

Allo-BMT Frequently Asked Questions

Do patients who have undergone autologous BMT (patient's own bone marrow used for transplant) present challenges in terms of genetic testing?

No. In these cases, the patient has received his/her own bone marrow so any sample submitted for genetic analysis will reflect the patient's genetic profile.

What special challenges do patients who have undergone an allogeneic bone marrow transplant (allo-BMT, bone marrow transplant using a donor) present in terms of genetic testing?

Typically, a blood or buccal (mouth) sample is collected from a patient for genetic testing. Blood samples from patients who have had an allo-BMT will not reflect his/her genetic profile, as the majority of circulating blood cells post-BMT are expected to be derived from the bone marrow donor. Although levels of contamination by donor DNA vary in buccal samples, there is a possibility that genetic testing for a patient who has undergone an allo-BMT will provide a result for the donor instead of the patient

What sample type is acceptable for a patient who has undergone a BMT?

Myriad will accept frozen, pelleted cells from cultured fibroblasts for patients who have undergone an allo-BMT. Cultured fibroblasts are derived from a skin punch biopsy. The process of culturing the skin fibroblasts greatly reduces the possibility of contamination from the donor's bone marrow. Sample requirements can be found below.

- The cultured fibroblast sample should be spun down and a frozen pellet of cells from the sample should be sent to the lab (shipped on dry ice).
- Sample needs to contain at least 1.0×10^7 cells.
- Samples should *not* be shipped out on a Friday or Saturday.
- Our lab is not validated to perform cell culture. Therefore, we are able to extract the DNA from the cells present in culture, but cannot culture additional cells from the fibroblast sample.
- The healthcare provider's lab should retain at least one flask and continue to culture the fibroblasts in case another sample is required.

How likely is it that the cultured fibroblasts will comprise cells derived from the bone marrow donor?

After careful review of the literature, Myriad has determined that the likelihood of significant donor contamination in this sample type is low, but will nonetheless perform genotyping on extracted DNA from the cell pellets to document that there is no detectable contamination. As always, ongoing quality assurance will be performed to ensure accurate results for your patients.

Can I re-test my patient who was previously tested via blood or buccal collection?

Yes. Myriad strongly encourages allo-BMT patients who were previously tested via blood or buccal to consider re-testing via cultured fibroblasts. Please contact the Medical Services department at 1-800-469-7423 x 3850 to coordinate this process. Myriad will perform this confirmatory analysis at no charge. (Please note that the laboratory performing sample collection and/or cell culture may charge the patient a fee for their service).