

## **CONCORDANCE TESTING COMPARING COMMERCIAL STR MULTIPLEX KITS WITH A STANDARD DATA SET**

Carolyn R. (Becky) Hill, Margaret C. Kline, David L. Duewer, John M. Butler

National Institute of Standards and Technology, Applied Genetics Group, Gaithersburg, MD 20899-8314

Concordance evaluations are important to detect allelic dropout or “null alleles” present in a data set. These studies are performed because there are a variety of commercial STR multiplex kits with different configurations of STR markers available to the forensic community [1]. The electrophoretic mobility of the markers can vary between kits because the primer sequences were designed to amplify different polymerase chain reaction (PCR) product sizes. When multiple primer sets are used, there is concern that allele dropout may occur due to primer binding site mutations that affect one set of primers but not another [2]. These null alleles become evident only when data sets are compared. Null alleles are a concern because they can result in a false-negative or incorrect exclusion of samples that come from a common source (if the samples have been typed with kits using different PCR primers). A base pair change in the DNA template at the PCR primer binding region can disrupt primer hybridization and result in a failure to amplify and detect an existing allele [2].

Multiple concordance studies have been performed at NIST with a standard sample set (~1450 in-house U.S. population samples) using various STR multiplex kits including Applied Biosystems Identifiler, MiniFiler, NGM, NGM SElect, SGM Plus, and Profiler Plus kits, as well as Promega PowerPlex 16, ESX 17 and ESI 17 Systems [3-8]. Various discordant results have been identified using concordance software developed at NIST, confirmed by DNA sequencing and reported to the forensic community on the null allele web page of STRBase [9].

A summary of the results of our concordance studies will be shown in order to help assess the benefits of performing concordance testing using a standard data set with STR multiplex kits that have different primer sequences for the same markers.

### **References:**

- [1] Hill, C.R., Kline, M.C., Duewer, D.L., Butler, J.M. (2010) Strategies for concordance testing. *Profiles in DNA (Promega)*, 13(1).
- [2] Butler, J.M. (2010) Fundamentals of Forensic DNA Typing, Elsevier Academic Press.
- [3] Butler, J.M., Schoske, R., Vallone, P.M., Redman, J.W., Kline, M.C. (2003) Allele frequencies for 15 autosomal STR loci on U.S. Caucasian, African American, and Hispanic populations. *J. Forensic Sci.* [48\(4\):908-911.](#)
- [4] Hill, C.R., Kline, M.C., Mulero, J.J., Lagace, R.E., Chang, C.-W., Hennessy, L.K., Butler, J.M. (2007) Concordance study between the AmpFISTR MiniFiler PCR Amplification Kit and conventional STR typing kits. *J. Forensic Sci.* [52\(4\): 870-873.](#)
- [5] Hill, C.R., Duewer, D.L., Kline, M.C., Sprecher, C.J., McLaren, R.S., Rabbach, D.R., Krenke, B.E., Ensenberger, M.G., Fulmer, P.M., Stort, D.R., Butler, J.M. (2011) Concordance and population studies

- along with stutter and peak height ratio analysis for the PowerPlex® ESX 17 and ESI 17 Systems. *Forensic Sci. Int. Genet.* 5(4): 269-275.
- [6] Coble, M.D. and Butler, J.M. (2005) Characterization of new miniSTR loci to aid analysis of degraded DNA. *J. Forensic Sci.* 50: 43-53.
- [7] Hill, C.R., Kline, M.C., Coble, M.D., Butler, J.M. (2008) Characterization of 26 miniSTR loci for improved analysis of degraded DNA samples. *J. Forensic Sci.* 53(1):73-80.
- [8] Hill, C.R., Butler, J.M., Vallone, P.M. (2009) A 26plex autosomal STR assay to aid human identity testing. *J. Forensic Sci.* 54(5): 1008-1015.
- [9] <http://www.cstl.nist.gov/biotech/strbase/NullAlleles.htm>