Mutations in SULT2B1 tied to ichthyosis in humans

June 5 2017

(HealthDay)—Scientists have discovered another gene mutation behind

certain cases of autosomal-recessive congenital ichthyosis (ARCI), according to a report published in the June 1 issue of the *American Journal of Human Genetics*.

Keith Choate, M.D., Ph.D., of the Yale University School of Medicine in New Haven, Conn., and colleagues utilized whole-exome sequencing and multigene panel screening to identify four distinct mutations in ARCI, including missense, nonsense, and splice site mutations.

The team noted loss of *SULT2B1* expression at RNA and protein levels in keratinocytes. They then reconstructed the morphologic skin alterations in a three-dimensional organotypic tissue culture model with *SULT2B1*-deficient keratinocytes and fibroblasts. Using thin layer chromatography, they demonstrated absence of cholesterol sulfate, and an increased level of cholesterol.

"Our study reveals an essential role for *SULT2B1* in the proper development of healthy human skin," the authors write. "Mutation in *SULT2B1* leads to an ARCI phenotype via increased proliferation of human keratinocytes, thickening of epithelial layers, and altered epidermal cholesterol metabolism."

More information: Abstract/Full Text (subscription or payment may be required)