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Allelic diversity and mutation at the hypervariable minisatellite locus DYF155S1 (MSY1)

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Abstract

Mutation rate and allelic diversity in DYF155S1 were investigated in Norwegian father/son materials. The detection of *de novo* mutations in 1071 father/son pairs showed an allele length mutation rate of about 2.5%. A further investigation of 300 pairs revealed two "boundary switch" mutations, giving a combined mutation rate of about 3.2%. About 60% of mutations lead to a one step change in MVR-code, while the others lead to two or more changes. A very high MVR-code variation was revealed. Most alleles had the modular structure 1-3-4 (49.5%) or 3-1-3-4 (39.5%). There was a clear association between allele size and modular structure, and the combination of allele length and modular structure revealed a further subgrouping of the alleles. Two groups of males from haplogroup 1 and 2, with individuals in each group also sharing haplotype in eight Y-STRs, were distributed in two different DYF155S1 population subgroups. Only two of the males shared an identical MVR-code. The high allelic variation demonstrated in these males suggests that DYF155S1 might be a powerful tool to differentiate males even if they are identical in Y-STRs and bi-allelic markers.

Keywords: DYF155S1; MSY1; Y-chromosome variation; Minisatellite mutation

1. Introduction

Minisatellite locus DYF155S1 is a hypervariable minisatellite composed of 25 bp AT-rich repeats with basepair substitutions at certain positions in the repeats [1]. Minisatellite Variant Repeat typing by PCR (MVR-PCR) [1,2] gives the number and type of repeats

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along the repeat array (MVR-code). The repeat array of this minisatellite consists of several blocks of identical repeats. The type, number and succession of such blocks are referred to as the modular structure of a given allele.

A highly variable Y-chromosome marker like MSY1 is of considerable interest as a tool in population genetics and forensic science. The aims of this investigation were to study mutation in DYF155S1, to reveal the allelic variation in a Norwegian population sample, and in a selected sample of males identical in Y-STRs and bi-allelic Y-markers.

2. Materials and methods

A total of 1071 confirmed father/son pairs from a large number of paternity cases were screened for allele length mutations by agarose gel-electrophoresis. 300 father/son-pairs with identical allele length were further analysed by MVR-PCR to detect mutations in MVR-code without allele length change (“boundary switch” mutations). 321 Norwegians were analysed by MVR-PCR to give the general variation in MSY-1. A selected material consisted of 27 unrelated males; 21 males belonging to haplogroup 2, and sharing haplotype in eight Y-STRs, and 6 males from haplogroup 1, sharing another haplotype in eight Y-STRs.

The MVR-PCR method published by Jobling et al. [1] was slightly modified with repeat-specific primers labelled with fluoroscent dyes. A three state MVR-PCR that detected type 1, 3 and 4 variants were performed. Other repeat variants were observed as non-existing peaks, and referred to as null-repeats. Different combinations of forward and reverse typing were used to get the MVR-codes of all alleles analysed by MVR-PCR.

3. Results

A total of 27 de novo allele length mutations were revealed in the 1071 father/son pairs. Two “boundary switch” mutations were revealed in the 300 pairs further analysed by MVR-PCR. This gives mutation rates of about 2.5% and 0.7%, respectively, and a combined MVR-code mutation rate of about 3.2%. The distribution of MVR-code changes shows that about 60% of the mutations lead to a one step change in the MVR-code, while the others lead to two or more changes in the MVR code.

The MVR-PCR typing revealed 267 different alleles in the 321 males, while the small groups of identical alleles consisted of five or less males. In three alleles additional 0 repeats were revealed. 49.5% of the alleles were of the 1–3–4 modular structure while 39.5% of the alleles were of the 3–1–3–4 type. Most of the alleles with other modular structures might be derived from the two main types by a single mutation at the boundary between modules.

There was a clear association between allele size and modular structure, giving a multi modal distribution with several subgroups of alleles. This grouping of the alleles probably reflects that alleles in the subpopulations share a closer common ancestry.

The two groups of males identical in bi-alleleic markers and Y-STRs were distributed in two different population subgroups with a limited size and modular structure within their

groups. This is as expected from alleles sharing a close common ancestry. Even if these alleles are identical in eight Y-STRs, only two of the alleles shared an identical MVR-code.

4. Discussion

A very high MVR-code variation in DYF155S1 was revealed in this investigation. This is in agreement with previous studies [1]. The mutation profile and mutation rate based on de novo mutations might provide a basis for methods using DYF155S1 variation to estimate age of, or relationship between, similar Y-haplotypes. The MVR-PCR analysis of males with identical haplotypes in bi-allelic and Y-STRs shows that MVR-code variation in DYF155S1 might be a powerful tool to subgroup or differentiate males identical in Y-STRs and bi-allelic markers.

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