Sir,

In vivo confocal microscopy of the cornea in Morquio syndrome

Mucopolysaccharidosis type IVa, known as Morquio syndrome, is a lysosomal storage disease associated with corneal haze.[1] A deficiency of *N*-acetyl-galactosamine-6-sulphate sulphatase leads to keratin sulphate deposition in the cornea and other tissues. Predominant systemic features include skeletal abnormalities, hearing loss and dental caries. We report corneal *in vivo* confocal microscopy (IVCM) observations in a case of Morquio syndrome.

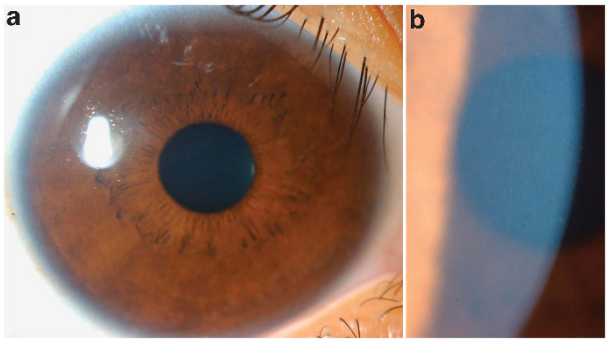

Figure 1 Slit-lamp biomicroscopy of the cornea shows a very mild diffuse fine granular corneal stromal appearance (vision 6/7.5 both eyes).

Case report

A 7-year-old male with Morquio syndrome underwent ophthalmic assessment at the Department of Ophthalmology, University of Auckland, New Zealand. The initial diagnosis was made at 9 months of age following identification of lumbar and lower-thoracic scoliosis. He subsequently developed hearing loss and a constellation of musculoskeletal abnormalities.

Unaided visual acuity was 6/7.5 in each eye and slit-lamp biomiocroscopy revealed a bilateral, diffuse, fine granular corneal stromal appearance (Figure 1). The optic discs and maculae were healthy.

Laser scanning IVCM demonstrated morphological changes throughout the corneal stroma (Figure 2). Immediately posterior to Bowman's layer, the stroma exhibited diffuse, irregular, hyper-reflectivity and in the anterior and mid-stroma, keratocyte cytoplasm was clearly visible with a fine, granular appearance. Keratocyte nuclei were rounded and exhibited vacuolation, particularly in the posterior stroma. The

