Hematopoietic cell transplantation in inborn errors of immunity Part II: others

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ABSTRACT

Inborn errors of immunity (IEI) are a large, heterogeneous group of disorders resulting from deficiencies in immune system development and/or function, leading to increased susceptibility to infections, autoimmunity, autoinflammation, lymphoproliferation, and malignancy. Some diseases are classical hematopoietic cell transplantation (HCT) indications, such as Wiskott-Aldrich Syndrome, but more than 150 different IEI have been transplanted. Each disease and patient should be addressed individually as the clinical manifestations are variable, as well as the pre- and post-transplant care. This consensus strongly recommends that these patients are referred to experienced centers performing HCT for IEI.

Keywords: Hematopoietic Stem Cell Transplantation. Primary Immunodeficiency Diseases. Wiskott-Aldrich Syndrome.

INTRODUCTION

Inborn errors of immunity (IEI) are a large heterogeneous group of disorders that result from deficiencies in the immune system development and/or function. IEI may present increased susceptibility to infections, autoimmunity, autoinflammation, lymphoproliferation, and malignancy[1]. The last report of the International Union of Immunological Societies expert committee on inborn errors of immunity described 559 IEI underlying phenotypes classified in 10 different phenotype groups (Table 1)[2].

Hematopoietic cell transplantation (HCT) has been performed in more than 150 different types of IEI and is considered the treatment of choice for a group with more severe manifestations. The first report of Brazilian experience with HCT for primary imunodeficiencies was published in 2018 and included data from transplants in 221 patients transplanted from July 1990 to December 2015 in 11 centers which participated in the Brazilian collaborative group[3]. The Brazilian Pediatric study group on HCT strongly recommends that centers transplanting patients with IEI should collaborate with international groups and follow international guidelines[4,5].

Table 1. Phenotypic classification of inborn errors of immunity as suggested by the International Union of Immunological Societies Inborn Errors of Immunity Committee.

Phenotypic classification
1. Immunodeficiencies affecting cellular and humoral immunity
2. Combined immunodeficiencies with associated or syndromic features
3. Predominantly antibody deficiencies
4. Diseases of immune dysregulation
5. Congenital defects of phagocyte number, function, or both
6. Defects in intrinsic and innate immunity
7. Autoinflammatory disorders
8. Complement deficiencies
9. Bone marrow failures
10. Phenocopies of Inborn errors of immunity

Source: Bousfha et al.[2].

Transplants in patients with IEI are highly complex and should be performed in centers with continuous and significant experience in these procedures and that participate in collaborative studies. We also recommend that international treatment protocols should be adapted taking into consideration national particularities and limitations. The main IEIs that can be treated with Hematopoietic stem cell transplantation (HSCT) are described in the Tables 2 and 3. Severe combined immunodeficiency will be discussed in another document.

Table 2. Hematopoietic cell transplantation indications for pediatric patients: Brazilian Society of Cell Therapy and Bone Marrow Transplantation Consensus recommendations for inborn errors of immunity.

Disease	Allogeneic				Autologous
	Familiar		Unrelated		
	MSD*	MMFD*	MUD	MMUD	
SCID	Yes	Yes	Yes	Yes	No
WAS	Yes	Yes	Yes	Yes	No
FHLH	Yes	Yes	Yes	Yes	No
CGD	Yes	Yes	Yes	Yes	No
PIRD	Yes	Yes	Yes	Yes	No

SCID: severe combined immunodeficiencies; WAS: Wiskott-Aldrich syndrome; FHLH: familiar hemophagocytic lymphohystiocytosis; CGD: chronic granulomatous disease; PIRD: primary immune regulation disorders; MSD: matched sibling donor; MMFD: mismatched family donor; MUD: matched unrelated donor; MMUD: mismatched unrelated donor; *family donors may be tested for the same genetic mutation as the recipient before hematopoietic cell transplantation. Some diseases have a large phenotypic variability, and siblings may inherit the same disease-causing mutation but exhibit different symptoms or severity. In some recessive diseases, heterozygous donors might be excluded. Source: Elaborated by the authors.

Table 3. Specific disease indications of hematopoietic cell transplantation in inborn errors of immunity.

