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Schnyder corneal dystrophy-Abnormal cholesterol homeostasis in a connective tissue

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Schnyder corneal dystrophy (SCD) is characterized by accumulation of lipids and lipid particles in the cornea similar to those lipid particles that accumulate in human atherosclerotic plaques. Cholesterol and phospholipid accumulate in the extracellular connective tissue space of both diseases in the form of crystals, liposomes, and droplets. Thus, there may be some pathogenic similarities between these disease-causing lipid accumulations. Cholesterol accumulation can result from an imbalance in deposition and removal. Some genetic disorders affecting HDL function (LCAT, ABCA1, and apoAI deficiencies) also result in corneal lipid deposition with similarities to SCD. In this regard, HDL-associated apolipoproteins (apo), apoAI and apoE, show increased accumulation in Schnyder corneas, and UBIAD1, the defective gene in SCD, interacts with the C-terminal portion of apoE. Because HDL functions to remove cholesterol from tissues, it is possible that cholesterol removal from the SCD corneas is impaired. On the other hand, UBIAD1, a prenyltransferase domain-containing protein, shows similarities to genes involved in regulation of the cholesterol synthesis pathway, and interacts with HMG-CoA reductase, the rate-limiting enzyme in cholesterol synthesis. Thus, it is also possible that increased cholesterol synthesis is the cause of cholesterol accumulation in SCD corneas. The predominantly mitochondrial location of UBIAD1 in corneal keratocytes suggests a possible link between mitochondrial and cholesterol metabolism. Identification of the gene defect in SCD makes it possible to test these different potential mechanisms of cholesterol accumulation, and to learn more about the regulation of cholesterol homeostasis in the cornea and other similar connective tissues such as the blood vessel wall.

Biography

Xueting Jin obtained her M.D. from the Medical Center of Fudan University, Shanghai, and qualified as a physician in 2012. During medical school, she participated in several research projects, mainly focused on the mechanism of atherosclerosis. She is currently a postdoctoral fellow at the National Heart, Lung, and Blood Institute, NIH, Bethesda, Maryland. Her research interests include disorders of cholesterol metabolism, focused on Schnyder corneal dystrophy and atherosclerosis.

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