A Software Tool for Mapping Human Genome by
Chromosome-Specific Two-Dimensional
Electrophoresis Method

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Many human diseases, whether or not they are hereditary, are accompanied by genomic aberrations such as deletions, amplifications, translocations, or loss of heterozygosity. These changes have been identified and analysed by directly observing chromosomes as well as test by means of hybridization or polymerase chain reaction (PCR) using markers or probes corresponding to each particular locus of the human genome. A method based upon gel-electrophoresis, called restriction landmark genomic scanning (RLGS), has recently been developed. Genetic alterations can be detected and analyzed by surveying the entire genomic DNA after separating DNA fragments in a single two-dimensional slab gel. The resolved DNA fragments are highly specific and migrate to a distinctive position during electrophoresis, where they can be visualized as spots on X-ray film. Typically, about three thousand DNA fragments (for simplicity, referred to as spots hereafter) can be identified with good reproducibility. Because of this high specificity and resolution, this procedure has been used to analyze and detect changes in the genomic DNA. However, although the RLGS profiles represent a set of landmarks as positions and intensities on an autoradiogram, there is no direct link between these spots and genetic

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information such as their chromosomal locations or known genetic markers. For practical purposes, each spot must be correlated with the specific locus or gene in a particular chromosome, to directly assign the observed changes in a specific area of the genomic DNA. At present, a data set from chromosome-specific two-dimensional gel-electrophoresis compiled into the total genomic DNA profile, which we call the Chromosome Assigned-RLGS (CA-RLGS) profile, contains 2,676 mono-chromosomal, 82 di-chromosomal, 101 multi-chromosomal, and 12 highly redundant spots derived from a ribosome-DNA cluster.

Since the goal of this study is to establish a database including such information linked with individual spots in the profile, extensive use of computerized tools is essential for collecting and analyzing data, and finally converting them into a database. In this presentation, we will demonstrate a preliminary version software tools that will be used for detecting spots on the X-ray film.