

CORRESPONDENCE

Familial Hypercholesterolemia (FH) Awareness amongst Physicians in Mumbai, India

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Sir,

Twin papers "Familial Hypercholesterolemia (FH) awareness amongst physicians in Mumbai, India" by Reddy & Ashavaid¹ and "Familial hypercholesterolemia with different types of xanthomas"² by Khot and Nagpure make an interesting reading in view of the current surging epidemic of coronary artery disease (CAD) particularly in young. There is a distinct possibility that some cases of homozygous familial hypercholesterolemia (HoFH) might be overlooked in young CAD by the busy physicians because of the rarity of this disease (one in one million).

Besides premature CAD it needs to be mentioned that some cases of HoFH might be presenting with valvular disease (most commonly stenosis / aortic regurgitation) in childhood/ adolescence.^{3,4} It needs no over emphasis that we might be missing some of these cases if not looked for keeping HoFH as cause for aortic valve disease in young, though they might be having clinical markers of HoFH like arcus juvenilis, xanthelasma, xanthoma and other pleomorphic cutaneous lesion of hypercholesterolemia.⁵ Thorough physical examination is extremely important in such cases.⁶ Keeping HoFH in mind is important because we now have drugs like PCSK9 inhibitors which are effective in lowering down markedly raised cholesterol. The only limitation of this drug is its exorbitant cost. Therefore it is extremely important on part of all physicians to suspect and screen for HoFH in young sibling and progeny of index case of HoFH. Further, one should think of possibility of HoFH in young people who present with acute coronary syndrome. The

diagnosis of HoFH should also be considered in consanguineous subjects presenting with aortic and/or mitral valvular disease in early childhood / adolescence.

References

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