

Human Y Chromosome as a Genetic Marker

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Abstract: The aim of this article was to determine genetic diversity (using a universal marker of Y-chromosome) within individual populations and identify the ancestral haplotype, participated in the ethnogenesis of the peoples.

Key words: Y-chromosome ■ Genetic diversity ■ Haplotype

INTRODUCTION

The main function of the Y chromosome is determination of sex and fertility in men. It is believed that the X and Y chromosomes evolved from ordinary pair of autosomes about 300 million years ago [1-18].

Previously it was thought that the Y chromosome contains a few genes and it functionally depleted and that its large part consists of noncoding DNA. Only in the last decade, there were studies that have greatly expanded our understanding of the structure and functions of the haploid genome. The first studies on the phylogeny of the Y chromosome began in the mid 80's, when polymorphisms of 12f2 [1] and 49a, f [18-22] were discovered. With the development of new technologies (PCR, sequencing, DHPLC) the number of known markers began to increase [13].

In nonrecombining region (NRY) a large number of polymorphic sites were discovered, including more than 400 biallelic markers [4, 23-27].

The special properties of the Y chromosome, such as the haploid state nonrecombining maintenance of large areas and the transfer of the paternal line, make it a unique tool for use in medical, population and evolutionary genetics.

Increasing the resolution of the phylogenetic tree of Y chromosome haplogroups will lead to a more subtle temporal and spatial resolution for the study of human migration [7].

Structure of the Y Chromosome: The size of the Y chromosome is about 60 million base pairs and most of it (95%) is presented with nonrecombining areas that do not have exchange of regions in the process of meiosis with X chromosome (NRY-non-recombining region of the Y or MSY - male-specific region of the Y). At the same time, NRY flanked by small areas for which recombination is common and fairly frequent in meiosis. Detailed physical map of the human Y chromosome has been proposed by Tilford *et al.* [28-35].

Nonrecombining part of the Y chromosome is divided into two parts - the heterochromatic and euchromatic region. In heterochromatic region of Y chromosome three sections are included, one of which is centromeric (~ 1 million bp). The second large region of more than 40 million base pairs is located in the distal portion of the long arm of the chromosome. Third heterochromatic block represents a small islet of 400 kb containing the tandem repeats of 3000 bp length 125 is in the proximal part of the long arm [29].

Total length of euchromatin regions is about 24 million bp, including 8 million bp located on the short arm (Yp) and 14.5 million bp on the long arm (Yq).

Euchromatin sequence is divided into three classes - transferred to the X chromosome (X-transposed), X-degenerate (X-degenerated) and amplified regions (ampliconic segments) [25].

The presence of the first class of sequences (X-transposed) 3.4 million base pairs in length in the Y chromosome is due to massive transfer of them to the X

