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BONE MARROW TRANSPLANT TESTING (BMT)

Bone marrow transplantation is an important adjunct to therapy in a number of hematologic, oncologic and genetic/metabolic disorders. Following a bone marrow transplant or peripheral blood transfusion, patient hematopoietic and lymphoid cells are replaced by cells derivedfrom the donor. The percentage of donor versus host cells is indicative of the success of the engraftment, and can be used to track, or characterize, the engraftment process.

In order to define the percentage of cells from two different individuals, host and donor cells must be distinguished from each other. This is done by DNA fingerprinting. This analysis requires three specimens: 1) a sourceof DNA from the recipient before transplantation; 2) a source of DNA from the donor; and 3) a source of DNA from the recipient after transplantation (blood or bone marrow).

TEST METHODS

DNA Fingerprinting: Bone marrow transplant follow-up is performed by studying microsatellite markers in varying regions of the genome, and a sex differentiating marker (D5S818, vWA, D13S317, THO1,D7S820, TPOX, CSF1PO, D8S1179, D21S11, D3S1358, D16S539, D2S1338, D19S433, D18S51, FGA and AMELX/Y). These markers differ greatly between people, and so can be used to distinguish between host and donor cells. PCR amplification of these targeted regions followed by capillary electrophoresis allows comparison of these markers in the donor and recipient.

INDICATIONS FOR TESTING

Assessment of engraftment of donor bone marrow following transplantation.

SPECIMEN REQUIREMENTS

Both pre-transplant (blood or cheek cells) and post- transplant (blood or bone marrow) samples are needed from the recipient, as well as a blood sample from the donor.

TEST SENSITIVITY

Mixtures of donor to recipient DNA can be detected at levels as low as 2%.

POTENTIAL OUTCOMES & INTERPRETATION OF TEST RESULTS

The pattern of markers, the DNA 'fingerprint', are compared pre- and post-transplant to determine if donor cells are being produced in the recipient after the transplant. This comparison is also used to determine the proportion or percentage of donor cells present in post-transplant samples from the recipient, a measure of engraftment.

For More Information

1. Blazar BR, Orr HT, Arthur DC, Kersey JH, Filipovich AH (1985) Restriction fragment length polymorphisms as markers of engraftment in allogenic marrow transplantation. *Blood* 66: 436-44.

2. Ginsbirg D, Antin JH, Smith BR, et al (1985) Origin of cell populations after bone marrow transplantation: Analysis using DNA sequence polymorphisms. *Journal of Clinical Investigation* 75: 596-603.

3. Min GL, Hibbin J, Arthur C, Apperley J, Jeffreys A, Goldman J (1988) Use of minisatellite DNA probes for recognition and characterization of relapseafter allogenic bone marrow transplantation. *British Journalof Haematology* 68: 195-201. .

4. Nollet F, Billiet J, Selleslag D, Criel A (2001) Standardization of multiplex Fluorescent short tandem repeat analysis for chimerism testing. *Bone Marrow Transplantation* 28: 511-18.

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