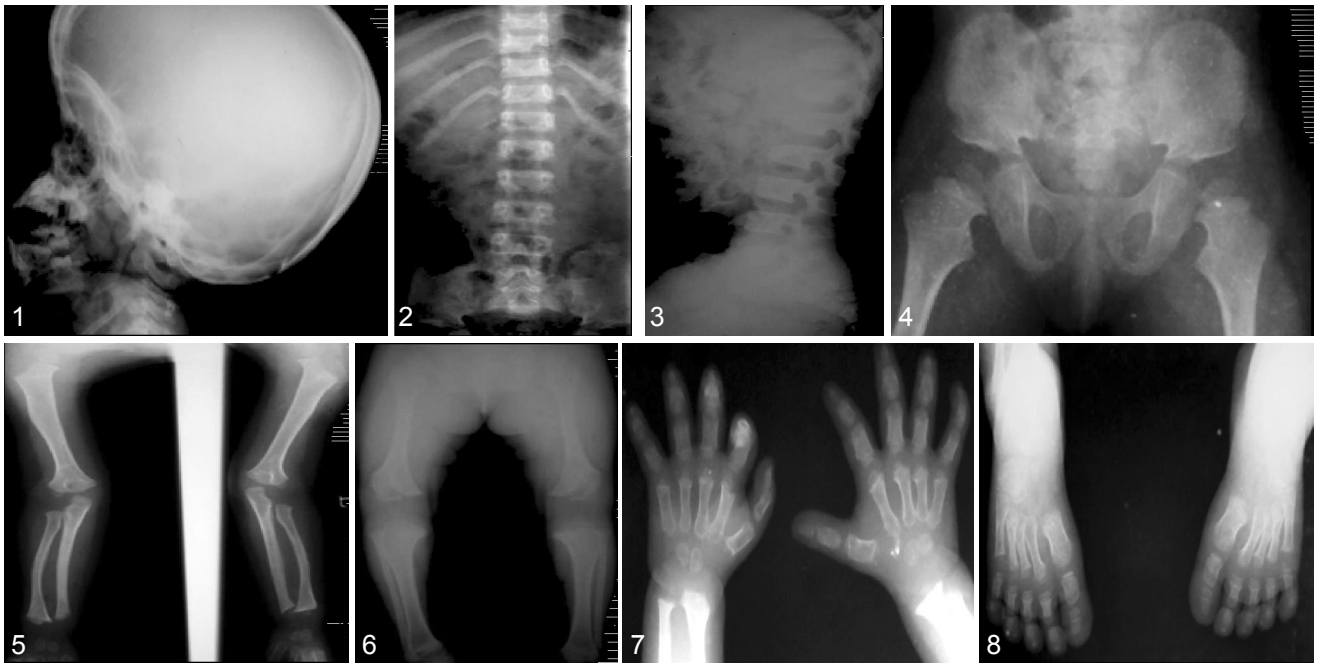

Image 1: Lateral Skull x-ray, Image 2: Spine (AP), Image 3: Spine (Lateral), Image 4: Pelvis, Image 5: Upper limbs, Image 6: Lower limbs, Image 7: Hands, Image 8: Feet

Questions

Q1. What are the skeletal abnormalities on the given images?

Q2. What is the diagnosis?

Q3. What are the differentials of this presentation?

Q4. What is the most common cause of sudden death in these patients and other complications with this disease process?

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Answers

Answer 1: Multiple skeletal abnormalities are visualized:
SKULL: Flat nasal bridge, large calvarium with frontal bossing, constricted basicranium
SPINE: Lateral: hypoplastic bullet / wedge shaped vertebra, rounded anterior beaking of vertebra in upper lumbar spine, scalloped posteriorly concave vertebral margin, wide intervetebral foramina, AP: narrowing of interpedicular space in lower lumbar spine or reversal of interpedicular distance
PELVIS: Square flattened iliac bones (tombstone configuration), champagne glass shaped pelvic inlet, lack of flaring of iliac wings, horizontal acetabula (= flat acetabular angle), Small and short sacrosciatic notches, short femoral necks
Extremities: short stubby limbs, Predominantly rhizomelic micromelia of long bones (femur, humerus), trumpet appearance of long bones = shortening with disproportionate metaphyseal flaring, ball-in-socket epiphysis = broad V-shaped distal femoral metaphysis in which epiphysis is incorporated
HANDS AND FEET: Short stubby fingers, Brachydactyly (short tubular bones of hand + feet), especially short proximal + middle phalanges

Answer 2: Heterozygous Achondroplasia

Answer 3: Homozygous Achondroplasia, mucopolysaccharidoses, thanatophoric dysplasia

Answer 4:

- Neurologic complications (compression of spinal cord and lower brainstem due to constricted basicranium and small foramen magnum resulting in apnea and sudden death)
- Hydrocephalus + syringomyelia (small foramen magnum)
- Recurrent ear infection (poorly developed facial bones)
- Crowded dentition + malocclusion

References

1. Horton WA, Hall JG, Hecht JT. Achondroplasia. Lancet 2007; **370(9582):** 162-72.

2. Radiology Review Manual. Fifth Edition. Wolfgang Danhert. 2003; 41-2.