

AN ISOLATED CASE OF HYPERLIPOPROTEINEMIA RESEMBLING TYPE II

B. M. S. BEDI,* A. CHANDRASEKAR,† K. VENKATESAN,‡ K. BALASUBRAMANIAN,‡
H. M. JAGANATHA,‡ V. ANANDAKRISHNAN‡ AND S. RAMAKRISHNAN ||

Summary

An isolated case of Frederickson's Type II a like-hyper-lipoproteinemia with cutaneous xanthomas and aortic stenosis was observed in a 14 year old girl of a South Indian family from Seven Malai, Kerala State. She had an increased serum cholesterol level, hyperlipidemia, hyperlipoproteinemia, normotriglyceridemia and tuberous and tendinous xanthomatosis with aortic stenosis. The members of three generations of her family were normal.

Lipoprotein disorders are classified into two major groups (1) hypolipoproteinemia and (2) hyperlipoproteinemia. Both these disorders can be familial as well as acquired. Frederickson^{1,2} has classified hyperlipoproteinemia into 5 main types based on the changes in the serum lipoprotein spectrum and other associated changes in the lipids of the body. Type II hyperlipoproteinemia is characterised by hypercholesterolemia, hyper or normotriglyceridemia and a prominent β -band on the lipoprotein electrophorogram. This type has been observed in a 14 year old girl from a South Indian family living in Seven Malai, Kerala. Available

members of the family in three generations have been thoroughly investigated for any lipoprotein disorder.

Case Report

A 14 years old girl, came to the department of Skin & V. D. of Jawaharlal Institute Hospital for treatment of persistent skin eruptions. On examination, yellowish orange papular lesions of various sizes and plaques were seen in the interdigital areas of hands, over the ankles, knees, wrists, front and back of elbows, right shoulder joint and both gluteal regions. The lesions were present from the age of 3 years, and had been progressive. Examination revealed arcus senilis bilaterally. Auscultation of the heart revealed a grade II ejection systolic murmur over the aortic area II conducted to carotid. Skin showed tuberous & tendinous xanthomata characteristic of type II hyperlipoproteinemia. Serum lipid profile showed serum cholesterol of 800 mg% with an increase in β -lipoprotein fraction. Triglyceride levels were normal. G. T. T. was also normal. Skin biopsy showed histological features of xanthoma. The

* Associate Professor & Head of the Dept. of Skin & V.D.

† Senior Resident, Department of Skin & V.D.

‡ Department of Biochemistry

|| Professor & Head of the Dept. of Biochemistry,

Jawaharlal Institute of Post-Graduate Medical Education and Research,
Pondicherry-605006

Request for reprints

Dr. B. M. S. Bedi

Department of Skin & V. D.

Goa Medical College, Panaji (Goa)

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xanthomatous tissue was also analysed for total & lipid cholesterol content. The cardiac condition was diagnosed as a mild symptomatic aortic stenosis. Patient's two brothers and one sister were biochemically normal. So also her parents and grandfather. Patient did not suffer from either myxoedema or nephrosis.

Result and discussion

Results of the various lipid estimations in the serum & xanthomatous tissue are given in Tables I & II. The index patient, had an increased total cholesterol of 800 mg% and a relative total lipid level of 2100 mg%. Her serum triglycerides and phospholipids were normal. Plasma was clear. Zonal electrophoresis showed a broad β -band with normal pre β band. The xanthomatous tissue showed 72% of cholesterol. The propositus alone showed

a mild aortic stenosis^{7,12}. She did not suffer from diabetes, myxoedema or nephrosis. Her grandfather aged 78 years had a cholesterol value of 300 mg%. Her parents, brothers and sister were biochemically normal. No member of three generations has developed a heart disease. All were apparently normal and healthy.

The investigations done in the family members clearly show that the patient did not inherit this hyperlipoproteinemia. It may represent an autoimmune disorder in which antibodies to blood lipoprotein are present in the circulation¹³. A normotriglyceridemia with a hypercholesterolemia and a hyperbeta-lipoproteinemia with tuberous and tendinous xanthomatosis is a typical finding in Type II a hyperlipoproteinemia.

