45th Annual International Congress of the
EGYPTIAN SOCIETY OF CARDIOLOGY
CardioEgypt 2018

Case Report
Antoine AbdelMassih (MD)

Case Summary and Investigations

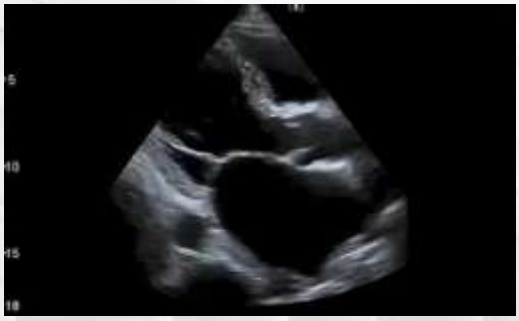
- A 5 y old male patient presented to the emergency department of Cairo University Children Hospital with severe respiratory distress poor peripheral perfusion. History was positive for familial hypercholesterolemia. General physical examination revealed multiple soft subcutaneous swellings over her hand, knuckles, Achilles tendon, wrist and ankle suggestive of xanthomas.

Previous labs withdrawn one week before the event

Patient's values		Patient's sister's values		
	mg/dl	mmol/l	mg/dl	mmol/l
Total cholesterol	657 mg/dl	16.8 mmol/l	630 mg/dl	16.15 mmol/l
LDL-C	572 mg/dl	14.6 mmol/l	554 mg/dl	14.2 mmol/l
HDL-C	61 mg/dl	1.6 mmol/l	50 mg/dl	1.3 mmol/l
Triglycerides	122 mg/dl	1.4 mmol/l	130 mg/dl	1.5 mm

He was on atorvastatin 80 mg Once daily

Echocardiography Revealed:

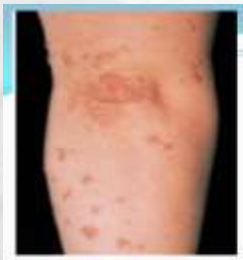


Fredrickson classification of hyperlipidemias

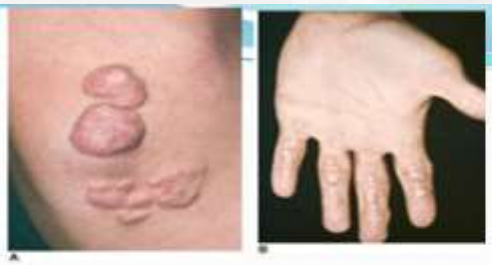
Hyperlipo-proteinemia		Synonyms	Defect	Main symptoms	Treatment
Type I	a	Buerger-Gruetz syndrome or familial hyperchylomicronemia	Decreased lipoprotein lipase (LPL)	Acute pancreatitis, lipemia retinalis, eruptive skin xanthomas, hepat osplenomegaly	Diet control
	b	Familial apoprotein CII deficiency	Altered ApoC2		
	c		LPL inhibitor in blood		
Type II	a	Familial hypercholesterolemia	LDL receptor deficiency	Xanthelasma, arcus senilis, tendon xanthomas	Bile acid sequestrants, statins, niacin
	b	Familial combined hyperlipidemia	Decreased LDL receptor and increased ApoB		Statins, niacin, fibrate
Type III		Familial dysbetalipoproteinemia	Defect in Apo E 2synthesis	Tuboeruptive xanthomas and palmar xanthomas	Fibrate, statins
Type IV		Familial hypertriglyceridemia	Increased VLDL production and decreased elimination	Can cause pancreatitis at high triglyceride levels	Fibrate, niacin, statins
Type V			Increased VLDL production and decreased LPL		Niacin, fibrate

Triglycerides vs. Cholesterol

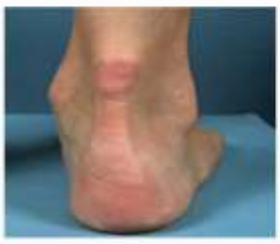
	++ Cholesterol	++ Triglycerides
Main carrying Lipoprotein	LDL	VLDL, Chylomicrons
Main complications	CAD	Pancreatitis
Types of Xanthomas	Tendon	Eruptive



Eruptive skin xanthomata characteristic of severe chylomicronemia.



Tuberoeruptive and tuberous xanthomata typical of familial dysbetalipoproteinemia. **A.** Knee **B.** Palm.



Tendon xanthomata typical of heterozygous familial hypercholesterolemia. Similar xanthomata occur in patients with familial defective apolipoprotein B-100, cerebrotendinous xanthomatosis, and sitosterolemia.

Xanthoma striatum palmare characteristic of familial dysbetalipoproteinemia.

