Short Tandem Repeat (STR) Assays: Bone Marrow Engraftment, Maternal Engraftment, Zygosity Testing and Maternal Cell Contamination

Description:

Short tandem repeats (STR) are small pieces of highly repetitive DNA sequences. However, the number of repeat units within a specific STR locus can be highly variable between individuals. The highly polymorphic nature of STR loci makes them incredibly useful in distinguishing between two individuals in a variety of assays. These assays utilize polymerase chain reaction (PCR) with capillary electrophoresis to determine the number of repeats at 25 different autosomal STR loci.

Specimens:

Bone Marrow Engraftment (BME):

BME is used to distinguish between donor and host DNA after bone marrow transplant. Specimens from both the bone marrow donor and recipient are required when performing BME for the first time. Subsequent tests require only recipient sample.

Donor & Pre-transplant Host Specimens:

- 3 mLs whole blood in lavender top (EDTA) Preferred
- 3 mLs whole blood in green top (NaHep)
- 3 mLs bone marrow in green top (NaHep)
- 2 cyto brushes*

Post-transplant Specimens:

- 3 mLs whole blood in lavender top (EDTA) Preferred
- 3 mLs whole blood in green top (NaHep)
- 3 mLs bone marrow in green top (NaHep)

White blood cell subcell type BME study:

Cell types available for sorting in the subcell type study are B cell, Myeloid cell, T cell, and NK cell.

White blood cell sub-population BME Specimens:

- At least 15 mLs whole blood in lavender top (EDTA) Preferred due to quality of separation
- At least 15 mLs bone marrow in lavender top (NaHep)

Please contact the Genetics and Genomics Diagnostic Laboratory to schedule this testing at 513-636-4474.

Note: A pre-transplant patient sample and a sample from the bone marrow donor are required to perform the first bone marrow engraftment test.

Maternal Engraftment:

Maternal engraftment evaluates if a child's sample contains DNA from their mother. Analysis requires orders and samples from child/patient and mother.

Maternal Specimens (Labeled with maternal name & DOB):

- 3 mLs whole peripheral blood in lavender top (EDTA) Preferred
- 3 mLs whole peripheral blood in green top (NaHep)
- 2 cyto brushes*
- 1 tube saliva*

Child/Patient Specimens (Labeled with child/patient's name & DOB):

- 3 mLs whole cord or peripheral blood in lavender top (EDTA) Preferred
- 3 mLs whole cord or peripheral blood in green top (NaHep)
- 3 mLs bone marrow in green top (NaHep)

Twin Zygosity Testing:

Twin Zygosity testing determines if twins are monozygotic (identical) or dizygotic (fraternal). Orders and Samples are needed for both twins.

Zygosity Specimens:

- 3 mLs whole blood in lavender top (EDTA) Preferred
- 3 mLs whole blood in green top (NaHep)
- 3 mLs bone marrow in green top (NaHep)
- 2 cyto brushes*
- 1 tube saliva*
- Amniotic fluid - MCC also required
- Chorionic Villi Sampling (CVS) - MCC also required
- Products of Conception (POC) - MCC also required

***Contact the lab at 513-636-4474 to obtain free cyto brushes, saliva collection kits or for other possible fetal specimen types**

Cincinnati Children's®
changing the outcome together

Genetics and Genomics Diagnostic Laboratory
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Email: LabGeneticCounselors@cchmc.org
www.cincinnatichildrens.org/genetics

Specimens (continued):

Maternal Cell Contamination (MCC):
MCC rules out the presence of maternal DNA within a fetal specimen that could interfere with genetic testing.
Maternal Specimens (Labeled with maternal name & DOB)

- 3 mLs whole blood in lavender top (EDTA) Preferred
- 3 mLs whole blood in green top (NaHep)
- 2 cyto brushes*
- 1 tube saliva*

Fetal Specimens (Labeled with maternal name, DOB & specimen type)

- Amniotic fluid
- Chorionic Villi Sampling (CVS)
- Products of Conception (POC)

***Contact the lab at 513-636-4474 to obtain free cyto brushes, saliva collection kits or for other possible fetal specimen types**

Methodology:

This testing can be performed on different specimens according to diagnostic indication:

- Testing on genomic DNA that was isolated from the above stated specimen is performed using the Thermofisher GlobalFiler PCR Amplification Kit to amplify short tandem repeat (STR) sequences. A total of 24 chromosomal loci were amplified for STR analysis. When available, at least three informative sites were used for chimerism calculations. Please contact the laboratory for detailed methodologies or the list of specific loci used for calculations.
- For patients with multiple donors, previous donors will no longer be calculated if undetected from all patient samples after a period of six months. This may affect the number of informative chromosomal loci.
- White blood cell subcell type study: The Genetics and Genomics Diagnostic Laboratory sort Lymphocyte subsets from specimen and the Cincinnati Children's Hospital Diagnostic Immunology Laboratory run cell counts and purities of the cell sort.

Sensitivity:

This test determines the allele size with >98% sensitivity at each locus.

Turn Around Time:

Engraftment by STR-blood, bone marrow or cytobrushes: **3-5 days**
Engraftment by STR-subcell type: **5 days**
Zygosity Testing by STR: **5 days**
Maternal Engraftment by STR: **5 days**
Maternal Cell Contamination by STR: **5 days**

Billing and CPT Codes:

Engraftment by STR- blood, bone marrow or cytobrushes: **81267, 81265**
Engraftment by STR- subcell type: **81268 (x3)**
Zygosity Testing by STR: **81265**
Maternal Engraftment by STR: **81265**
Maternal Cell Contamination by STR: **81265**

Please call 1-866-450-4198 for any pricing or billing questions.

Results:

Results will be reported to the referring physician or health care provider as specified on the test requisition form.

Shipping Instructions:

Please enclose test requisition with sample. All information must be completed before sample can be processed. Place samples in Styrofoam mailer and ship at room temperature by overnight Federal Express to arrive Monday through Saturday.

Ship to:

Genetics and Genomics Diagnostic Laboratory 3333 Burnet Avenue NRB 1013
Cincinnati, Ohio, 45229
513-636-4474

References:

Clark, J., S. Scott, et al. (2014) "Monitoring of Chimerism following Allogeneic Haematopoietic Stem Cell Transplantation (HSCT): Technical Recommendations for the use of Short Tandem Repeats (STR) based techniques, on behalf of the United Kingdom National External Quality Assessment Service for Leucocyte Immuno-phenotyping Chimerism Working Group." British Journal of Haematology 168:26-37.

Frankel, W., A. Chan, et al. (1996) "Detection of Chimerism and Early Engraftment after Allogeneic Peripheral Blood Stem Cell or Bone Marrow Transplantation by Short Tandem Repeats." American Journal of Hematology 52(4): 281-7.

Pindolia, K., N. Janakiraman et al. (1999) "Enhanced Assessment of Allogeneic Bone Marrow Transplant Engraftment using Automated Fluorescent-Based Typing." Bone Marrow Transplantation 24(11): 1235-41.

Thyagarajan, B., S. Young, et al. (2009) "Systematic Analysis of Interference due to Stutter in Estimating Chimerism following Hematopoietic Cell Transplantation." Journal of Clinical Laboratory Analysis 23(5): 308-13.