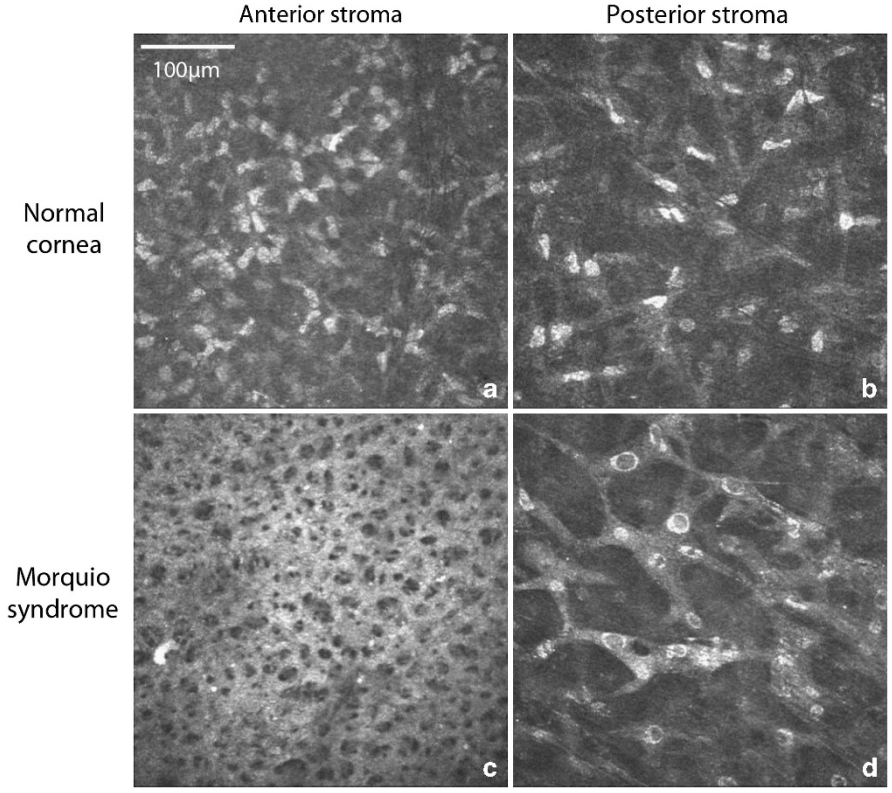

Figure 2 Laser scanning *in vivo* confocal microscopy (IVCM) of the cornea in Morquio syndrome (with normal corneal appearance for comparision) shows diffuse irregular hyper-reflectivity in the anterior stroma, immediately posterior to Bowman's layer. The posterior stroma exhibits rounded, prominent, keratocyte nuclei with vacuolation, and clearly visible keratocyte cytoplasm. (Each IVCM image frame measures 400 μm $\times$ 400 μm; image depth: (a) 68 μm, (b) 416 μm, (c) 66 μm, and (d) 398 μm).

corneal epithelium, sub-basal nerves and endothelium appeared normal.

Comment

This is the first report of IVCM of the cornea in Morquio syndrome. The granular appearance of keratocyte cytoplasm corresponds to the intracellular fibrillogranular inclusions identified by electron microscopy.[2]

In contrast to cross-sectional analyses of pathological specimens in light and electron microscopy, the *en face* images provided by IVCM offer an alternative perspective, enabling additional, non invasive analysis of keratocyte morphology. The appearance of round, vacuolated keratocyte nuclei, predominantly in the posterior stroma, has also been reported in Maroteaux–Lamy[3] and Scheie syndromes.[4] The hyper-reflectivity immediately posterior to Bowman's layer has also been reported in Scheie syndrome.[4]

Previous histological reports have also identified changes in corneal epithelial and endothelial cells.[2,5] The ability of IVCM to image corneas at an early stage of disease may account for the absence of these abnormalities in this case.

This report confirms that IVCM appearances of the cornea in Morquio syndrome are similar to those in other mucopolysaccharidoses.

Conflict of interest

The authors declare no conflict of interest.

References

1 Kasmann-Kellner B, Weindler J, Pfau B, Ruprecht KW. Ocular changes in mucopolysaccharidosis IV A (Morquio A syndrome) and long-term results of perforating keratoplasty. *Ophthalmologica* 1999; **213**(3): 200–205.
2 Iwamoto M, Nawa Y, Maumenee IH, Young-Ramsaran J, Matalon R, Green WR. Ocular histopathology and ultrastructure of Morquio syndrome (systemic mucopolysaccharidosis IV A). *Graefes Arch Clin Exp Ophthalmol* 1990; **228**(4): 342–349.
3 Patel DV, Ku JYF, Kent-Smith B, McGhee CNJ. In vivo microstructural analysis of the cornea in Maroteaux-Lamy syndrome. *Cornea* 2005; **24**(5): 623–625.
4 Grupcheva CN, Craig JP, McGhee CNJ. In vivo microstructural analysis of the cornea in Scheie's syndrome. *Cornea* 2003; **22**(1): 76–79.
5 Leslie T, Siddiqui MAR, Aitken DA, Kirkness CM, Lee WR, Fern AI. Morquio syndrome: electron microscopic findings. *Br J Ophthalmol* 2005; **89**(7): 925–926.

S Stewart, CNJ McGhee and DV Patel

Department of Ophthalmology, New Zealand National Eye Centre, Faculty of Medical and Health Sciences, University of Auckland, Auckland, New Zealand
E-mail: dipika.patel@auckland.ac.nz

Eye (2012) **26,** 1394–1395; doi:10.1038/eye.2012.155; published online 3 August 2012