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MUCOPOLYSACCHARIDOSIS - CASE REPORT

Paediatrics

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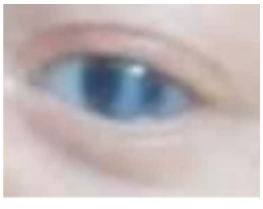
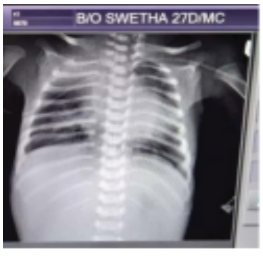
KEYWORDS

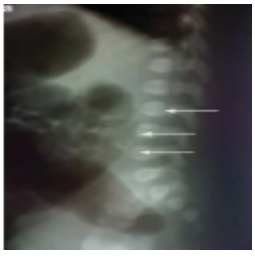
Background Mucopolysaccharidoses are hereditary,progrsesive diseases caused by mutations of genes coding for lysosomal enzymes needed to degrade glycosaminoglycans (acid mucopoly sacch arides),the major GAGs are chondroitin -4 sulphate, heparansu lph ate,chondroitin-6-sulphate,dermatansulphate,keratin sulpha te, hyaluronan

The prevalence of MPS-I in1/1000000 is caused by mutations of the IDUA gene on chromosome 4P16.3 encoding α-L-iduronidase, mutation analysis was revealed 2 major allele, W4O2X, Q70k account for more than half the MPS-I allel's in the white population.

Case report :
A new born born toG3P2L2A1 mother with 2oconsanguinage marriage through the normal vaginal delivery presented with respiratory distress onD1 then on D3 of life baby develops recurrent convulsions, then D5 of life baby develops jaundice, O/E facial dysmorphism ike large bulging head,low set ears, flat bridged nose, with broad tip, claw like hands, corneal clouding present,, on abdominal examination hepatospleenomeglay present, no history of similar complaints in the family.

Investigation :
CBC (TLC16500 increased) RFT, LFT, blood sugars, serum electrolytes normal, CSF – analysis normal, thyroid profile – normal, TMS – Normal 2D Echo – thickening of inter ventricular septum, right ventricular hypertrophy, PAH, (a). X-ray – thick ribs placed horizontally, (b) immature, ovoid configuration of vertebral bodies, neurosonogram – hydrocephalus, urinary GAG's – positive, highly suggestive of muco polysacharidosis.



Treatment :
Injection antibiotics were given according to local antibiotic sensitivity pattern Inj. Mitochondrial cocktail anticonvulsants Phenobarbitone were given, nebulisations with adrenaline were given,

DISCUSSION :
Coarse face, visceromegalyl, corneal clouding X-ray, 2D-Echo findings, urinaryhigh glycosaminoglycon's levels are suggest mucopolysacharidosis.suspecting hurler disease

CONCLUSION :
Prenatal diagnosis for all MPS is carriedout on cultured cells from amniotic fluid (or) chorionic villous biopsy,if any family member has been diagnosed specific

Mutations,chromosomal rearrangements unique to hunter syndrome all other family members should be tested for IDS gene for carrier state .

REFERENCES
1. Nelson Text Book of Pediatrics, 20th Edition.