Type IV Mucopolysaccharidosis (Morquio Syndrome) – a rarity in Ophthalmology

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Abstract

A 9 years male child who was born of consanguineous marriage was brought with a complaint of progressive painless diminution of vision in both eyes. Detailed ocular examination only revealed bilateral diffuse corneal stromal haze. Systemic evaluation showed coarse facial features, prominent chest and knock knees. Multiple digital X-rays revealed features of mucopolysaccharidosis. Genetic analysis with leucocyte lysosomal enzyme study showed reduced activity of N-acetylgalactosamine-6-sulfate sulfatase suggestive of a rare mucopolysaccharidosis, type IV A (Morquio syndrome). Parental counseling was done regarding the disease inheritance, need for follow up and long term prognosis.

Keywords: Morquio syndrome, mucopolysaccharidosis, corneal clouding