dehydration, weakness, hypotension, hypoglycemia, hyponatremia and hyperkalemia are the presenting features of classical salt wasting disease (2-3).These symptoms first develop in the affected infant at around 2 weeks of age and if untreated result in shock and death in few days. Prenatal androgen excess in classical disease, leads to development of ambiguous genitalia in affected females while males appear normal at birth Almost all of these symptoms were present in our case. Treatment consists of glucocorticoid (hydrocortisone 10-15 mg/m2/d) and mineralocorticoid (Fludrocortisone 0.05-0.2 mg/d) replacement and salt supplementation (3). Surgical management of ambiguous genitalia is done for significantly virilized females between 2-6 months of age. (3)

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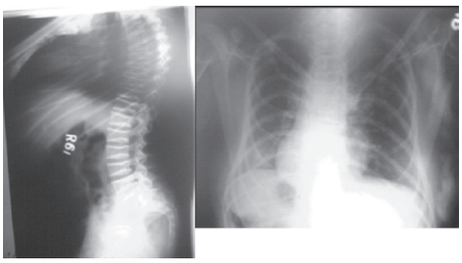
E-published: March 2010

SPOT DIAGNOSIS (IMAGE GALLERY)



Short Stature - A skeletal dysplasia

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A 15 years old girl came to our hospital with short stature. She had a short trunk with upper segment to lower segment ratio of 0.77:1.We worked out on the lines of disproportionate short stature with short trunk. She was normal at birth and first presenting feature was in second decade of life. She had coarse faces, intelligence quotient of 87, bilateral sensorineural deafness, clear cornea, progressive orthopedic anomalies from last 3-4 years, and change in behavior with increase in aggression from last 3 years, disturbed sleep and changing speech from last 2 years. X-rays are depicted in the figure.

What is the diagnosis?

Answer: X-rays are suggestive of Mucopolysaccahroidosis, {MPS}. There was increase in urinary excretion of heparin sulphate. The overall clinical scenario and biochemical markers were suggestive of MPS III {Sanfilippo disease}.

Mucopolysaccahropidosis type III is an autosomal recessive disorder, caused by deficiency in one of the four enzymes involved in the lysosomal degradation of the glycosaminoglycan- heparan sulphate. On the basis of enzyme deficiency there are four different biochemical subtypes, MPS III A, B, C and D with excessive excretion of Heparan sulphate in urine in all these types. Phenotype variations are less common in Sanfilippo than other types of MPS. A milder type may appear totally normal at birth. There is severe progressive central nervous system {CNS} involvement with mild somatic changes. Such disproportionate involvement of the CNS is unique of Sanfilippo. Delay in the diagnosis of MPS III usually occurs because of mild physical features, slow progression of severe CNS involvement and hyperactivity unlike other forms of MPS. Clinical course can be divided into three phases. First phase starts between 1 and 4 years, initially child is normal, later on there is developmental delay. Second phase starts around 3-4 years with severe behavior problems, progressive mental deterioration and dementia. In the third phase behavior problems decreases, there is motor retardation, spasticity and swallowing difficulties. Death usually occurs in 2-3 decade of life, although survival in the fourth decade has been reported {1}.

Presumptive diagnosis is on the basis of clinical and radiological features. Urine screening is done by Berry spot and Acid Turbidity tests. But screening tests may be falsely negative in Sanfillipo. Accurate and confirmed diagnosis is made by 2-dimensional electrophoresis, NMR spectroscopy {2}. Further quantitative estimation of

Heparan sulphate can be done in urine. Other diagnostic tests are enzyme assay, which helps in carrier detection also. Amniotic fluid or chorionic villus biopsy can also be done for prenatal diagnosis in case of a positive family history {3}.

Authorship details: SK diagnosed the case` PG referred the case for the pediatric consultation and managed the skeletal deformities, RK diagnosed the skeletal deformities.

Conflict of Interest: None

Funding: Nil

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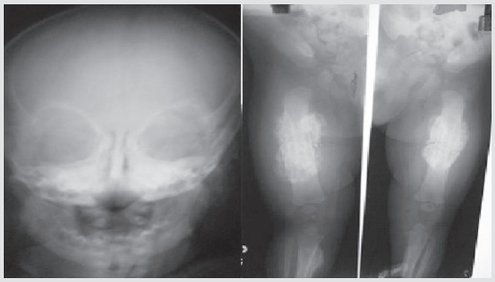
E-published: January 2010

SPOT DIAGNOSIS (IMAGE GALLERY)



Excessive crying and Bone disease

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A 4 month old male child presented with fever, excessive crying and swelling of the face. Plain radiographs were done as shown above.

What is the diagnosis?

Caffey disease or Infantile cortical hyperostosis is a benign, rare, proliferating bone disease affecting infants {1}. Classically, Caffey disease occurs in the first year of life { Less than 5 months}. It is characterized by clinical triad of fever, soft tissue swelling and hyperirritability and a clinching radiograph picture of underlying cortical hyperostosis {2}. Although the etiology of caffey disease remains unclear, many clinical and pathologic features are suggestive of inflammatory process {3}. All races are affected, seen equally in boys and girls {1}. Laboratory investigation may show an elevated ESR, an elevated serum alkaline phosphatase, moderate leukocytosis, thrombocytosis and anemia {4}. Clinical course of caffey disease is highly variable, ranging from self limited to protracted illness. Clinical differential diagnosis includes Osteomyelitis, Parotitis and parotid gland abscess and bone tumors of the affected area. Management is essentially palliative aimed at pain relief but some authors claim a good response to high dose immunoglobulin {5}. Corticosteroids have been used to hasten bone remodeling and Indomethacin has been used to control flare ups.

Contributors credits: AN collected the data and drafted the article and revised the manuscript for important intellectual content. He will act as guarantor of the study. UNR helped in manuscript writing. The final manuscript was approved by all authors

Source of Funding: nil

Competing interests: nil

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E-published: February 2010