P-147 - SITOSTEROLEMA IN COSTA RICA: REPORT OF THE FIRST CASE

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**INTRODUCTION:** Sitosterolemia is a rare autosomal recessive disorder of lipid metabolism characterized by increased intestinal absorption and a decrease in the biliary excretion of cholesterol and plant sterols such as sitosterol. Increased plasma concentrations of plant sterols (especially sitosterol, campesterol, and stigmasterol) are observed once foods with plant sterols are included in the diet and have accumulated in the body. ABCG5 and ABCG8 are the only genes in which pathogenic variants are known to cause sitosterolemia. Because the proteins sterolin-1 (encoded by ABCG5) and sterolin-2 (encoded by ABCG8) form a heterodimer transporter, affected individuals have biallelic pathogenic variants in either ABCG5 or ABCG8. Affected individuals can develop: a) Xanthomas (cholesterol deposits) in the tendons and around the pressure points, b) Hypercholesterolemia, atherosclerosis of early onset and c) Hematological abnormalities, including hemolytic anemia

**OBJECTIVE:** We present a 36-year-old female patient with a history of hypercholesterolemia, with painful masses in the Achilles tendon, knees and hands, and joint pain since childhood, with the family history of consanguineous parents and a younger brother with similar but milder symptoms. Description of the first biochemically and molecularly confirmed case of Sitosterolemia in Costa Rica.

**MATERIALS AND METHODS:** Biochemical analysis including: Serum lipoprotein electrophoresis, cholesterol, cholestanol and triglyceride measurements was performed. Hemoglobin analysis and Next Generation Sequencing (NGS) studies were performed. Imaging tests such as abdominal ultrasound and brain magnetic resonance imaging (MRI) were performed.

**RESULTS:** The proband had large xanthomas at the level of heels and Achilles tendon, elbows, knees and hands. Abdominal ultrasound and brain MRI do not report changes. Clinically there were no neurological symptoms. Biochemically she presented abnormal lipoprotein electrophoresis with elevated ultrasensitive CRP, hypercholesterolemia due to elevation of LDL. Molecular study reported a homozygous, pathogenic variant. c.647_657dupGTGAGCGAGG (p.Arg220Alfs*37) in the ABCG8 gene. Therapy with a restricted sterol diet and Ezetimibe was started.

**DISCUSSION AND CONCLUSIONS:** Sitosterolemia is a defect in the metabolism of cholesterol and plant sterols such as sitosterol, which can manifest from childhood and worsen with aging. The right diagnosis will allow a better approach and the correct management, as in this case.