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The cornea in mucopolysaccharidosis IH-S: structural and ultrastructural study

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Type I mucopolysaccharidoses (MPS I) include three autosomal recessive disorders (Hurler, MPS IH; Scheie, MPS IS; and Hurler-Scheie, MPS IH-S) caused by the deficient activity of the lysosomal hydrolase α-L-iduronidase with the consequent accumulation of dermatan and heparan sulfate in the lysosomes of several cell types [1]. MPS IH-S is an attenuated disease and the patients show minor facial and skeletal dysmorphism, regular intelligence, mild cardiac and respiratory disease, hepatosplenomegaly, and a normal lifespan. The most common feature is corneal opacification [2], whose morphological basis was not studied in detail. In this work we performed a structural and ultrastructural analysis of the cornea in a patient with MPS IH-S. The patient underwent penetrating keratoplasty and the corneal button was immediately processed for light and electron microscopy. From the micrographs a morphometric analysis was also performed.

The corneal epithelium showed superficial cells with few microfolds and evident intercellular spaces. The wing cell layer was formed either by cells with well-evident tonofilaments and small peripheral clear vesicles, or with bilobed nucleus and large paranuclear vesicles filled with granular material. The basal cells showed polygonal shape, with many small vesicles, placed generally in the supranuclear cytoplasm: the intercellular space was enlarged by granular material. The Bowman's layer was either normal in thickness and structure, or thinner and formed by granular material of variable electron density. The stroma was formed by irregular lamellae of differently oriented collagen, by a large number of keratocytes filled with vesicles, and by intercellular granular material. The corneal endothelium showed degenerative changes.

The morphometric analysis of the collagen fibrils diameter provided a mean diameter of 21.71±2.09 nm. Hemidesmosomes were less numerous in the basal cells when compared to the normal cornea. Stromal keratocytes were reduced in their number, particularly in the anterior stroma. Our data showed in MPS IH-S patient pronounced changes of the epithelium, of the Bowman's layer and of the stroma, consistent with the corneal opacity. As the etiology of the disease is a deficiency of α-L-iduronidase and the consequent accumulation of glycosaminoglycans, we are of the opinion that the stromal keratocytes are the first cells to be involved in the pathogenesis of the corneal disease. The accumulation of the aberrant products seems able to induce morphological changes of both the Bowman's layer and the corneal epithelium.

References

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