Transplant: Frequently Asked Questions

Always consult your physician before taking any action based on the information presented on this page.

What is a Hematopoietic Stem Cell Transplant (HSCT)?

An HSCT is a medical procedure that destroys the stem cells in a patient's marrow and replaces them with stem cells from a matched or partially matched related or unrelated donor. The closer the match, the less likely that the new stem cells will recognize the patient's cells as foreign and attack them, a complication known as graft-versus-host disease (GvHD). At the present time, stem cell transplantation is the only long-term cure for the blood defects in FA. Stem cells can be taken from a donor's bone marrow or peripheral blood, or can be obtained from cord blood harvested at the time of a baby's birth. To prepare for transplant, the patient's own bone marrow is destroyed, making space for the new, healthy stem cells to engraft.

Box A

Pre-transplant laboratory tests

- Confirmatory diagnostic testing for FA (DEB or MMC most commonly)
- Confirmatory HLA typing
- Bone marrow aspirate and biopsy with cytogenetic evaluation
- Infectious disease assessments
  - Prior exposures (cytomegalovirus; hepatitis A, B and C; HIV; HTLV1/2; EBV; syphilis)
  - Presence of active infections (CT scan of sinuses, chest, and abdomen; dental evaluation)
- Organ function assessments
  - Lung (pulmonary function tests, oxygen saturation)
  - Heart (EKG, echocardiogram)
  - Liver (liver enzymes, ultrasound)
  - Kidney (chemistries, nuclear medicine studies such as glomerular filtration rate or GFR, ultrasound)

How do we know when/if it's time to go to transplant?

It is important to note that not every person with FA will need a transplant. Although likely, it is not a certainty. Someone needs a transplant when his/her counts are low enough to require it*. If a patient with FA appears to be a good candidate for transplant based on history and physical examination, a number of routine tests should be performed immediately prior to transplant to verify eligibility and to determine if any adjustments are needed in the treatment. For example, poor kidney function could result in important drug dose adjustments or an anomaly on chest CT might result in additional evaluations, antibiotics, or delay in transplant until resolved. See Box A for a list of the types of tests performed at most transplant centers.

*More info? For specific information on counts and indications that it's time for transplant, see pgs. 223-224 of the Fanconi Anemia Guidelines for Diagnosis and Management.

Are there alternatives to transplant?

While transplant is generally recommended as first-line therapy for bone marrow failure, MDS or leukemia in patients with FA, there are a couple of alternatives for delaying transplant or for patients considered too 'high risk' to undergo transplant therapy. These alternatives may be androgens or hematopoietic growth factors. Gene therapy trials (for FANCA) are also open on two continents to determine if the patient's marrow may be corrected by this method. The patient and family should discuss the risks and benefits of alternative approaches with the hematologists at an FA comprehensive care center.

See p. 238 in the Guidelines book for more info on HSCT alternatives.

Where should we go for transplant?

Because of the unique complications associated with HSCT and the late effects associated with FA itself, it is recommended that whenever possible, patients be cared for at selected centers with comprehensive care clinics specific to FA. Though only a few of these specialized centers exist worldwide, patients who travel to these centers

help advance FA research as much as they themselves benefit from the centers' comprehensive care. The dramatic improvements in transplantation for patients with FA over the past decades, for example, would not have been possible without research that benefited from the concentration of patients at a few centers. Treating patients at selected centers may also help clinicians and researchers improve the management of FA-associated conditions that develop later in life, particularly cancer. See Box B for some useful questions to consider when choosing where to seek treatment. More info? See pgs. 224-229 in the Guidelines book for more information.

What are the success rates of transplant?

Many factors influence the likelihood that a transplant will be successful. Younger children with marrow aplasia (no myelodysplastic syndrome [MDS] or leukemia) do very well, with a better than 90% chance of a good outcome. Transplants are harder in older people and people with MDS or acute myeloid leukemia (AML), or people who have abnormal organ function or past infections. Your doctor will be able to tell you what the chances for success would be in your individual case.

How long will transplant take?

The actual transfer of cells usually takes less than 10 minutes. Most transplant centers will expect the patient to remain near the facility for a minimum of 100 days. While complications can occur after this period, the first 100 days are considered the highest risk period associated with HSCT. During the initial hospitalization for the transplant procedure, patients are isolated (to a room) to reduce exposure to infectious agents. Once the marrow has recovered sufficiently, patients are allowed out of their hospital rooms unless intervening problems prevent this.

Factors that influence the length of time spent away from home include the number of transplant complications such as GvHD and infections, access to a BMT facility closer to the patient's home, the comfort of the referring physician, and evidence of immune recovery. These factors should be discussed on a case-by-case basis.

Ok, it's time to go to transplant. What about logistics, like insurance and a place to stay while getting treatment?

If the insurance company is associated with a transplant center that has limited or no expertise in FA, the insurance company will often give approval for the patient to travel to an experienced FA center once the insurance company understands the differences in the centers' experience and the importance of experience in patient survival. Insurance denials or less than complete coverage for transplant at an FA-experienced transplant center (because they are "out-of-network") can often be contested successfully. FA centers can help with getting insurance approval.

Other FA families are excellent resources when it comes to navigating insurance issues as well as other practicalities like lodging during transplant. If you have a specific question, put it out there on the Family Support Group on Facebook for suggestions from others. Marie Sweeten, FARF Family Services Director, is also available to help answer questions.

Box B

Questions to help assess a transplant center's experience with FA.

- What is the total number of transplants that the center has performed specifically in patients with FA?
- How many FA transplants have been performed each year for the past 5 years? How many of those patients are still alive?
- What treatment regimen do you propose? Please tell me the exact doses of each drug and the radiation dose (if applicable). How many patients have been treated with this regimen at this center? How many are still alive?
- What is the risk of acute and chronic GvHD in FA patients using this regimen? How do you plan to prevent GvHD?
- How long will you follow the patient (me/my child/my spouse)? Who will follow the patient (me/my child/my spouse) long term?

How can I learn more?

Of course, speaking with your doctor(s) is the best way to educate yourself and make decisions. The Fanconi Anemia: Guidelines for Diagnosis and Management are also an excellent resource. The complete Guidelines are available at www.fanconi.org or can be mailed to you. Request your copy(ies) at info@fanconi.org. Finally, updates in stem cell transplantation are provided every year at the Family Meeting at Camp Sunshine in June. ■