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Background: Colon cancer is cancer of the large intestine (colon), the lower part of digestive system. Adiponectin induce apoptosis and prevent growth and proliferation cell. Many studies show an association between low level of adiponectin hormone and insulin resistant, obesity that is because of adiponectin polymorphism and increased risk of colorectal cancer. The goal of this study is to evaluate incidence polymorphism of adiponectin gene rs 2241766 in Iranian and to check the role of this polymorphism in increased risk of colorectal cancer.

Methods: Genotyping of the insulin gene were determined in 107 colorectal cancer patients and 107 controls using polymerase chain reaction and restriction fragment length polymorphism genotyping assays (PCR-RFLP). The statistical analysis was done with SPSS 16.

Results: Results show that polymorphism adiponectin rs 2241766 is not a predisposing factor to increased risk to CRC (Pvalue = 0.29). The genotype GT compared with TT, is not associated with increased risk of CRC (OR = 1.02, CI = 0.52–1.97 Pvalue = 0.95) but GG versus TT (OR = 0.015 CI = 0.29–0.78 Pvalue = 0.025) is associated with decreased risk of CRC. Incidence of mutant allele has no significant difference between patients and controls (OR = 7.34 95% CI = 0.45–1.19 Pvalue = 0.21).

Conclusion: These findings suggest that mutant allele of adiponectin polymorphism rs 2241766, is associated with decreased risk of CRC.

Keywords: Colorectal cancer, Adiponectin gene, PCR-RFLP

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E.poster – [A-10-1090-1]

A novel mutation in the alpha 1 globin gene in an Iranian α -thalassemia carrier

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Introduction: Alpha-thalassemia is an inherited disorder that affects the production of normal hemoglobin. More than 30 different point mutations and small deletions and insertions have been reported in alpha globin gene. The most common causes of α -thalassemia are deletions, and point mutation are less common, but they may occur at high frequencies in certain areas.

Materials and methods: After obtaining informed consent, blood samples (10 mL) were collected in tubes containing EDTA and DNA was extracted from peripheral blood leukocytes by salting out method. Multiplex gap-PCR and direct sequencing techniques were used to analyze alpha globin genes.

Results: The proband (31 years old) was referred with low hematological indices and normal HbA2 and HbF. Multiplex gap-PCR

showed the presence of The sequencing of $-\alpha 3.7$ deletion in heterozygous form. The sequencing of α -globin revealed c.349G>A (p.Glu 116 Lys). The mutation has not been reported in globin gene server previously.

Conclusion: This is the first CD116 (GAG>AAG) mutation described on the alpha1 globin gene. The severe reduction of hematological indices lead us to suppose this proband has a mutation in compound heterozygous with $-\alpha 3.7$ deletion.

Keywords: Alpha-thalassemia, CD116 mutation, Compound heterozygous

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E.poster – [A-10-1098-1]

Detection of the most common β globin polymorphisms by single-tube multiplex ARMS /RFLP

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Introduction: Multiplex polymerase chain reaction (PCR) is a variant of PCR in which two or more loci are simultaneously amplified in the same reaction. This method has been successfully applied in many areas of DNA testing including analyses of deletions, mutations and polymorphisms. We have developed a simple and rapid multiplex ARM-PCR technique for analysis β globin gene cluster polymorphisms instead of PCR and digestion.

Materials and methods: DNA samples were obtained from β -thalassemia carriers referred to our clinic for prenatal diagnosis (PND). Genomic DNA was extracted by using salting out method. In this study we have developed single-tube multiplex ARMS-PCR for for three sites RFLPS (HincII/3' Ψ β , Avall/ β , HinfI/ β) that could be easily identified by size.

Results: Our results show that normal and mutant PCR products are readily scored by the presence or absence of the correctly sized band following electrophoresis on gels. Comparison of ARMS-PCR and PCR with digestion showed 100% concordance.

Conclusion: The simplicity and university of this multi ethnic multiplex PCR assay significantly reduce the cost and complexity of PCR RFLP techniques. Multiplex polymorphic markers determine whether family members have inherited normal or mutation-harboring chromosome in one test.

Keywords: Multiplex ARMS-RFLP, β gene cluster

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Poster – [A-10-1111-1]

Association between Heme Oxygenase-1 gene promoter polymorphisms and metabolic syndrome in Iranians

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