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Characterization of X-linked Hypohidrotic Ectodermal Dysplasia (XL-HED) Hair and Sweat Gland Phenotypes Using Phototrichogram Analysis and Live Confocal Imaging

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ABSTRACT

Hypohidrotic ectodermal dysplasia (HED) is the most common type of ectodermal dysplasia (ED), which encompasses a large group of syndromes that share several phenotypic features such as missing or malformed ectodermal structures, including skin, hair, sweat glands, and teeth. X-linked hypohidrotic ectodermal dysplasia (XL-HED) is associated with mutations in ectodysplasin (EDA1). Hypohidrosis due to hypoplastic sweat glands and thin, sparse hair are phenotypic features that significantly affect the daily lives of XL-HED individuals and therefore require systematic analysis. We sought to determine the quality of life of individuals with XL-HED and to quantify sweat duct and hair phenotypes using confocal imaging, pilocarpine iontophoresis, and phototrichogram analysis. Using these highly sensitive and non-invasive techniques, we demonstrated that 11/12 XL-HED individuals presented with a complete absence of sweat ducts and that none produced sweat. We determined that the thin hair phenotype observed in XL-HED was due to multiple factors, such as fewer terminal hairs with decreased thickness and slower growth rate, as well as fewer follicular units and fewer hairs per unit. The precise characterization of XL-HED phenotypes using sensitive and non-invasive techniques presented in our study will improve upon larger genotype-phenotype studies and the assessment of future therapies in XL-HED.