Materials and methods: The CC, CT and TT genotypes frequency of C677T polymorphism of MTHFR gene were 77, 22 and 1% in preeclamptic women and 73, 19.5 and 7.5% in controls and significantly were not different(P = 0.06). But the frequency of TT genotype was significantly higher in controls. OR = 8.5 (95% CI 1.1–71, P = .018). There was no significant difference detected in T allele distribution between pre-eclamptic women(12%) and controls(17%) too.

Conclusion: Our results showed that there is not any correlation between the C677T polymorphism and preeclampsia, but the TT genotype of C677T polymorphism seems to be a protective factor for preeclampsia.

Keywords: Preeclampsia, Methylenetetrahydrofolate reductase, C677T, Polymorphism

doi:10.1016/j.clinbiochem.2011.08.735

Poster – [A-10-748-1]

Co-inheritance of αααanti 3.7 triplication with β-thalassemia trait in an Iranian family

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Introduction: The pathophysiology and clinical severity of β-thalassemias are associated with the degree of α/nonα-chain imbalance. A triplicated α-globin gene locus can exacerbate the effects of α-chain excess caused by a defective β-globin gene, although this coinheritance in different individuals results in variable phenotypes. In the present study we report the molecular analysis of an Iranian subject with a thalassemia intermedia phenotype, heterozygous for β-thalassemia.

Methods: DNA extraction from peripheral blood leukocytes was performed by salting out method. Mutation analysis of the β-globin gene was detected by ARMS-PCR and multiplex-PCR was used to detect alpha triplication (αααanti3.7).

Results: The mother of index case was a carrier of β-thalassemia with IVSII-I mutation in heterozygous form and her husband was hematologically normal with no mutation in β-globin gene. The propositus, a 2.5 years old child presented a transfusion dependent thalassemia intermedia phenotype. Multiplex PCR detected the presence of extra α-globin gene in the patient and her mother but her father was normal.

Conclusion: The clinical and hematological pictures of β-thalassemia heterozygotes with a triplicated α-globin gene arrangement is variable, ranging from an asymptomatic presentation to a mild to moderate thalassemia intermedia phenotype. This finding has important implications for genetic counseling and prenatal diagnosis programs. This family may be at risk for another child with severe thalassemia intermedia. The genetic and phenotypic characteristics of the patients described here indicate the need to consider the possibility of a triplicated α-gene allele in patients with heterozygosis for β-thalassemia who show an unexpected severe phenotype.

Keywords: Alpha triplication, Thalassemia intermedia, β-thalassemia trait

doi:10.1016/j.clinbiochem.2011.08.736

Poster – [A-10-757-1]

Polymorphism in exon 11 of Breast Cancer Gene (BRCA1) in Betta Kuruba tribal population of India

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Introduction: Breast cancer is a cancer formed in tissues of the breast. Worldwide, it is the second leading cause of cancer deaths in women. Specific mutations in genes like HER2, BRCA1, BRCA2 and p53 have been linked to it. Mutations in BRCA1 and BRCA2 are associated with a significant increase in the risk of breast cancer. BRCA1 officially refers "breast cancer 1, early onset." It belongs to a class of "tumour suppressor genes". The prevalence of breast cancer as well as the BRCA1 and BRCA2 mutation varies with races and ethnicity. Betta Kuruba is a scheduled tribe of India who lives in the high reaches of Western Ghats of South India.

Methods and methods: 570 nucleotide bases (880th–1449th base) of exon 11 of genomic DNA from blood sample of 31 Betta Kuruba control subjects and 9 breast cancer diagnosed cases were sequenced. Agarose gel electrophoresis, PCR and cycle sequencing were carried out. Thermal cycler, Genetic Analyser 3730, Seqscape 2.5, GenBank, Ensembl, Netprimer and Primer 3 were used.

Results: A single nucleotide polymorphism (SNP) was identified at position 1412. In cases, its prevalence was 77.78% for heterozygote and 11.11% for mutated homozygote. In control, the frequency for heterozygote was 29.03% and for mutated homozygote is 25.81%. Frequency for mutant allele T in cases and control is 0.5 and 0.42, respectively.

Conclusion: A SNP was identified in position 1412 of exon 11 of BRCA1. The mutation or heterozygosity was significantly observed. The high frequency of mutant allele and the significant frequency of heterozygosity in the control group indicated certain underlying environmental risk factors that may trigger the process of cancer development.

Keywords: Breast Cancer, BRCA1, Betta Kuruba, Exon 11, Single nucleotide polymorphism

doi:10.1016/j.clinbiochem.2011.08.737

Poster – [A-10-766-1]

Determination of insulin-like growth factor-I effect on apoptosis in CHO cells in biopharmaceutical production

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Introduction: Optimizing of mammalian cell culture technology is essential for economical production of biopharmaceuticals such as monoclonal antibodies and recombinant biopharmaceutical proteins. A major problem faced in bioreactor culture is cell death which decreases overall biopharmaceutical yield. One of the major modes of cell death is apoptosis that is a form of programmed cell death (PCD) which is major problem in culture of animal cell line with industrial significance such as Chinese Hamster Ovary (CHO) cells. The ability to