## SickKids Genome Diagnostics

### **IDENTITY TESTING: ZYGOSITY TESTING**

Individuals can be distinguished from one another by DNA fingerprinting, which compares variable DNA markers in different regions of the genome. Each person has his or her own pattern of DNA markers - a DNA 'fingerprint'. PCR amplification followed by capillary electrophoresis allows comparison of these markers between different individuals.

DNA fingerprinting can also be used to evaluate the purity of tissue samples obtained for other kinds of tests. Maternal cell contamination (MCC) of fetal cell samples obtained for prenatal analysis can be assessed in this way, and DNA fingerprinting can also be used to distinguish between the X and Y chromosomes for fetal sex determination.

#### **TEST METHODS**

Identity testing is performed by studying 15 DNA microsatellite markers and a sex-differentiating marker (D5S818, vWA, D13S317, THO1, D7S820, TPOX, CSF1PO, D8S1179. D21S11, D3S1358, D2S1338, D19S433, D16S539. D18S51, FGA and AMELX/Y). PCR amplification of these targeted followed by capillary regions electrophoresis allows comparison markers in the samples of these provided.

#### **ZYGOSITY TESTING**

Zygosity testing compares DNA marker patterns between siblings to assess whether individuals from a multiple gestation (e.g. twins, triplets) are monozygotic (identical) or dizygotic (fraternal). The testing can be done after birth or prenatally.

Dizygotic twins are identified if their DNA fingerprints do not match at two or more markers. When the DNA fingerprints match completely, the twins are most likely monozygotic.

DNA fingerprinting does not match the entire genome of the children, so there is a small probability that a pair of twins are dizygotic despite matching at all DNA markers analyzed. A risk estimate of this occurrence is provided with each report.

#### For More Information

Understanding Gene Testing: <u>http://www.accessexcellence.</u> org/AE/AEPC/NIH/index.html

Waye JS (1993) Forensic identity testing using highly polymorphic DNA markers: current status and emerging technologies. *Transfusion Medicine Review* 7: 193-205.

To locate a genetics center near you, please visit the Canadian Association of Genetic Counsellors website at <u>www.cagc-accg.ca</u> or the National Society of Genetic Counsellors website at <u>www.nsgc.org</u>

# **SickKids**

1. This test was developed and its performance characteristics validated by Diagnostics the Genome Laboratory at the Hospital for Sick Children. It has not been cleared or approved by the U.S. Food and Drua Administration. The FDA has determined that such clearance or approval is not necessary. This test is used for clinical purposes.