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### Coupling innovative imaging: in vivo multilaser confocal microscopy and ex vivo confocal Raman spectroscopy of cornea and skin in nephropathic cystinosis

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#### ABSTRACT

**Purpose:** Nephropathic cystinosis is a rare, autosomal-recessive inherited disease, characterized by lysosomal accumulation of cystine crystals in almost all tissues. Aim: to describe an innovative in vivo confocal microscopy (IVCM) of crystals in the skin, the cornea and the conjunctiva as well as raman spectroscopy of the crystals in skin and cornea **Methods:** A 36 year-old women with advanced nephropathic cystinosis underwent penetrating keratoplasty for severe visual loss and chronic ulceration in her left eye. The only dermatology symptom was skin dryness. Cornea and skin was analysed with IVCM using the innovative multilaser (488, 658 and 785 nm) confocal microscope Vivascope 1500 and the handeld monolaser Vivascope 3000 (MAVIG GmbH). In order to obtain the chemical composition, ex vivo Raman spectroscopy (LabRAM ARAMIS,Horiba Jobin-Yvon,France) was performed on corneal button retrieved during keratoplasty and on a skin sample, both immediately frozen in liquid nitrogen without adjuvant

**Results:** Multilaser and monolaser IVCM showed reflective crystals in the corneal epithelium, stroma, tarsal conjunctiva and forearm skin with the highest resolution obtained at 488nm. Ex vivo Raman spectra were obtained in skin and cornea **Conclusion:** Combination of IVCM with Raman spectroscopy may improve the diagnosis and follow-up for other metabolic diseases with skin and corneal thesaurismoses like amylosis, Wilson disease, Fabry disease or mucopolysaccharidosis