New insight into RASopathy-associated lymphatic defects

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The RAS pathway is a cellular signaling pathway that regulates growth and development in humans. RASopathies are a group of diseases characterized by defects in RAS signaling.

Many patients with RASopathies present with defects in the lymphatic system, which removes excess fluid from tissues, absorbs fats from the digestive system, and transports immune cells.

To determine how alterations in the RAS pathway affect development of the lymphatic system, researchers at Yale University generated transgenic mice that expressed mutations associated with a RASopathy known as Noonan syndrome.

In this issue of the Journal of Clinical Investigation, Michael Simon and colleagues report that excess RAS pathway activation triggers increased activity of a protein known as ERK.

Mice with RASopathy-associated mutations exhibited lymphatic defects similar to those seen in humans, but the defects could be reversed by treatment with an ERK inhibitor.

These findings demonstrate that excessive ERK activation underlies lymphatic defects in RASopathies and suggest that ERK inhibition could be a useful therapeutic strategy.

More information: Endothelial ERK signaling controls lymphatic fate

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