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Mutation in the Gene Encoding Sterol Regulatory Element Binding Protein (SREBP-2) in Hyper-Cholesterolaemic Subjects

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KEYWORDS Mutations; missense mutation; SREBP-2 gene; hypercholesterolamia; Coronary artery disease

ABSTRACT The cholesterol homeostasis is regulated by the cleavage of membrane bound transcription factor sterol regulatory element binding proteins (SREBPs). We have analyzed 50 hypercholesterolaemic patients for genetic mutations in SREBP2 gene by direct sequencing of exon 5-10 of SREBP-2 gene that encodes the crucial functional domains. One missense mutation (V623M) in the regulatory domain was identified in the exon10 of SREBP-2 gene. However, no mutations were found in the regulatory domain in 200 controls samples. This is first report from India suggesting that the missense mutation (V623M) in human SREBP-2 gene may be true pathogenic mutations of hypercholesterolamic state.