Frequently Asked Question: Can a sample be submitted for DNA extraction/testing after a patient has had a bone marrow / peripheral blood stem cell transplant?

Summary:
With the exception of requests for chimerism studies for bone marrow transplantation monitoring, we **cannot** accept peripheral blood samples from individuals who have had non-autologous bone marrow transplantation / peripheral blood stem cell transplantation for the purpose of DNA extraction and testing. Contact the Molecular Genetics Laboratory (MGL) to discuss possible alternative sources of DNA.

Detailed discussion:
Bone marrow transplantation (BMT) and peripheral blood stem cell transplantation (PBSCT) are procedures that replace bone marrow with healthy marrow stem cells. There are three types of transplants:

- **Autologous**: patients receive their own stem cells
- **Syngeneic**: patients receive stem cells from a monozygotic (i.e., identical) twin/triplet
- **Allogeneic**: patients receive stem cells from another person (related or unrelated to the patient)

Transplantation is commonly used to treat hematopoietic malignancies (e.g., leukemia, lymphoma, multiple myeloma) or to replace bone marrow after it has been damaged by chemotherapy. Other indications for transplantation include hematologic diseases, severe immunodeficiencies and metabolic disorders.

Often, prior to the transplantation, high-dose chemotherapy and/or radiation is given to the transplant recipient. The purpose of this is to help eradicate the patient's diseased bone marrow and reduce post-transplant immune reactions; in the process, these treatments destroy the patient's existing bone marrow. Stem cells harvested from the transplant donor are then injected into the bloodstream of the transplant recipient. These stem cells travel to the bone marrow where it is hoped they will begin the process of engraftment. When engraftment is achieved, the donated stem cells begin to produce blood cells in the transplant recipient.

For certain patients, pre-transplant chemotherapy and/or radiation doses are significantly lower and the transplant recipient's bone marrow is not completely ablated. This is known as a non-myeloablative or reduced-intensity transplant ("mini-transplant"). Unlike in traditional BMT or PBSCT, cells from both the donor and the recipient may co-exist following transplantation and engraftment of the donor stem cells.

When a peripheral blood sample is received in MGL, DNA is extracted from the leukocytes (white blood cells). Following a traditional BMT or PBSCT, once complete engraftment occurs, the leukocytes in the blood of a transplant recipient are expected to have arisen from the stem cells of the donor. Therefore, the DNA obtained from the peripheral blood of non-autologous BMT/PBSCT recipients is actually the DNA of their stem cell *donor*, not the DNA of the transplant recipient. Therefore, it cannot be used to assess the transplant

recipient for constitutional genetic changes the likes of which are assessed using the genetic tests that are performed in MGL.

It may be possible to obtain DNA useful for these clinical studies from alternative tissue sources obtained from transplant recipients. Contact the on-service geneticist to discuss the test desired and the details of the patient's non-autologous transplant (syngeneic or allogenic; traditional or mini-transplant, etc).