**Evaluation of Additional Y-STR Loci to Resolve Common Haplotypes**

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We are investigating the advantages of additional Y-STR loci beyond those currently available in commercial kits such as Promega’s Powerplex Y [1] and Applied Biosystem’s Yfiler [2]. The approach for selecting and evaluating these loci is detailed as well as their ability to resolve samples with common types that could not be resolved with the commercial Y-STR kits. A total of 62 Y-STRs have been characterized with a subset of U.S. population samples [3,4]. From these studies, the Y-STR loci that appear to be the most presently useful include DYS449, DYS461, DYS505, DYS522, DYS527 (a duplicated locus), DYS532, DYS534, DYS576, DYS607, DYS650, DYS652, DYS709, DYS710, DYS712, DYS 715, and DYS 724 (a duplicated locus).

Another added benefit of these additional Y-STRs is their ability to increase the power of discrimination between closely related male individuals, such as fathers and sons. We have examined 350 father-son sample pairs from Caucasian, African American, Hispanic and Asian populations using the 17 Y-STR loci in the Yfiler kit. A total of 19 mutations were observed as well as deletions and duplications [5]. In addition, we are updating SRM 2395 Human Y-chromosome DNA profiling standard certificate with additional Y-STR loci in order to support the genetic genealogy community which is continually adding new Y-STRs.

**Why use Y-STRs?**

Y-chromosome STRs are attractive to the forensic community due to their male-specific amplification. This quality is useful in sexual assault situations when the female victim’s DNA is in fair excess to the male perpetrator’s DNA. Y-STRs are also beneficial in difficult cases with no maternal evidence or only fingernail scrapings from the victim. Other applications for Y-STRs include paternity testing, tracing paternal lineages to aid in missing persons investigations, historical studies, and to help link families through genetic genealogy.

Most forensic DNA laboratories will only use commercially available STR kits for quality control issues.

**Y-STR Testing Kits**

<table>
<thead>
<tr>
<th>Kit Name</th>
<th>Loci</th>
<th>Number of Loci</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>PowerFlex® Y</td>
<td>54</td>
<td>54</td>
<td>Male-specific, 12-locus fluorescent STR kit</td>
</tr>
<tr>
<td>AmpFlSTR® Yfiler™</td>
<td>48</td>
<td>48</td>
<td>Male-specific, single amplification 17-plex PCR assay</td>
</tr>
</tbody>
</table>

**Y-STR Mutation Rates**

To-date, we have examined 350 father-son pairs (700 samples). Buccal swabs were extracted with DNA IQ, quantified with an Alu qPCR assay (Nicklas and Buel 2003), and typed with Identifiler and Yfiler STR kits to obtain information on 15 autosomal STRs and 17 Y-STRs. Autosomal allele sharing confirmed paternity. As noted previously (Butler et al. 2005), duplications and deletions can occur on the Y-chromosome, which may be seen in both father and son.

**Y-STR Mutation Rates**

In almost 350 father-son sample pairs, we observed 19 differences between father and son with the 17 Y-STR loci in the Yfiler kit. Eight mutations resulted in the loss of a repeat in the son and 11 loci gained a repeat. All samples resulted in single mutation except one sample which was a two repeat loss at Y-GATA-H1. Also, one sample pair was found to have two mutations (DYS356 and DYS468). Additional mutations in father and son pairs for the 17 Y-STR loci have been reported in the literature.

**Selection and Characterization of New Y-STR Loci**

We plan to develop large multiplex reactions with the best additional Y-STR loci. Potential new Y-STR loci were selected based on best candidates from previous studies (Redd et al. 2002, Kayser et al. 2004, Leat et al. 2006). The selected loci were examined in a U.S. population screen consisting of 31 Caucasians, 32 African Americans, and 32 Hispanics. The number of alleles and calculated gene diversity values are shown below for 8 Y-STR loci. Loci highlighted in yellow have been sequenced and are included in the SRM 2395 certificate. We are currently sequencing loci highlighted in yellow.

**Summary**

- We are examining new Y-STR loci beyond those available in commercial kits.
- As expected, more Y-STR loci increase the ability to resolve samples from one another particularly those with a common type.
- Studies are underway to see if the sample pairs we are going to measure mutation rates and to assist understanding which and how many Y-STRs may be optimal for differentiating between closely related individuals.

**Acknowledgments and Disclaimer**

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