GENETICS AND GENODERMATOSES

SITOSTEROLEMIA: A DIAGNOSIS OF EXCLUSION?

Isha Singh [(1)]

Mgm Hospital Kamothe, Dermatology, Mumbai, India [(1)]

Background: Sitosterolemia is a rare autosomal recessively inherited disease caused by mutations affecting ABCG5 or ABCG8, which are located on human chromosome band 2p21. Around 100 cases have been reported in the literature wordwide till date.)

Observation: The usual clinical enzymatic colorimetric methods cannot discriminate between cholesterol and plant sterols leading to difficulty in diagnosis. We report an 18 year old female born of a non-consanguineous marriage, residing in India, presenting with multiple plane-palmar, intertriginous, eruptive, tendinous xanthomas, xanthelesma with multiple joints pain and difficulty in doing daily chores. Patient had a short stature. Patient was an isolated case in her family with such extensive lesions. And her diagnosis was and still is very challenging.

Keynote: Sitosterolemia shares several clinical characteristics with familial hypercholesterolemia. The clinical manifestations include large painful xanthomas, arthritis, premature atherosclerosis, short strature, anemia, and macrothrombocytopenia. Prompt recognition of these symptoms is important to prevent fatal medical conditions like coronary heart disease and pancreatitis.