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EC CLINICAL AND MEDICAL CASE REPORTS

Opinion

Tangier Disease: The Current Clinical Treatment Approach

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Received: November 02, 2022; **Published:** December 29, 2022

Tangier disease is a rare, autosomal recessive storage disorder of serum lipoprotein metabolism. The disease is named after an island off the coast of Virginia (Tangier island Chesapeake Bay) where the first patients were identified. Esterified cholesterol is found to accumulate in macrophages and Schwann cells of peripheral nerves, plasma cholesterol is observed to be low and plasma high density lipoprotein almost completely absent [1]. The large build up of esterified cholesterol occur in tissues like the arteries, liver, spleen, and tonsils leading to clinical manifestations such as coronary artery diseases, muscle wasting, peripheral neuropathy, reduced visual acuity, hepatomegaly, splenomegaly, lymphadenopathy and enlarged yellow tonsils [2].

Major serum abnormalities observed in homozygous abnormal patients include hypertriglyceridemia, hypocholesterolemia and decreased apolipoprotein A-I (apoA-I) concentrations [3].

The molecular defect in Tangier disease has been explained by some propounded hypotheses namely (a) accelerated catabolism of high density lipoproteins (HDL) despite normal rates of apoA-I synthesis, (b) abnormal structure of apoA-I, (c) impaired interconversion of proapoA-I to mature apoA-I and (d) a failure of proapoA-I to associate with HDL [3].

The cause of the disorder is as a result of mutations in the cell membrane protein ABCA1 (ATP binding cassette transporter) on chromosome 9q31 [4,5]. This protein is responsible for the secretion of excess cholesterol from cells into the HDL metabolic pathway resulting in a profound deficiency of HDL followed by accumulation of cholesterol in tissue macrophages.

The disease symptoms may include: atherosclerosis, dry skin, corneal clouding, mild hypertriglyceridemia, stomach pain and type 2 diabetes [6].

The diagnosis of the disease involves blood tests for high density lipoprotein (HDL) and one of its proteins [apolipoprotein A1 (ApoA1)], genetic testing and biopsy. Some of such tests include electromyograms (nerve and muscle studies), abdomen and carotid arteries ultrasounds. angiogram of the heart, echocardiogram, exercise stress test, and eye examination [7].

The current clinical treatment approach indicates no specific treatment for Tangier disease. However, reports show that management of the disease may involve (i) surgery to remove an affected area, such as tonsils or spleen (ii) foods that raise HDL level such as avocados, beans, fatty fish, fiber fruits, nuts, whole grains, and olive oil (iii) life style measures that may include frequent exercising, eating monounsaturated fat and not saturated type, maintaining healthy weight, and avoiding tobacco products (iv) use of therapeutic agents and such potential agents include miglustat [8] and cholesterol lowering agents.

In conclusion, although Tangier disease currently seems to be untreatable disorder, report indicating miglustat to have ameliorate effect on some clinical manifestations of the disease may trigger new research interest in investigating for pharmacological active agents against the disease.

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Volume 6 Issue 1 January 2023