The Short Tandem Repeat Locus vWF2 Consists of Two Polymorphic Sub-Loci

Albert P. Haddad and Rosemary L. Sparrow
Australian Red Cross Blood Service- Victoria, Southbank, Victoria, Australia

Intron 40 of the vWF gene contains three highly polymorphic short tandem repeat (STR) regions, VWA31, VWF1 and VWF2. This study demonstrates that the VWF2 locus is more complex than originally thought, consisting of two polymorphic sub-loci. Consequently, this makes VWF2 one of the most useful STR loci for human identification and parentage testing investigations.

Sequence analysis of the VWF2 region revealed the presence of two distinct regions of TCTA motif repeats; the first between nucleotides 2257 and 2267 (assigned the name of VWF2-a sub-locus) and the second between nucleotides 2295 and 2344 (assigned, VWF2-b). This has the implication that VWF2 alleles of the same nucleotide length may in fact have different nucleotide sequences.

Ninety nine unrelated individuals, representing seven different VWF2 alleles, were tested for VWF2-a and VWF2-b. VWF2-a sub-locus was found to have five alleles representing 0 to four motif repeats, whilst VWF2-b sub-locus had seven alleles of eight to 14 motif repeats. In the 198 chromosomes analyzed, 23 allelic configurations (VWF2-a and VWF2-b) were identified. Theoretically, there are 35 possible allelic configurations.

Determination of the VWF2-a and VWF2-b sub-loci provides a powerful method for enhanced discrimination compared to regular VWF2 locus typing.