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Optical diagnosis of a metabolic disease: cystinosis.

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ABSTRACT

Nephropathic cystinosis (NC) is a rare autosomal recessive storage disease characterized by the lysosomal accumulation of cystine crystals throughout the body, particularly in blood cells, the cornea, skin, kidneys, the central nervous system, and the muscles. The skin and the cornea are the most accessible sites to explore, and in vivo reflectance confocal microscopy (IVCM) helps identify crystals in both but does not provide any information to help define their composition. Raman spectroscopy (RS) allows cystine to be easily recognized thanks to its characteristic signature with a band at 499 cm^{-1} . Two dermatology confocal microscopes were used to visualize crystals in both the skin and the ocular surface of a cystinosis patient, and an ex vivo Raman examination of a skin biopsy and of the cornea was performed and removed during a corneal graft to confirm the cystine composition of the crystals. Recently, RS has been performed in vivo and coupled with IVCM. In the future, it is suggested that crystals in NC and other deposits in storage diseases could be identified with this noninvasive in vivo technique that combines IVCM to recognize the deposits and RS to confirm their chemical nature.