Association between TP53 gene ARG72PRO polymorphism and gastric cancer in Ardabil province, Iran.

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Introduction: Gastric cancer (GC) as the 3rd most common malignancy in Iran, accounts for ~50% of all GI cancers who cause 55% of all cancer-related deaths in Iran. The rates of GC reported from Ardabil province, Iran, are among the highest in the world. Upper gastrointestinal cancer accounts for more than 50% of all cancer deaths in this area. Codon 72 polymorphism of the tumor suppressor gene TP53 has been associated with a higher risk in the development of several types of cancer. The polymorphism results in a variant protein with either an arginine (CGC) or a proline residue (CCC). We aimed analyze the association of the TP53 codon 72 polymorphism with the risk of developing gastric cancer in a high-risk population around the world.
Materials and Methods: We enrolled 87 patients with mean age 65.9 (range: 37-87; std.=11.1) affected with primary gastric cancer (GC) and same age- and sex-matched healthy control participants. The analysis has been done by PCR-RFLP on DNA extractions from peripheral blood.
Results: In case group the genotype was 16.1%, 42.5%, and 41.4% for Arg/Arg, Arg/Pro, and Pro/Pro, respectively. And for controls those were 18.5%, 40.2%, and 41.4%. In comparing case and control group, no significant correlation was found (p=0.9). Also, there was any significant correlation between codon 72 status and pathologic data.
Conclusion: Because of the high frequency of GC in our province, the investigations about the role of genetic susceptibilities for GC are very important. In spite of finding no relationship between P53 polymorphisms, studying other genetic variations is recommended.

Study of extracellular DNA associated with erythrocytes in healthy peoples and with chronic pyelonephritis.

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Introduction: At present, the origin of extra cellular DNA in plasma (serum) in human blood by the release of living cells do not arise any doubts.Therefore, we currently focus on the composition of these nucleic acids and their correlation with different diseases or phases of the physiological state of man. Thus the use of analysis of extra cellular DNA may have to address all four objectives of Laboratory Medicine - screening, diagnosis, monitoring and forecasting. For screening and as a noninvasive diagnostic method it is possible to use definition of level of extra cellular DNA and its predecessors-ASF (acid-soluble fraction) in the blood of patients with renal pathology.In our work we have distinguished two objectives: The study of content of extra cellular DNA and ASF associated with erythrocytes in the blood of healthy individuals and patients with common renal pathologies - chronic pyelonephritis (CP)Previously, it was found that an increased level of nucleic acids in blood was observed in a number of cancers, radiation damage, autoimmune diseases, aging and pregnancy.
Material and Method: 50 Patients (age 25- 55 years) with clinically verified CP and 50 healthy peoples were included in the study. Epidemiological data (as Personal, family, smoking and drinking history, nutritional information) and clinical pathological data were collected from the medical records and face to face survey questionnaire Their blood sample was collected for study of extra cellular DNA & ASF (Acid Soluble Fraction) by using Marusheva L.E equilibrium test and spectrophotometery.
Result: Our analysis shows an increase of ASF compared with control by 2.02 times which represents a significant decay of extra cellular DNA on its fragmentation, since endonuclease activity in blood significantly increased in CP.The number of extra cellular DNA associated with erythrocytes is decreases by 2.18 times compared with control.This result confirms our data on the accelerated degradation of extra cellular DNA in blood in CP.
Conclusion: Extra cellular DNA & ASF associated with erythrocytes in blood of healthy people and in patients with Chronic Pyelonephritis was differentiated by increased level of ASF by 2.02 times & decreased level of extra cellular DNA content by 2.18 times..

Apert Syndrome

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Introduction: Apert syndrome is a genetic disease, caused by a mutation transmitted autosomal dominant gene FGFR2 (location: 10q25-q26). Major clinical manifestations include oxicefalie, brachycephalism, hipertelorism, protruding cardiovascular anomalies and genito-urinary and musculoskeletal sindactilie. Objective: This paper aims at presenting a patient with Apert Syndrome, focusing on multidisciplinary aspects of disease management (genetic screening, pediatric, orthopedic, dental and neurological), and problems occurring in providing genetic counseling. The patient is the 3rd child of a couple young mother 30 years, father 36 years, neconsangvin. Pregnancy developed normally birth occurred at term, naturally, cranial presentation and postnatal development is normal, appropriate age of 1 month.
Results: Based on clinical picture and paraclinical Apert Syndrome is suspected. Certainly positive diagnosis: molecular diagnosis, identification of gene mutations in FGFR2.
In conclusion we mark the importance of clinical examination and radiological diagnosis Syndrome Apert, patients requiring a multidisciplinary assessment (neurosurgeon, radiologist, oral-maxillo-facial surgery, orthopedic surgeon, geneticist, a psychiatrist) for preoperative evaluation and identification of other malformations associated / complications (hydrocephalus).