# A Computational System for human chromosome analysis

## Summary of the Project (PHASE 1)

Technological developments have opened up new possibilities of exploring diverse segments in the scientific areas of DNA namely, cytogenetics. Human cytogenics emerged in the middle of this century. It took off in 1959 with the discovery of the association between Down syndrome and chromosome 21. Nowadays, there are various techniques to study human chromosomes that focus on morphology which is the main technique in G-banding, or molecular cytogenetics known by FISH (Fluorescent In-situ hybridization).

Recently, we have been testimony to an increased demand in genetic sequencing by the considerable investment made throughout the world in various projects. For example, the project of Human Genoma “Plant Genome Initiative”, among others. FAPESP has confirmed it´s support of such project developments by way of financing research projects in various segments such as: Human Genoma of Cancer, Genoma Cana, Genoma Xanthomonas, among others.

In the same line of research and development, but on a smaller scale of complexity, the world markets have offered some computational systems dedicated to the analysis of Chromosomes. These systems present functions like generating chromosome representing in pairs called “pairing”; According to international standards, known as “karyotype” and the analysis of molecular cytogenetics of some chromosomes that derived from the FISH studies. Because of FISH it is possible to provide a cytogenetic diagnosis of embryos and fetuses at elevated risks of Chromosomal abnormalities. If the FISH study is executed in the initial stage of cell division, some diseases can be detected as can the sex of the future individual. This study uses chromosomal waves luminescent that fit in parts of the DNA chain from a chromosome emitting light in certain wave lengths. Optic filters are then used to select the light with the wave length that one wishes to analyze. The capturing and analysis of the standard light is the essence of the FISH study.

Despite the increasing demand, there are not many systems offered that automate or assist in the collecting of data for the geneticists in the generation of Karyotypes and the FISH

study. Our objective is to develop a computerized system utilizing vision techniques through the computer. This would assist geneticists in the analysis of how human chromosomes form, which would then lead to visualizing chromosomal pairing. Finally, this system would also assist in the collection and analysis of data for the FISH studies.

## Summary of the Project (Phase II)

The development of technology has enabled the exploration of a diverse number of segments in the area of science that study the DNA chains, namely, cytogenetics. Human cytogenetics has emerged in the middle of this century, around 1959 when the association between Down syndrome and Trisomy 21. Currently there are various techniques for studying human chromosomes, morphologically is the main technique for G-banding, as well as by methods of molecular cytogenetics known by FISH (Fluoescent in situ hybridization). It is through the FISH study that we can obtain a cytogenetic diagnosis of embryos and fetuses at high risk of chromosomal aberrations. If the FISH study is conducted at the initial stage of cell division, some illnesses can be detected as well as the sex of the future individual being developed. This study uses the “chromosomal waves” luminescents that attach themselves to part of the DNA chain of some of the chromosomes letting off light in different lengths of the wave. Then ethic filters are then utilized to select the light with wave lengths that are chosen to be analyzed. The capturing and the analysis of the standard luminescence is the essential part of the FISH study.

Despite the increasing demand, the number of automated systems which assist in the endeavors of geneticists in the generation of Karyotyping and the FISH study is small. The objective of the present project is to develop a computational system with adequate pricing in the Brazilian market (and Mercosul) utilizing techniques of visualization by computers (Computational vision as cited in Hom (1986). This system would be capable of assisting geneticists in analysis of human chromosomal formation in order to assist in the pairing process as well as assist in the FISH Study. With the assistance of this system and the remote transfer of digital images acquired by specialists in the citogenetics area, and the use of the internet, we intend to increase the homogeneous results of chromosomal analysis.

The objective of the system can by summarized by the following:

- Develop a software program to capture digital images of chromosome samples

- Develop a software program for the Karyotype generation

- Develop a software program for the execution of the FISH study

- Exchange information remotely via internet for the emission of second opinions