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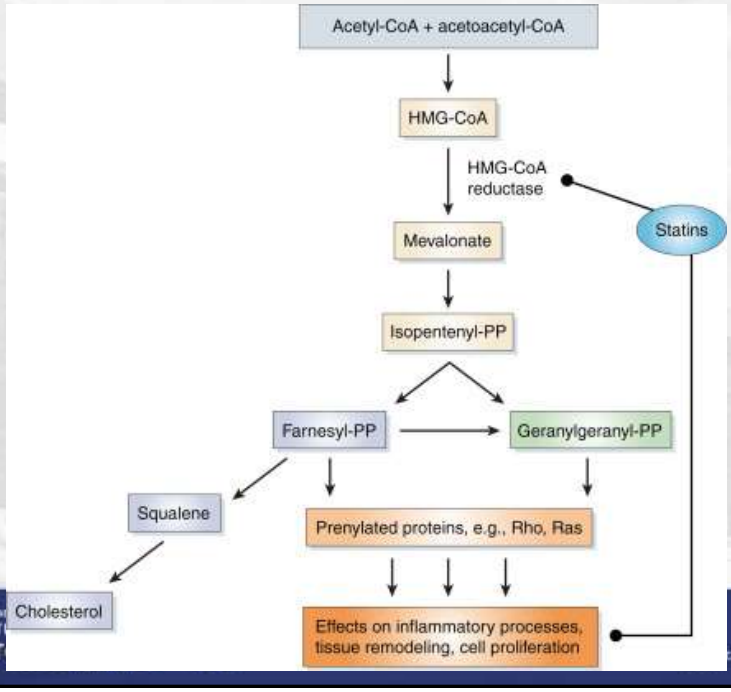

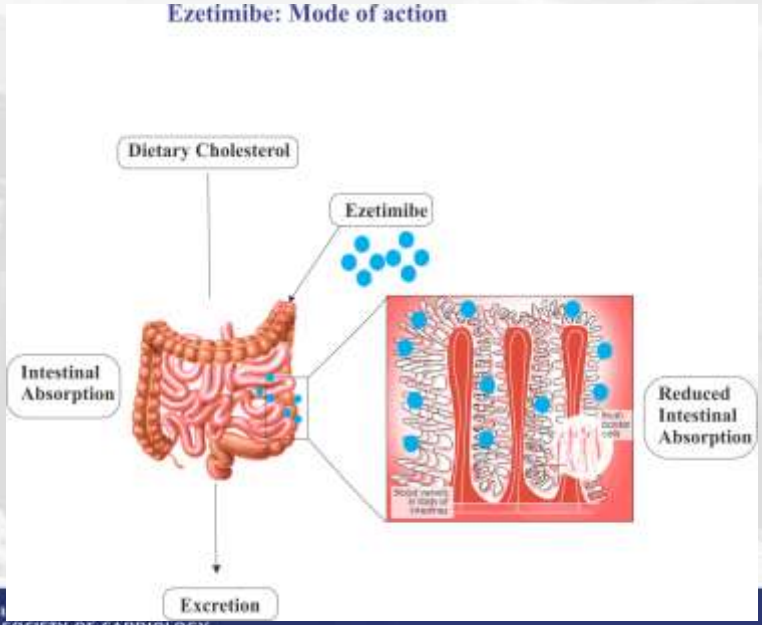

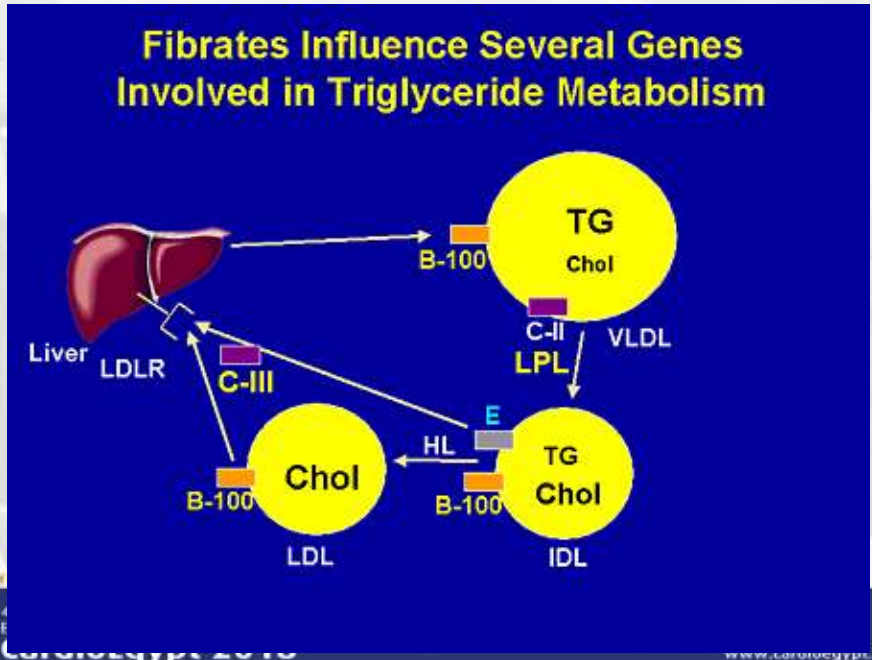

Any Pt with familial Hypercholesterolemia should be screened every 2 years with ECG and preferably stress ECG and if signs of CAD are apparent a MSCT should be carried out (Harada-Shiba et al. 2012)

Take home messages

- 1-Familial hypercholesterolemia is mainly manifested through tendon xanthomas and complicated by premature CAD
- 2-Drugs that are commonly used are Statins and Ezetimibe
- 3-Cardiovascular risk should be assessed every 2 years with ECG and ruled out or confirmed by MSCT

•Thank You

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