DDC
Texas Medical Center Digestive Diseases Center

WEEKLY GI RESEARCH WEBINAR

"Dosage-sensitive modifiers of Notch signaling in a mouse model of Alagille syndrome"

Hamed Jafar-Nejad, M.D.

Associate Professor, Department of Molecular and Human Genetics

Baylor College of Medicine

Alagille syndrome (ALGS, OMIM #118450) is a multi-system developmental disorder affecting the hepatobiliary and cardiovasclar systems, among others. The majority of ALGS patients harbor dominant mutations in components of the Notch signaling pathway (~95% JAG1, ~1-2% NOTCH2). However, there is extreme variability in disease presentation and prognosis that cannot be explained by the nature of the causative mutations. Moreover, no mechanism-based therapies are available. Using a mouse model for ALGS which we established several years ago, we seek to identify and characterize dosage-sensitive genetic modifiers of the ALGS phenotypes, hoping to shed light on its pathophysiology and to establish potential therapeutic targets.

References: (1) Thakurdas S, Lopez M, Kakuda S, Fernandez-Valdivia R, Zarrin-Khameh N, Haltiwanger RS, and Jafar-Nejad H (2016). Jagged1 heterozygosity in mice results in a congenital cholangiopathy which is reversed by concomitant deletion of one copy of Poglut1 (Rumi). Hepatology 63(2): 550-65. (2) Adams JM and Jafar-Nejad H (2019). The roles of Notch signaling in liver development and disease. Biomolecules. 9: 608 (review) (3) Adams JM and Jafar-Nejad H (2019). The roles of Notch signaling in liver development and disease. Biomolecules. 9: 608 (review)

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