Natasa Chrysodonta, J Clin Exp Cardiolog 2016, 7:6(Suppl)
http://dx.doi.org/10.4172/2155-9880.C1.035

An update on the management of Alagille syndrome

Natasa Chrysodonta
Hinchingbrooke Hospital, UK

Alagille syndrome is an autosomal dominant disorder, also known as arteriohepatic dysplasia Alagille-Watson syndrome, or syndromic bile duct paucity. The syndrome expressivity is highly variable but when fully expressed patients have cardiac malformations, skeletal and ophthalmological abnormalities in conjunction with cholestasis and bile duct paucity. It has been identified that the multisystem involvement is due to defects in the Notch signaling pathway, with the main mutation identified in JAG1. Despite relative good prognosis, mortality by the age of 20 years reaches 70%. The major contributor to the previous is the complex congenital heart disease in addition to the hepatic pathology in these patients. This emphasizes the need for early and appropriate treatment in this population. This review examines the evidence surrounding the management of this syndrome, primarily from a cardiovascular perspective.

Biography

Natasa Chrysodonta is currently a foundation year 2 doctor in the United Kingdom. She has completed her medical degree at the University of Bristol and is currently undertaking an MSc in Genomic Medicine in Queen's Marry University of London.

nc8636@my.bristol.ac.uk

Notes: