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ABSTRACTS

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United Kingdom who won the Nobel prize for his investigation on cloning is named by chance. Then, the development and investigation of Medical Genetics, in Universities and Private Centers of the country will be mentioned. Also the evolution up to date, the genetic counseling, the cytogenetics, the immunogenetics, the molecular genetics and others throughout the continent, and the relations that we keep with centers of more developed countries.

The Biothics imposed by our laws, principles and usages, with their advantages and inconveniences will be discussed. In the background, basis of preconceptional genetics will be explained considering our Genetics Foundation in Cordoba, Argentina as the pioneers in this scientific programme based on the preventive care that the couple should undergo in order to offer their gametes in optimum conditions to create children with the minimum risk of congenital mistakes, until another possibility of identification and rectification of pathological genes in the germinative cells is discovered. Comments and statistics will be described in details.

Red blood cell and genetic studies

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Since the discovery by Landsteiner of the ABO blood groups early in the century, the human red blood cells (RBC) have been extensively used in areas of genetic research. Studies of haemoglobins in sickle cell disease and thalassaemia and of RBC enzymes in general, and glucose-6-phosphate dehydrogenase (G6PD) in particular, are just a few examples of how investigations on RBC have been useful in our understanding of genetic and pathophysiological principles in human medicine.

Comparative studies on mammalian RBC have provided valuable information in cell metabolism and oxygen affinity of haemoglobin and therefore, not unexpectedly, RBC of some animals have been used as models for human diseases. For example, sheep RBC exhibit at least three principal genetic variants that affect the membrane transport characteristics of potassium, amino acids and nucleosides; RBC of some dogs have very high concentrations of reduced glutathione. It is against this background that we have begun studies on the RBC metabolism in some native mammals in Australia-the marsupials. Major findings have been: 1. Almost seven fold variation in G6PD activity and two fold variation in glucose-phosphate isomerase (GPI) activity in the RBC of two species of wombats. 2. Higher sensitivity of wombat RBC (With low levels of G6PD and GPI) to oxidant stress induced by certain chemicals. 3. A significant nagative correlation between the activity of catalase and methaemoglobin formaiton by H_2O_2 and also between the catalase activity and the degree of haemolysis by H_2O_2 in marsupial RBC.

Frequencies of different glucose-6-phosphate dehydrogenase mutations in southern Chinese populations

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Glucose 6-phosphate dehydrogenease (G6PD) deficiency, an X-linked genetic disease, is the most common enzymopathy causing hemolytic disorder in the world. It is associated with neonatal jaundice and may lead to mental retardation in severe cases. The incidence of G6PD deficiency varies in different ethnic groups. In

southern China, deficient activity of G6PD is common with the average prevalence around 2-3%. To date, more than 50 mutations at G6PD gene been identified, most of them are point mutations, which indicate genetic heterogeneity of G6PD deficiency.

Seven missense mutations (1388, 1376, 1024, 493, 487, 392 and 95) were identified from G6PD deficiency patients in Taiwan, which account for around 88% of patients. To determine the distributions and frequencies of these mutations in southern Chinese, we used a PCR-based restriction analysis to detect the seven known G6PD mutations among G6PD deficient patients from Taiwan, Hong Kong, Singapore, Guangdong and Guangxi, Dried blood spot collected on filter were used for PCR experiments.

Five mutations (1388, 1376, 1024, 392 and 95) account for 73% to 87% of G6PD mutant alleles in the Chinese from above areas. Two mutations (493 and 487) are rare in southern Chinese except in Taiwan. Mutation 493 is found exclusively in Chinese from Taiwan, which may be due to founder effect of fresh mutation, or originate from one of the aboriginal native of Taiwan, who merged into Chinese population in Taiwan by intermarriage previously. Our study indicates multiple origins of G6PD mutations that spread in southern Chinese populations. Moreover, this easy and non-radioactive assay method provides a simple way to confirm positive G6PD deficient cases detected by neonatal screening test using the same dried blood spot. This method is also proven suitable for large scale epidemiological study of G6PD mutations in different populations.

Molecular genetic approaches for control of vector transmitted human diseases

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Mosquito transmitted human diseases such as malaria, filariasis, yellow fever, dengue and dengue-hemorrhagic fevers, and several viral encephalitides pose some of the most serious public health problems in the world today. Malaria alone causes clinical illness, often severe, in 300 - 500 million people and between 1.4 - 2.6 million deaths each year. The epidemiology and distribution of many of these diseases has dramatically increased in recent years as a result of (i) development of widespread resistance of vector populations to traditional chemical control and of pathogens to commonly used drugs, (ii) large-scale movement of human populations from rural to urban and/or to previously uninhabited areas, often leading to the emergence of slums lacking even rudimentary public health facilities and with high human densities and, (iii) inadvertent introduction of vector and parasite populations from endemic to non-endemic areas, sometimes across continents, due to increased human mobility and commerce. Consequently, many of these diseases have become virtually intractable.

A variety of chemical, biological and genetic methods have been used from time to time to control vector populations, often with limited success. Nevertheless, past releases for genetic manipulation of field populations have established the validity of the scientific principle of genetic control. Recent work on the development of molecular genetic strategies, based on the recombinant DNA technology and spread of appropriate genes for inability to transmit a disease through suitable transposable elements and/or the rickettsial endosymbiont, *Wolbachia*, to control and/or manage these diseases, offers exciting possibilities and will be discussed.

In order to achieve effective population control through molecular genetic approaches it is critical that (i) the genomes of mosquito species, particularly those that are known vectors, are well characterized, (ii) saturated, molecular genetic and physical maps are constructed, (iii) the refractory genes for vector incompetence are effectively mapped and cloned, and (iv) vector transformation with molecular constructs