# Technical Note on Typing DYS518

**October 24, 2024**

## Background

DYS518 is included in the Yfiler™ Plus kit (Thermo Fisher Scientific) and the mYSTR and CombiSTR Plus (NimaGen) MPS kits. It is not included in the minimal set of markers for YHRD-inclusion (1). DYS518 is considered a rapidly mutating locus with a mutation rate of 1.3e-2 (2), thereby making it potentially useful in differentiating among relatives. Y-STR loci are components of Y-STR haplotypes consisting of a series of linked allele numbers. Small databases of allele number-based haplotype frequencies are available (1), but sequence-based frequency databases have not yet been developed.

## Nomenclature

A 2010 report described DYS518 as a [AAAG]n [GGAG]1 [AAAG]4 N6 [AAAG]m repeat motif (3). This nomenclature includes the two variable [AAAG] repeats on either end, along with **five** neighboring non-variable repeats [GGAG]1 and [AAAG]4, resulting in allele numbers with a “n plus m plus 5” format.

A later 2014 report described DYS518 as a [AAAG]3 [GAAG]1 [AAAG]n [GGAG]1 [AAAG]4 N6 [AAAG]m repeat motif (4). This nomenclature follows the ISFG recommendations for complex markers by including the two variable [AAAG] repeats on either end, but with **nine** neighboring non-variable repeats [AAAG]3 [GAAG]1 [GGAG]1 [AAAG]4, resulting in allele numbers with a “n plus m plus 9” format.

Since the earlier nomenclature did not include four non-variable repeats that the later nomenclature did, allele numbers using this earlier nomenclature would need to add four to be concordant with results using the later nomenclature.

## Location

The STR locus for DYS518 begins and ends at the same genomic location regardless of which nomenclature was used. And the total number of nucleotides found within the STR also remains constant regardless of which nomenclature is used. For example, the GRCh38 reference sequence begins at 15207988, ends at 15208145, and contains 158nt regardless of which nomenclature was used (see Table 1).

**Table 1. Two different nomenclatures used in literature applied to the GRCh38 reference sequence.**

| Bracket Scheme | Reference Sequence | STR Location | Repeat Count | Ref |
|---|---|---|---|---|
| [AAAG]n [GGAG]1 [AAAG]4 N6 [AAAG]m | [AAAG]16 [GGAG]1 [AAAG]4 N6 [AAAG]13 | 15207988>>>158nt<<<15208145 | 34 | 3 |
| [AAAG]3 [GAAG]1 [AAAG]n [GGAG]1 [AAAG]4 N6 [AAAG]m | [AAAG]3 [GAAG]1 [AAAG]16 [GGAG]1 [AAAG]4 N6 [AAAG]13 | 15207988>>>158nt<<<15208145 | 38 | 4 |

## Allele Numbers

Allele numbers are defined in CE-based analysis (i.e. analysis of the DNA fragment length) using sizing standards. However, allele numbers are defined in MPS-based analysis (i.e. analysis of the DNA sequence) by subtracting the nucleotides in the sequence that do not, by convention, contribute to allele number determination. Since current allele databases have been built based on CE-derived allele numbers, allele number assignments in MPS should seek concordance with CE-based allele numbers.

**Table 2. Genotype of 2800M DNA standard bracketed by two different nomenclatures.**

| Bracket Scheme | STR Region | Allele numbering Format | Allele Number |
|---|---|---|---|
| [AAAG]n [GGAG]1 [AAAG]4 N6 [AAAG]m | [AAAG]16 [GGAG]1 [AAAG]4 N6 [AAAG]13 | n + m + 5 | 32 |
| [AAAG]3 [GAAG]1 [AAAG]n [GGAG]1 [AAAG]4 N6 [AAAG]m | [AAAG]3 [GAAG]1 [AAAG]16 [GGAG]1 [AAAG]4 N6 [AAAG]13 | n + m + 9 | 36 |

U1-C08-**2800M**_S59:

CAGGTGAATCACTTGAACCTGGGAAACAGAGGTTGCAGTGGGCCAAGATCTCGTCATTGCACTCCAGCC TGGGCAACACAAGTGAAACTGCTTCTCGAAA AAAG AAAG AAAG GAAG AAAG AAAG AAAG AAAG AAAG AAAG AAAG AAAG AAAG AAAG AAAG AAAG AAAG GGAG AAAG AAAG AAAG AAAG GAAGAG AAAG AAAG AAAG AAAG AAAG AAAG AAAG AAAG AAAG AAAG AAAG AAAG AAAG AAAG GAAGAGGGAGGGAAGAAGAAAAGAAAGAAGGAAGGAAGGAAGGAGGGAAAAGAAGGAAAGAAAAGAA AGAAATCACCCATGGTAAGAGCTGAAGATGTGA

## Literature Cited


1. https://yhrd.org/pages/resources/locus_information
2. Hao, Shicheng; Chen, Jinghan; He, Hang; Yuan, Li. Mutation Studies of 31 Highly Mutated Y-chromosomal Short Tandem Repeat Systems in the Han Population of Northern China. *Journal of Forensic Science and Medicine* 10(2):pp.155-162, Apr-Jun 2024. DOI: 10.4103/jfsm.jfsm_33_24
3. M.E. D'Amato, L. Ehrenreich, K. Cloete, M. Benjeddou, S. Davison. Characterization of the highly discriminatory loci DYS449, DYS481, DYS518, DYS612, DYS626, DYS644, and DYS710. Forensic Sci. Int. 4 (2010) 104-110.
4. J. Mulero, J. Ballantyne, K. Ballantyne, B. Budowle, M. Coble, L. Gusmao, L. Roewer, M. Kayser. Nomenclature update and allele repeat structure for the markers DYS518 and DYS449. Forensic Sci. Int.: Genetics 13 (2014) e3.