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Role of Handheld In Vivo Reflectance Confocal Microscopy for the Diagnosis of Fabry Disease: A Case Report.

Cinotti E, Provvidenziale L, Fimiani M, Perrot JL, Cambazard F, Rubegni P.; Diseases. 2018 Jun 27;6(3). pii: E55. doi: 10.3390/diseases6030055.

ABSTRACT

Fabry disease (FD) is a rare X-linked lysosomal storage disorder caused by the deficient activity of the lysosomal enzyme α -galactosidase that leads to a systemic accumulation of globotriaosylceramide. Handheld in vivo reflectance confocal microscopy (HH-RCM) is a useful modern technique in diagnosis and follow-ups of many skin diseases. This noninvasive device provides high-resolution and high-contrast real-time images to study both the skin and the ocular surface structures that can help clinicians to confirm the diagnosis of FD. HH-RCM could be helpful even for the follow-ups of these patients, enabling us to monitor the effect of enzyme replacement therapy on corneal cells and keratinocytes.

KEYWORDS: confocal microscopy; diagnosis; fabry disease; lysosomal diseases; screening

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