Flemish Population Data, Mutation and Locus Structure of STR D12S1090.

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Locus D12S1090 (GDB: 376560) is part of the Lifecodes Multiplex-I kit and contains, according to the manufacturer, 25 different alleles. We typed 204 chromosomes from the Flemish population for this locus, and for the two other loci included in the same kit: D3S1744 (GDB: 686595) and D18S849 (GDB 684741) with respectively 9 and 10 different alleles. The distributions for our population fit well with the distributions made available by the manufacturer for 220 North-American caucasians. For D12S1090, heterozygosity was 90.8 % and power of exclusion was 84.1%. These high values are comparable with other highly variable loci (e.g. ACTBP2) and makes this locus very attractive for parentage testing.

When performing a paternity test, a mutation was observed for locus D12S1090 (fourteen other loci included paternity, yielding a probability of paternity of W = 99.991 %). Since mutations can contain clues to the understanding of the mutation mechanisms, we cloned and sequenced the paternal and mutant child alleles. As is often the case, this mutation of paternal origin was an expansion with the addition of one tetrameric repeat unit. No differences were found in surrounding sequences.

The structure of the locus is complex. The repeat has a sequence rich in AT repeats on both sides and is interrupted by two single and one dinucleotide insertion. The repeat, ATCT, is composed of 3 ″weak″ basepairing nucleotides (A and/or T) and one ″strong″ basepair (G or C). Almost all repeat motifs, independent of length, follow the rule (A+T) > (G+C). It is likely that these observations are related to the polymorphic behaviour of these sequences. Triplet repeats like CAG (G+C>A) are the cause of a growing number of neurological diseases and some can expand in one generation with more than 1000 repeat copies. Very probably, other mechanisms underlie this mutational behaviour.