LETTER TO EDITOR (VIEWER'S CHOICE)

INFANTILE GLAUCOMA WITH COARSE FACIAL FEATURES AS AN EARLY COMPLICATION OF HURLER-SCHEIE

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Baby VM was the first child born to the non-consanguineous parents with birth weight of 3600 gm. At 8 months, child developed difficulty in looking during bright light. On examination, she was found to have mild gibbus, claw hand and joint stiffness. Ophthalmic examination confirmed infantile glaucoma with intraocular pressure of 4 kPa in both eyes. She was referred at IHG at 13 months of age with 6,600gms of weight with coarse facial feature, wide opening of mouth with normal physiological and neurological development (Fig 1A). Striking features were noisy breathing, snoring and occasional protruding tongue. Further confirmation of MPS was done by urine Glycosaminoglycans (GAG) quantitative study which was 32 mg/mmol of creatinine (NR: 23.3$\pm$4.1 mg/mmol of creatinine). She was reinvestigated for qualitative urine GAG study by one-dimensional electrophoresis that showed concentrated band at the position of dermatan sulfate. This pattern is seen in urine sample of MPS patient with Hurler/Scheie, Hunter, Maroteaux-Lamy, Sly syndrome, alternatively in multiple sulfatase deficiency or mucolipidosis. Further confirmative study for MPS was carried out by lysosomal enzymes from leucocytes for α-L-Iduronidase, Arylsulfatase-B and β-glucuronidase. The activity in leucocytes was found to be 12.9 nmol\hr\mg of protein (NR: 26 - 54 nmol\hr\mg of protein), 83.88 nmol/hr/mg of protein (NR: 41.82 - 99.48 nmol\hr\mg of protein), and 319.01 (NR: 281.5 - 444.5 nmol\hr\mg of protein) respectively. However, due to mild clinical feature with glaucoma as the only presenting sign no follow-up was made until 4 years of age. Proband developed kyphoscoliosis and hypertrichosis by this time (fig 1-B). Radiographic study of spine showed compressed spinal cord with the possibility of MPS (Fig. 1C). Her head circumference was 49 cms with body weight of 14 kg and normal intelligence. Re-investigation for lysosomal enzymes from plasma, showed low level of α-L-Iduronidase (2.5 nmol/4hr/ml plasma, control value of 17.6 and 22.7 nmol/4hr/ml plasma) with normal Arylsulfatase-B and β-Glucuronidase (42.2 nmol and 116.94 nmol respectively). Leukocyte study for α-L-Iduronidase in the proband showed significantly reduced activity and both parents have confirmed carrier level of alpha-Iduronidase (7.5 nmol in mother and 12.6 nmol in father). Thus, the diagnosis of Hurler/Scheie (I-HS) was confirmed in the proband correlating clinical signs, reduced activity of α-L-Iduronidase and carrier level of enzyme in both parents.

Glaucoma is a known complication in MPS storage disorder; however, as an isolated presenting problem it is rare. In present case, the diagnosis of I-HS was not confirmed until 4 year of age when proband developed kyphoscoliosis as the only skeletal abnormality with normal growth and intelligence. The most common cause of Glaucoma in MPS is the accumulation of dematan sulphate (DS) that interfere with aqueous outflow due to arrest of trabecular meshwork development of the anterior chamber angle.(1) Thickness of the extracellular matrix in sclera and cornea also occurs due to accumulating DS that results in further constriction of the anterior chamber. (2,3) In present case due to milder form of MPS I-H, it is expected to be less accumulation of DS, which is responsible for the distortion of the meshwork in the corneo-scleral junction, lysosomal engorgement and corneal clouding. Intermediate level of alpha-Iduronidase in leucocytes at 13 months also explains the slow progression of the substrate accumulation seen in milder variety of MPS I-H that is Hurler-Scheie. Joint stiffness was noticed in the proband while other features like deafness and valvular heart disease were not apparent. Kyphoscoliosis as a presenting feature at 4 yrs of age is due to MPS accumulation in the dura resulting in compression of the cervical cord. (4) Hence, the only presenting sign of MPS I-HS was glaucoma, which may be more commonly seen as an early complication in Hurler disease.

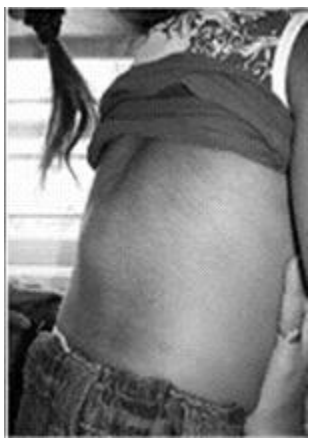

Fig:-1A: Clinical feature of proband at 13 months

Fig:-1B and 1C: Kyphoscoliosis, hypertrichosis and Radiograph at 4 yrs of age

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