Clinical Genetics

AB047. Clinical ocular manifestations of Taiwanese patients with mucopolysaccharidoses VI (Maroteaux-Lamy syndrome): a single institution experience

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Background: Mucopolysaccharidoses (MPS) is a group of lysosomal storage disorders caused by specific enzyme deficiency that leads to accumulation of glycosaminoglycan (GAGs). GAGs are widely distributed in many tissues and organs, result in different clinical features. In this study we conducted the manifestation changes of refractive error and corneal clouding in patients with mucopolysaccharidosis VI (MPS VI) under enzyme replacement therapy (ERT).

Methods: A retrospective analysis was carried out on two Taiwanese MPS VI patients treated with ERT during 2001–2012. The clinical characteristics, corneal clouding with slit lamp equipment and refractive error determined by using the ophthalmic refractometer, and corrected visual acuity, and intraocular pressure (IOP) were assessed to understand the evolution of ocular features following treatment with ERT.

Results: In patient 1, hyperopia was noted before and after ERT. There was no obvious improvement of corneal clouding after ERT. In patient 2, hyperopia was initially noted before ERT but unable to be measured due to severe corneal opacity. Clinical observation showed no improvement in corneal clouding after ERT and eventually his best corrected visual acuity worsened and keratoplasty was needed in both eyes. The patient 2 also had ocular hypertension, suspected secondary to MPS VI. However, due to severe corneal clouding, optic disc changes and visual field was unable to be evaluated.

Conclusions: Although many literatures show ERT may be effective in preventing and/or clearing corneal stromal GAGs accumulation, in our experience, no significant improvement of corneal clouding were observed after ERT. Hyperopia and glaucoma were also noted in our patients and without changes after ERT. Severe corneal clouding can lead to difficulties in diagnosing and monitoring severity of hyperopia and glaucoma. It is a great challenge for ophthalmologists and scientists to find out an appropriate systemic evaluation for ocular features in MPS VI patients.

Keywords: Mucopolysaccharidoses (MPS); corneal clouding; refractive error; enzyme replacement therapy (ERT)

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