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# Adult Cystinosis: Electron Microscopy of the Conjunctiva

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**Abstract.** An ultrastructural study was made on the conjunctiva of a 38-year-old man with adult cystinosis. Intracellular cystine crystals were found not only in fibroblasts but also in numerous macrophages. Both fibroblasts and macrophages showed increased pinocytosis. Occasionally there were macrophage degenerations with extracellular cystine crystals present. Whether this increased macrophage reaction indicates a specific difference to childhood cystinosis remains a matter of speculation.

The three types of cystinosis (adult, juvenile, and childhood form) constantly present with crystalline cystine storage in the cornea and conjunctiva. In contrast to the childhood form, adult cystinosis is a benign disorder without renal dysfunction, hyperaminoaciduria, or uveal involvement [1]. The diagnosis can be established by slit-lamp examination. The corneal and conjunctival ultrastructure in a cystinotic child has been studied in detail [2], but to our knowledge electron microscopy of the conjunctiva in the adult form has yet only been mentioned in one [3] of the published cases [1, 3–7].

## Case Report

In 1973, a 38-year-old metal worker of Greek origin (M 502/81) was examined because of a metallic corneal foreign body. Examination revealed glistening crystals in the cornea and aggregates of more yellowish appearing, dotlike crystalline material along the conjunctival blood vessels. Visual acuity was normal, and the patient was not aware of these eye changes. He did not complain about photophobia. He did not show any pigmentary changes in his hair or skin. In 1976 the corneal and conjunctival deposits had not significantly changed. In 1981 we noticed some dilatation of the conjunctival lymph vessels and possibly some increased density of the crystalline deposits, both in the cornea and conjunctiva. Further laboratory tests to exclude nephropathy and multiple myeloma were negative. Urine tests for aminoacids were negative, too. The son of the patient showed no abnormalities clinically and his laboratory tests were also normal.

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## Observations

A biopsy was taken from the bulbar conjunctiva, although the clinical appearance of the crystalline deposits left no doubt about the diagnosis. The specimen was immediately bisected and one part fixed in alcohol, while the other was fixed in glutaraldehyde, postfixed in 2% osmium tetroxide, and embedded in Araldite epoxy resin.

### *Light Microscopy*

Unstained paraffin-embedded sections of the conjunctiva showed birefringent crystals. H & E stained sections as well as 1-μ-thick sections of the glutaraldehyde-fixed material presented accumulations of histiocytic cells, containing "empty" crystalline spaces in their cytoplasm. Extracellular crystals were not obvious.

### *Electron Microscopy*

Ultrathin section of the glutaraldehyde-fixed tissue were stained with uranyl acetate and lead citrate and examined with a Zeiss EM 9 S-2 electron microscope.

Crystalline spaces filled with a fine granular material were found mainly in macrophages but also in fibroblasts (Fig. 1). Both cell types showed vesicle formation in their cell surface. Within the surrounding conjunctival stroma there were deposits of a granular electron dense material, which occasionally was also intracellular in the fibroblasts and macrophages. There seemed to be an active pinocytosis of this material into the macrophages, which were characterized by subplasmalemmal linear densities.

The intracellular crystals were closely related to membrane-limited granular material. Higher magnification of the cytoplasm gave the impression that the crystals developed in these lysosomelike structures (Fig. 2). Those macrophages that contained marked amounts of crystals showed mainly rectangular crystals of varying diameter. Occasionally there were degenerate macrophages with extracellular deposition of crystals (Fig. 3). In one place "empty" spaces of crystalline appearance were noticed between collagen bundles.

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