Male Individualization Based on Y-Chromosomal Short Tandem Repeats: A Comparative Information Theoretical Analysis of 16 Y-STR Loci in Central Anatolia and Iraqi Populations

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KEYWORDS Central Anatolia and Iraqi Populations. Entropy. Pointwise Mutual Information. Population Genetics. Y-STR Polymorphisms.

ABSTRACT The aim of this study is to investigate the discrimination capacity of 16 Y-Chromosomal Short Tandem Repeat markers (Y-STRs) based on their joint entropy for the purpose of male individualization on samples taken from Central Anatolia and Iraqi Populations. The Y-chromosome polymorphism of sixteen STR loci (DYS19, DYS385a/b, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438, DYS439, DYS448, DYS456, DYS458, DYS635, Y-GATA H4) were studied. Genomic DNA was extracted from buccal swabs using the QIAamp Mini kit and was co-amplified by using Applied Biosystems AmpF*l*STR® Yfiler™ PCR Amplification Kit. The Iraqi data set was readily available in the literature which is based on blood samples randomly collected from 100 healthy unrelated males living in middle or south of Iraq. The researchers observed 106 unique haplotypes in Central Anatolia data set. The genetic diversity values across the 16 Y-STR loci ranged from 0.564 (DYS391) to 0.876 (DYS385a/b). The complete male individualization with only 16 Y-STR markers in a genetically diverse local population is possible. In this study, haplotype diversity was 1.0 and discrimination capacity was 100 percent. The high discrimination capacity of the 16 Y-STR markers makes them valuable for male individualization for forensic purposes in Central Anatolia Region of Turkey. The researchers also show that, the *pointwise mutual information* and the *joint entropy* between allele pairs measure the discrimination power of markers more accurately than individual genetic diversity values and provide a better insight into the interaction between the genetic profile of the population and the given Y-STR marker set.

Perspectives Revisited - The Buccal Cytome Assay in Mobile Phone Users

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KEYWORDS DNA Damage. Cytokinetic Defects. Cell Death. Buccal Epithelial Cells

ABSTRACT Buccal cell preparations previously scored for micronuclei were re-investigated for genomic instability and other biomarkers to assess DNA damage, cell-proliferation and cell-death in healthy mobile phone users (n=25; 30.96±2.09y) using mobile phones for 3-5y and the non-mobile phones users (n=25; 32.28±2.01y) according to the buccal micronucleus cytome (BMCyt) assay which was then not available. The frequency of micronuclei (13.66x), nuclear buds (2.57x), basal (1.34x), karyorrhectic (1.26x), karyolytic (2.44x), pyknotic (1.77x) and condensed chromatin (2.08x) cells were highly significantly (p=0.000) increased in mobile phone users whereas the binucleated cells (4.03x) and repair index (8.36x) showed significant decrease (p=0.000). DNA damage and nuclear anomalies scored in BMCyt assay are indicative of genetic damage that has not been repaired and this may predispose the mobile phone users to malignancy and cytotoxicity ramifications. Therefore, despite the benefits of communication technology, measures need to be taken so that better connectivity is not at expense of health.

Int J Hum Genet, 15(4): 183-189 (2015)

Angiotensin-Converting Enzyme Gene Insertion/Deletion Polymorphism in Patients with Pulmonary Thromboembolism

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KEYWORDS Pulmonary Embolism. Angiotensin-Converting Enzyme. Renin-Angiotensin System. Gene Polymorphisms. Venous Thromboembolism

ABSTRACT The aim of the present study is to investigate the relationship between angiotensin-converting enzyme (ACE) gene polymorphism and pulmonary embolism by comparing the frequency of ACE gene polymorphism between cases diagnosed with pulmonary embolism with that of the control group. The study included 73 patients and 73 healthy subjects as the control group. Isolated DNAs were genotyped using the polymerase chain reaction (PCR) method for the identification of the ACE insertion/deletion (I/D) polymorphism. The genotypes were determined according to the bands observed in the agarose gel electrophoresis. The frequency of ID genotype was 39.7 percent, the frequency of insertion/insertion (II) genotype was 17.8 percent, and the frequency of the deletion/deletion (DD) genotype was 42.5 percent in the patient group. In the control group, the frequency of the II genotype was 21.9 percent, the frequency of the ID genotype was 38.4 percent, and the frequency of the DD genotype was 39.7 percent. There were no statistically significant differences between the patient group and the control group in terms of the frequencies of II, ID, and DD genotypes ($p > 0.05$). The findings of the present study showed no association between ACE gene polymorphism and the risk of developing the pulmonary embolism. Due to the limited number of patients however, these results must be confirmed by further studies incorporating larger [illegible] of patients

Two Independent Genetic Origins of β^+-Thalassemia Due to -31 A to G Mutation in Thai and Japanese Populations

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KEYWORDS Allele Specific PCR Assay. Beta-Globin Gene. β^+- Thalassemia. β^{-31A-G} Mutation. Genetic Origin. Haplotype Analysis

ABSTRACT Haplotype associated with the -31 (A-G) β^+ -thalassemia gene in seven Thai individuals were examined and compared with that described originally in Japanese. Seven polymorphic restriction sites within β-globin gene cluster were determined using allele specific polymerase chain reaction (ASPCR) methods newly developed for rapid β-globin haplotyping. A concordant result of DNA polymorphisms examined using ASPCR and conventional PCR-restriction fragment length polymorphism (PCR-RFLP) method was observed. It was found that all these seven Thai β^+-thalassemia alleles were associated with the β-globin haplotype (+ - - - - - +), which is different from that described for a Japanese subject (- + + - + + -). This indicates two independent origins. As compared to the PCR-RFLP method, β-globin haplotyping using ASPCR developed is easier, rapid, less time-consuming and requires no restriction digestion. The methods should also prove useful in population genetic study and linkage analysis of β-hemoglobinopathy.

Congenital Heart Defects and Chromosomal Abnormality

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KEYWORDS Congenital Defects in Heart. Abnormalities in Chromosomes. Genetic Counseling

ABSTRACT Chromosomal abnormality is one of the causal factors in the formation of the congenital heart defects. 65 patients (33 male and 32 female) with heart defects were referred for karyotyping and counseling. Chromosomal abnormalities were detected in 27 (41.5%) and 38 had a normal karyotype. Numerical abnormality was found in 21 (77.8%) and structural in 6 (22.2%), numerical was detected in 14 females and 7 males, and structural in 4 female and 2 male patients. Numerical abnormality was one with 47,XX+13; 2 with 45,X and 18 with 47,XX+21 (11) or 47,XY+21(7). Structural abnormality was derivative 9 in 2, deletion 11q, derivative 14, Robertsonian translocation between 14 and 21 and ring 18 mosaicism in one each. Parental origin of the structural abnormality revealed that two were maternal and one was paternal. In the present study, association could be detected between chromosome 21 and the female probands with chromosomal abnormality and heart defects.