**ABSTRACT**

**BACKGROUND:** Patients with infantile nephropathic cystinosis have progressive accumulation of cystine in tissues leading to delayed extrarenal complications. No simple tool is available to evaluate the level of body cystine accumulation. **OBJECTIVE:** We sought to determine the value of in vivo reflectance confocal microscopy of the skin in patients with infantile nephrogenic cystinosis.

**METHODS:** Nine patients and control subjects were recruited for this study. Images were acquired by means of a near-infrared reflectance confocal laser scanning microscope.

**RESULTS:** Scattered bright particles within the papillary dermis were observed in all patients but not in control subjects. The density of particles ranged from numerous (++++) to very few (+/-) and their distribution was heterogeneous. Electron microscopy confirmed that these particles corresponded to cystine crystal deposits within dermal fibroblasts. The density of cystine crystals within the dermis was greater in older patients, in patients with a high leukocyte cystine concentration, and with delayed cysteamine therapy. There was no correlation between the density of cystine deposits and renal disease or hypopigmentation but high levels of deposition occurred in association with extrarenal manifestations.

**LIMITATIONS:** This is a preliminary study on a small sample of patients. Repeated examination and longer follow-up is necessary.

**CONCLUSION:** In vivo reflectance confocal microscopy of the skin appears to be a noninvasive means of assessing body cystine accumulation in infantile cystinosis and could be used as a complementary marker of treatment response in addition to leukocyte cystine measurement.