

DNA Fingerprinting Assays for Evaluating Marrow Engraftment and Chimerism

The UNC Hospitals Molecular Genetics Laboratory performs DNA fingerprinting assays to evaluate the extent of marrow engraftment following marrow transplantation. This assay can also be used to evaluate microchimerism after solid organ transplantation.

Biology & Clinical Utility of DNA Fingerprint Assays

The success of allogeneic marrow transplantation depends on stable engraftment of the donor marrow. The extent of engraftment can be evaluated using the same types of identity tests that are used in forensic and parentage tests. In these assays, highly polymorphic portions of the human genome are PCR-amplified and sized to identify allelic markers of donor versus recipient tissue. These informative markers are then used to evaluate marrow samples after transplant to calculate the proportion of donor versus recipient cells that are present. This assay allows us to identify patients with delayed or failed engraftment. In solid organ transplant recipients, microchimerism studies can be done on blood or other tissues to measure the proportion of donor lymphocytes. In some cases, the presence of donor cells is associated with immune tolerance and a favorable outcome.

Laboratory Testing for Transplant Chimerism and Marrow Engraftment:

Three samples are needed representing **donor**, **recipient**, and the **post-transplant** blood or bone marrow on which donor versus recipient cell proportion is measured. Testing is typically performed on **bone marrow** (1ml, EDTA) or **blood** (3ml, EDTA). Other sample types may be suitable such as fresh or frozen tissue, cheek brushings, or paraffin embedded tissue.

DNA is extracted and then subjected to PCR amplification using multiplex or individual primer sets flanking polymorphic short tandem repeats (PowerPlex, Promega). Products are visualized by capillary gel electrophoresis to determine which markers are informative, and then to evaluate the proportion of donor versus recipient alleles in the post-transplant sample of interest. Mixtures as small as 5% can be reliably detected and quantitated.

Reference:

- Nuckols JD, et al: Evaluation of an automated technique for assessment of marrow engraftment after allogeneic bone marrow transplantation using a commercially available kit. *Am J Clin Pathol* 113:135-40, 2000.

Questions?

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