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RESEARCH ARTICLE

CASE REPORT ON EVALUATION OF OPHTHALMOLOGICAL FINDING IN HURLER DISEASE

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ABSTRACT

Aims: The mucopolysaccharidosis (MPS) are a heterogeneous group of rare disorders characterised by accumulation of glycosaminoglycans within multiple organ systems. This case reports evaluates occular findings and manifestations in a hurler disease patient.
Methods and materials: A 8yr old girld diagnosed as a type1 muccopolysaccharidosis presented with systemic manifestations include coarse facies, dysostosis multiplex, hepatosplenomegaly, and intellectual impairment. Ophthalmic findings with visual acuity of 6/12 and 6/9 was having corneal clouding and showing fundus picture of tourtious vessels , optic nerve head swelling. Initial intra ocular pressure was 18mmHg and 20mmHg for right and left eye respectivel and corneal clouding. She also had left eye latent squint of 15degree essotropia and hypertropia. Electroretinogram was done including karyotyping for further evaluation. And case was reviewed.
Results: The patients visual aquity was treated with glasses and regular follow-up of intra-occular pressure and status of cornea and retina was observed.
Conclusion: Ocular complications causing significant reduction in vision are common in MPS. The majority of MPS I and MPS VI patients have corneal opacification, which can lead to difficulties in diagnosis and monitoring of glaucoma, optic disc changes, and retinopathy.

INTRODUCTION

The mucopolysaccharidoses(MPS) are heterogeneous group of rare disorders characterized by accumulation of glycosaminoglycans within multiple organ systems including Eye. The MPS have been historically subdivided into different types depending on clinical manifestations and encompass a wide spectrum of phenotypes, ranging from those disorders, which are fatal in the first months of life to those compatible with a normal lifespan. The MPS result from inherited abnormalities of specific lysosomal enzymes involved in degradation of GAG. Ophthalmic complications are common in MPS, mostly seen in type 1 MPS(Hurlers Syndrome) caused by abnormalities of the enzyme -l-iduronidase.

SUBJECT AND METHODS

Case Report

This 9yr Girl of diagnosed case of Hurler's Syndrome was referred from dep. of Paediatrics for routine eye work-up. She has a phenotypic manifestation of skeletal and respiratory difficulties. On examination she has Hepatoslenomegaly. Her father gives past history of one episode of seizure. She is having a small brother with normal phenotypic manifestation.

Examination

Her visual aquity was 6/12 and 6/9 for right eye and left eye with best possible visual aquity of 6/9 and 6/6 respectively with mild cornea hazyness. Intraocular pressure was 18mmHg and 20mmHg for right and left eye respectively. Left eye was 15 degree esotrophia and hypotrophia.

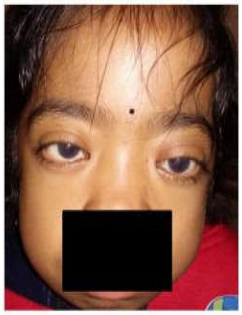

Fig 1: Showing the picture of the Hurler's Syndrome child with 15 degree Eso and Hypotropia

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Fundus finding shows tourchous vessels with optic disc swelling and secondary optic atrophy. With normal O.C.T finding. X-ray shows Skeletal deformity and Claw hand eformity

CT and M.R.I shows *Communicating Hydrocephalus* and *Prominent Sylvian Fissure*. U.S.G whole abdomen shows Hepatomegaly and Splenomegaly. Urinary glycosaminoglycans(GAG) - 171 mg GAG/mmol

TSH – 0.321mIU/l

24 hour urinary protein 0.05 nmol/mg protein/hr with deficiency of leukocyte alpha-L-iduronidase Electroretinogram (ERG) shows reduced b wave and there is normal Visual Evoked Potential

