What is the diagnosis?

Exomphalos. It encompasses a spectrum of abnormalities ranging from a small defect with the gut protruding into the cord (a hernia into the cord) through umbilical defects less than 5cm wide (Exomphalos minor) to larger umbilical defects (Exomphalos major or giant omphalocele). (1) An Omphalocele is a herniation or protrusion of the abdominal contents into the base of the umbilical cord. The herniation of intestines into the cord occurs in about 1 in 5000 births and herniation of liver and intestines in 1 in 10000 births. (2) Although many are isolated defects, some are part of constellation of malformation (such as Beckwith-Weidman syndrome or Trisomy 18), and a few are associated with maternal intake of valproic acid. (3) Prenatal ultrasound detects these defects after 14 weeks gestation as during 1st trimester midgut normally is herniated. (3,4) Surgical closure of the defect may be accomplished with a primary closure or a staged repair once the neonate is hemodynamically stable. (4) Urgent surgery is not indicated for the exomphalos with an intact amniotic sac. Ruptured exomphalos requires urgent surgery. (1).
A 6 years old male child presented with short stature, mouth breathing and snoring. He had height of 94cms (Less than 3rd centile), upper segment to lower segment ratio of 1.2:1 and weight of 14 kgs (Less than 3rd centile). Anterior fontanelle was open measuring 5×6 cms and posterior fontanelle was open measuring 3×3 cms with sutural separation. There were marked frontal and parietal prominences. The eyes were also prominent and protruding but sclera was not blue. The nose was beaked and the nasal bridge was depressed. The teeth were malformed and were in double row with high arched palate. The hands were short and stubby and the nails were broad and dystrophic though all the phalanges were normal. There was mild bowing of the legs and no history of any fractures in the past. On investigation, complete blood count, calcium, phosphorus and alkaline phosphatase were all within normal limits. The growth hormone assay was done in view of short stature and was found to be deficient, even on provocative insulin and L-dopa tests. Radiographs of limbs showed generalized increase in bone density and thickening of bone cortices without obliteration of medullary cavity. The radiograph of skull showed open cranial sutures and fontanelle and increased density of base of skull.

Pycnodysostosis. It is an autosomal recessive disease associated with short stature, fractures, large head with frontal and parietal bossing, open anterior fontanelle and cranial sutures, obtuse mandibular angle, prominent eyes with bluish sclera, underdeveloped facial bones, persistence of deciduous teeth into adulthood often with caries, short, broad hands and feet with dystrophic nails and trunk deformities such as kyphosis, scoliosis, increased lumbar lordosis, and narrow chest. (1) Raised plasma calcitonin concentrations have been reported. (2) A generalized increase in bone density is seen on radiographs, but medullary canals are evident. The skull shows open anterior fontanelle and sutures with small facial bones, non-pneumatised paranasal sinuses and a flattened mandibular angle. Terminal phalanges in the hand are partially or totally aplastic with loss of ungual tufts. The deficiency of growth hormone is due to hypoplasia of pituitary gland caused by the compression of gland by the thickening of sella turcica. Several mutations have been found in the gene encoding cathepsin K, a cysteine protease that is highly expressed in osteoclasts. Dynamic study of tetracycline uptake reveals slowing down of bone resorption and formation. (3) Features which differentiate osteopetrosis from pycnodysostosis

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