TABLE I
Investigation on Blood

Generation Members	Total lipids * mg%	Serum cholesterol † mg%	T. G. mg% ‡	Lipoprotein spectrum
I Grand father	1200	300	64	Slight in B
Grand mother (deceased)	—	—	—	—
II Propositus father	1100	250	68	Normal
Mother	580	257	80	Normal
III Propositus	2100	800	66.5	B
Sister	564	250	82.0	Normal
Brother	490	244	85.0	Normal
Brother	700	230	80.0	Normal

* Estimated by Sulfo-phospho-vanillin method 3

† Serum cholesterol estimated by the method of Zak cited by Varley 4

‡ Estimated by the micromethod of Van Handel & Zilversmit 5

|| Stained as described by Sunderman 6

TABLE II
Investigations on Xanthomatous Tissue (Average of 5 investigations \pm SD)

Material	Lipids % dry tissue	Cholesterol % dry tissue
Xanthomatous tissue of the propositus	81 \pm 9.0	72 \pm 6

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REFERENCES

1. Frederickson DS, Levy, RI and Lees RS : Familial lipoproteinemias, *New Eng J Med*, 276 : 32, 1967.
2. Beaumont JL, Carlson, LA, Cooper, GR, et al : Classification of hyperlipidemias and hyperlipoproteinemias, *Circulation XLV* : 501, 1972.
3. Christopher SF, Ted WF, Ralph TD and Cecelia AQ. Improved determination of total serum lipids by the Sulfo-phosphovanillin reaction, *Clinical Chemistry* 18 : 673, 1973.
4. Varley H : *Practical Clinical Biochemistry*, 3rd Ed. The English Language Book Society and William Heinemann Medical Books Ltd., London, 1966, P. 236.
5. Van Handel E and Zilversmit DB : Micro-method for the direct determination of serum triglycerides, *J Lab Clin Med* 50 : 152, 1957.
6. Sunderman FW, and Sunderman, FW (Jr) : *Serum proteins and dysproteinemias*, 1st Ed. Pitman Medical Publishing Co., London & J.B. Lippincott Company, Philadelphia, 1964, p. 233.
7. Cook CD, Smith, HL, Giesen, CW et al : Xanthoma tuberosum, aortic stenosis, coronary sclerosis and angina pectoris : Report of a case in a boy of thirteen years of age, *Am J Dis Child*, 73 : 326, 1947.
8. Rigdown RH and Willeford G : Sudden death during childhood with xanthoma tuberosum : Review of literature and report of a case, *J.A.M.A.* 142 : 1268, 1950.
9. Barr DP, Rothbard S and Elder MA : Atherosclerosis and aortic stenosis in hypercholesterolemic xanthomatosis, *J.A.M.A.*, 156 : 943, 1954.
10. Stanley P Chartrand G and Davignon A : Acquired aortic stenosis in a twenty - five year old girl with xanthomatosis : successful surgical corrections, *New Eng J Med*, 273 : 1278, 1965.
11. Kumbhani AP, Daftary VG Yawalkar SJ et al : Xanthomatous aortic stenosis in familial hypercholesterolemia, *Ind Heart J* 19 : 170, 1965.
12. Rothbard S Hagstrom JWC and Smith JP : Aortic Stenosis and myocardial infarction in hypercholesterolemic xanthomatosis, *Am Heart J*, 73 : 687, 1967
13. Myant NB and Slack J : Type II hyperlipoproteinemia clinics in *Endocrinology and Metabolism* 2 : 87, 1973.

TRUE

Serious CNS, renal and cardiac complications can occur not infrequently when urticaria is part of serum sickness symptomatology. However, these system involvements are not rare in urticaria particularly those associated with angioedema. Cerebral edema, hemiparesis, convulsions and nerve palsies as part of urticaria-angioedema have been convincingly reported. ECG changes have been observed in many patients during an urticarial attack as well as transient albuminuria.

Reference : Warin RP and Champion RH ; *Urticaria*, W. B. Saunders Company Ltd., London, 1974, p. 81-83.