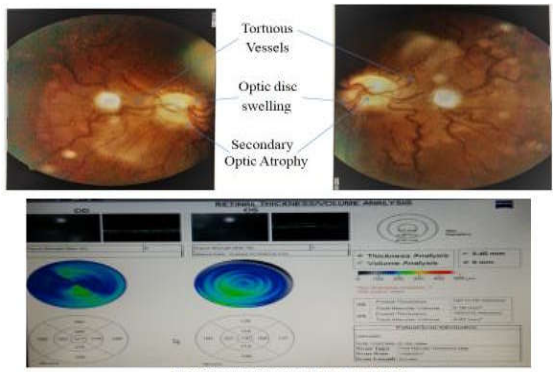

Fig 2: Showing Fundus picture and O.C.T

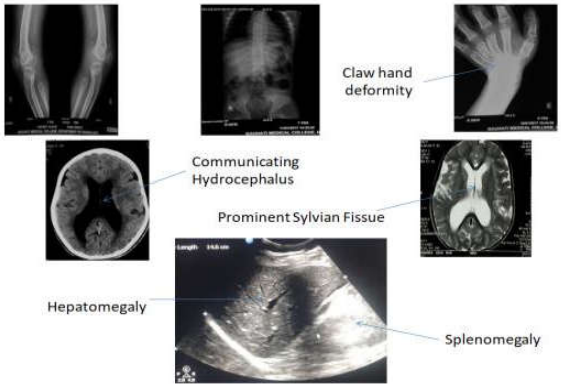

Fig 3: Showing X-ray, CT , M.R.I and USG Whole Abdomen pictures

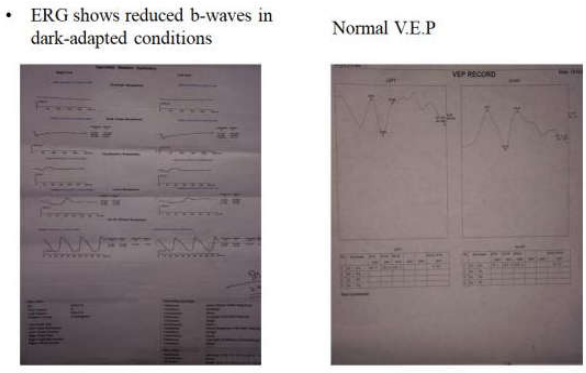

Fig 4: Showing E.R.G with reducrd b-wave and normal V.E.P

RESULTS AND DISCUSSION

Refraction was done and prescribed for glasses with prism bar. Regular monitoring of I.O.P, Corneal status and Retinal changes were monitored on regular follow-up.

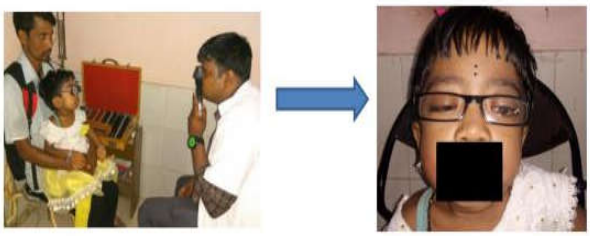

Fig 5: Showing Refraction of the patient and with glasses

CONCLUSION

This study demonstrates that corneal opacification is a common feature in MPS I. Retinopathy with Disc swelling is known to occur to a variable degree in all MPS I subgroups. Optic atrophy seen in minimal cases. Electro diagnostic evidence of retinopathy is seen in this MPS I Hurler patient. There have been no previous studies of the incidence of ocular hypertension or glaucoma in MPS patients. Most of them have open angle glaucoma. Angle closure glaucoma has been reported in few cases. In this case there was not much rise in I.O.P but regular follow-up was done. Visual impairment is common in MPS patients. In this case it was corrected by spectacles. Patients with MPS require regular ophthalmic assessment to detect, monitor, and treating ocular complications. The profound effect of poor vision on a young patient, who may also be suffering multiple physical and intellectual problems, so the ocular management of these patients plays a crucial part of their multidisciplinary care.

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