

Abstract ID: - 85

Abstract Topic: - Clinical Genetics

Abstract Title: - Mucopolysaccharidosis type VII in 2 years male child: A case report.

Presenting author name: - Siddharth Tripathi

Presenting author institute: - MGM Medical college and hospital

Co-authors name: - Dr Sudesh Kumar, Prof(Dr) Nandita Chatopadhyay

Co-authors institute: - MGM Medical College and Hospital, MGM Medical College and Hospital

Aims: - Mucopolysaccharidosis type VII OR Sly disease is occurred due to GUSB gene encoded deficiency of Beta-glucuronidase lysosomal enzyme deficiency which is one of the rarest varieties. [1,2,3]. Due to deficiency of beta-glucuronidase ungraded glycosaminoglycans including chondroitin, dermatan and heparan sulfate accumulate in multiple organs, plasma and urine leading to visceromegaly, skeletal changes and neurological deterioration.

Methods: - Riaz 2 years male child, admitted with progressive abdominal distension and intermittent loose stool for 2 months. Frequency of stool was 8-10 times/day, liquid in consistency without blood. Abdominal distension is progressive. Mother has also complained of swelling of face, trunk and leg. Baby was delivered vaginally at hospital at complete 9-month pregnancy. Child had attained normal development. Family history was not significant.

On examination child was conscious, afebrile. In anthropometry, HC-49cm, height 80 cm and weight 11 kg and mid parental height 166 cm. In general examination no pallor, icterus, edema and lymphadenopathy were noted. In head-to-toe examination rough facial feature, frontal bossing, nasal depression, full lip. Eye hypertelorism, macroglossia, short stubby hand without spine deformity. In systemic examination per abdomen liver was massive enlarge and splenomegaly. other systemic like cardiac, respiratory and neurology was not significant finding.

Results: - Investigation - Complete blood count showed - Hb-10.7 g/dl, RBC- 4.46 lakh/cmm, and total leucocyte count was normal. Kidney, Liver and Thyroid function test was normal. Urine reducing sugar was negative. Abdominal ultrasonography showed hepatosplenomegaly and hypo echoic area in spleen. In fundoscopy no abnormality was found. On the above presentation and basic investigation mucopolysaccharidosis was made as provisional diagnosis.

Outcome: So skeletal survey and urine GAG test was done. In skeletal survey, changes were observed including thickening of skull, deformity of Sella turcica, spatulated ribs, beak shaped vertebrae and proximal tapering of metacarpal. Urine showed for GAG- positive and increased urinary excretion of dermatan and hearan sulfate. The patient was referred to higher center for further treatment where Leucocyte beta -glucuronidase deficiency was diagnosed.

Conclusions: - For the better outcome with progressive metabolic disease, high index of suspicion, early diagnosis and confirmation is required.

Keywords: - Investigation - Complete blood count showed - Hb-10.7 g/dl, RBC- 4.46 lakh/cmm, and total leucocyte count was normal. Kidney, Liver and Thyroid function test was normal. Urine reducing sugar was negative. Abdominal ultrasonography showed hepatosplenomegaly and hypo echoic area in spleen. In fundoscopy no abnormality was found. On the above presentation and basic investigation mucopolysaccharidosis was made as provisional diagnosis.

Outcome: So skeletal survey and urine GAG test was done. In skeletal survey, changes were observed including thickening of skull, deformity of Sella turcica, spatulated ribs, beak shaped vertebrae and proximal tapering of metacarpal. Urine showed for GAG- positive and increased urinary excretion of dermatan and heparan sulfate. The patient was referred to higher center for further treatment where Leucocyte beta -glucuronidase deficiency was diagnosed.