Ashwani K Singal, J Genet Syndr Gene Ther 2017, 8:3 (Suppl)
DOI: 10.4172/2157-7412-C1-014

3rd Annual Congress on
RARE DISEASES AND ORPHAN DRUGS
October 30-November 01, 2017 San Antonio, USA

Ashwani K Singal
University of Alabama at Birmingham, USA

Mitochondrial bioenergetics in porphyria: Studies in peripheral blood cells

Porphyria is a group of metabolic disorders due to altered enzyme activities within the heme biosynthetic pathway. It is a systemic disease with multiple potential contributions to mitochondrial dysfunction and oxidative stress. Recently, it has become possible to measure mitochondrial function from cells isolated from peripheral blood (cellular bioenergetics) using the XF96 analyzer (*Seahorse Bioscience*). Using various inhibitors and activators of mitochondrial respiration, this technique measures various components of O2 consumption rate (OCR) in peripheral cells such as basal, ATP linked, proton leak, maximal, reserve capacity, non-mitochondrial, and oxidative burst, all measured as pmol/min./100,000 monocytes. We performed cellular bioenergetics on 18 porphyria (9 PCT, 6 acute, and 3 protoporphyria) patients and 39 age/gender matched healthy controls. Of porphyria cases, 5 were active (1 PCT and 4 acute) and 13 in biochemical remission. Monocyte bioenergetics was significantly decreased in active porphyria vs. porphyria in remission and vs. healthy controls. Among 6 acute porphyria, a negative correlation (-0.8 to -0.93) was observed between urinary porphobilinogen and various components of monocyte OCR. In two pseudporphyria patients, monocyte OCR was similar to healthy controls and higher than active porphyria. These novel and interesting preliminary findings suggest existence of mitochondrial dysfunction in porphyria and potential non-invasive biomarker for disease activity. Studies are suggested to examine mechanisms of these findings as basis for deriving mitochondrial based therapies in management of porphyria.

Biography

Ashwani K Singal is working as Associate Professor of Medicine in division of Hepatology and Director of Porphyria Center at the UAB, Birmingham AL. His clinical research interests include alcohol and non-alcohol fatty liver disease, porphyria, and renal dysfunction in liver cirrhosis. He has over 110 publications, on editorial board of reputed journals, and research award committees of the AGA and AASLD. His research is funded from the Transplant Institute of the UAB, ACG, NIAAA and NIDDK from the NIH, and pharmaceutical industry.

aksingal@uab.edu
ashwanisingal.com@gmail.com

Notes: