



SKIN MANIFESTATIONS OF INTERNAL DISEASE

## FAMILIAL DYSBETALIPOPROTEINEMIA- A CASE REPORT

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**Background:** Xanthomas are a common presentation of disorder of lipid metabolism usually associated with abnormalities of cholesterol metabolism. Tuberous xanthomas are commonly associated with hypercholesterolemia, increased levels of LDL and familial dysbetalipoproteinaemia. Tendinous xanthomas are found in association with severe hypercholesterolaemia and elevated levels of LDL, typical of pure hypercholesterolaemia and FH.

**Observation:** A 27 year old male patient presented with multiple, painless, firm, yellowish plaques and nodules over buttocks, elbow joints, knee joints, foot and metacarpophalangeal joints of various size and shape. CVS examination revealed class III symptoms MI and Impending cardiac failure with CAD effort Angina class III with impending failure. Lipid profile showed Sr.Cholesterol 740 mg/dl Sr. TGL-563 mg/dl and Sr. LDL-589 mg/dl. Histopathological features are consistent with xanthoma. Similar findings were observed in his father and elder sibling. Clinical features and laboratory values indicated the diagnosis of familial

Dysbetalipoproteinaemia. This case is presented for its rarity.

**Key message:** Xanthomas are a disorder of lipid metabolism usually associated with abnormalities of cholesterol metabolism can give us a clue for underlying medical conditions.

