Genetic Markers

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Role of genetic and environmental factors in predisposition of many common diseases has been widely recognized. The genetic component predisposes and an environmental component help in the manifestation of the disease. Genetic studies helps in detecting predisposed persons so that they may be advised against exposure to environmental risk factors[1]. An identification of individuals who may be predisposed to such diseases could provide valuable information which may be of immense help not only in the management of the disease in the affected person but also in planning the future strategies for the family.

Duodenal ulcer is a common gastrointestinal problem believed to exist in families. Many pedigrees have been reported in which several members had an ulcer. The disease was found to oceurtwo to two and a half times more frequently among siblings of ulcer patients than among ulcer free subjects from a comparable population[2]. Duodenal ulcer was 2.6 times more frequent in sibs of patients than in others[3]. Genetic factors have long been suspected because of an increased frequency of the disease among the first degree relatives of the patients[4], greater concordance between monozygotic and dizygotic twins[5] and the associationofABO blood groups with the disorder[4]. Secretor and non-secretor status[4], ABO blood groups[6], HLA typing[7], serum pepsinogen[8] and serum alpha 1 antitrypsin[9] serves as genetic markers of the ulcer diathesis. The magnitude of association between blood group 0 and non-secretor status with duodenal ulcer appears weak[6], while raised serum pepsinogen and low alpha 1 antitrypsin have been found to identify those at an increased risk for the development of the disease[10]. Due to scarcity of data which could accurately indicate whether there exist any associationbetween the various genetic markers and duodenal ulcer, it appears appropriate to establish a possible co-relation between them, more so because duodenal ulcer is common in the local population and the etiology is yet to be determined either to be of genetic or non-genetic in origin.

In a recently concluded study, it was observed that only 28% of the patients had raised serum pepsinogen, while serum alpha 1 antitrypsin was low in 35% of the patients. Other findings include dominant blood group 0, lower mean age, an early onset of the disease, an increased frequency of gastrointestinal bleeding and ulcer perforation thereby confirming the strong association of duodenal ulcer with all the markers. It appears that the genetic etiology of the disease existed in just 28% of the patients while the rest comprising a large majority (72%) had ulcer as a result of neuro endocrinological or environmental factors which are also known to cause the disease[11].

A deficiency of pmtease inhibitor alpha 1 antitrypsin might be linked to causation or persistence of duodenal ulcer. This postulate is supported by the findings that patients identified with partial deficiency of protease inhibitor had relapse and duodenitis persisted inspite of lonpterm follow-up and treatment with H2 receptor antagonist[12].

Duodenal ulcer patients manifesting hyperpepsinogenemia or low serum alpha 1 antitrypsin reqUire further investigation in the family to identify siblings and progeny carrying the trait and thus, at higher risk, so that they could be shielded fmm aggravating environmental and other factors:

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