Severe combined immunodeficiency (SCID)	Standard of care
Hypomorphic SCID / leaky-SCID	Hematopoietic stem cell transplantation indication depends on history of infections or autoimmunity and the patient's performance status
Wiskott-Aldrich syndrome	Best results if performed before 5 years old
Phagocyte disorders: chronic granulomatous disease; leucocyte adhesion deficiency	Best results in younger age and well-matched donors
HLH: familial hemophagocytic lymphohistiocytosis (mutations in: PRF1, UNC13D, STX11, STXBP2); Chediak-Higashi syndrome; Griscelli syndrome type 2 (RAB27A mutation); X-linked lymphoproliferative disease	Standard of care Best results with controlled inflammatory symptoms
Primary immune regulation disorders: IPEX syndrome; CTLA4, LRBA, STAT3 GOF; very early onset inflammatory bowel diseases (interleukin-10, interleukin-10-R)	Hematopoietic cell transplantation may be an option Symptom control before transplant results in better results Cases should be discussed in reference centers

Source: Elaborated by the authors.

WISKOTT-ALDRICH SYNDROME

Wiskott-Aldrich syndrome (WAS) is a rare X-linked genetic disorder usually characterized by a triad of symptoms: thrombocytopenia, eczema, and combined immunodeficiency (affecting both cellular and humoral immunity). About 40% of patients develop autoimmune complications, such as hemolytic anemia, neutropenia, vasculitis, inflammatory bowel disease, kidney disease, or arthritis. There is also an increased risk of malignancies, particularly B-cell lymphoma. After diagnosis, a clinical score based on the presence of thrombocytopenia, eczema, immunodeficiency, autoimmunity, and malignancy can help guide the decision to proceed with transplantation. A score of 3 or higher is considered classical WAS, with autoimmunity and/or malignancy corresponding to a score of 5 (Table 4).

Table 4. Wiskott-Aldrich syndrome score: disease severity classification based on clinical presentation.

	X-linked thrombocytopenia		Wiskott-Aldrich syndrome		
Scores	1	2	3	4	5
Thrombocytopenia	+	+	+	+	+
Immunodeficiency	-	- or +	+	++	-˜++
Eczema	-	- or +	+	++	-˜++
Autoimmune or malignancy	-	-	-	-	+

Source: Elaborated by the authors.

HCT is the treatment of choice for most patients with WAS, correcting hematologic and immunologic defects. Studies have reported excellent overall survival rates, with a five-year overall survival of 91% in patients undergoing HCT. Younger age at transplantation is associated with superior outcomes, consolidating the recommendation for early HCT[6]. Current indications include:

- Patients with a WAS score of 3 or higher;
- Children with severe refractory thrombocytopenia (platelet count < 10,000/µL), especially those younger than 2 years old;
- Patients with mutations leading to the absence of WAS protein expression, even in the absence of severe initial symptoms, to prevent the development of severe comorbidities;
- Adults with classic WAS who have survived without correction or X-linked thrombocytopenia patients who develop late-onset autoimmunity or malignancy.

The recommended conditioning regimen is myeloablative, typically combining busulfan, fludarabine, and r-ATG, with busulfan pharmacokinetics monitoring when available (Table 5). Higher levels of donor chimerism, particularly in the myeloid compartment, are associated with better outcomes, especially regarding the correction of thrombocytopenia and autoimmunity[5].

Table 5. Hematopoietic cell transplantation conditioning and graft-*versus*-host disease (GVHD) prophylaxis regimens for pediatric patients: Brazilian Society of Cell Therapy and Bone Marrow Transplantation Consensus recommendations for inborn errors of immunity.

Donor type	Conditioning	GVHD prophylaxis
Myeloablative		
MSD	BU (AUC 85–95) + Flu 160 mg/m² + ATG 5–10 mg/kg or alemtuzumab 0.5–1 mg/kg	CSA + MTX or MMF
MUD		CSA + MTX or MMF
Haplo		Cy 50 mg/kg + 3 + 4 + CSA + MMF
CB		CSA + MMF or MP
Reduced toxicity		
MSD	BU (AUC 60–70) + Flu 160-180 mg/m² + ATG 5–10 mg/kg or alemtuzumab 0.5–1 mg/kg	CSA + MTX or MMF
MUD		CSA + MTX or MMF
Haplo		Cy 50 mg/kg + 3 + 4 + CSA + MMF
MSD	Flu 150–160 mg/m² + Mel 140 mg/m² ± ATG or alemtuzumab 0.5–1 mg/kg	CSA + MTX or MMF
MUD		CSA + MTX or MMF
Haplo		Cy 50 mg/Kg + 3 + 4 + CSA + MMF

MSD: matched sibling donor; MUD: matched unrelated donor; HAPLO: haploidentical donor; CB: cord blood; CSA: cyclosporin; MTX: metothexate; MMF: mycophenolate mofetil; MP: methylprednisolone. Source: adapted from the European Blood and Marrow Transplant Inborn Errors Working Party recommendations[5].

The use of haploidentical donors, along with post-transplant cyclophosphamide for graft-*versus*-host disease (GVHD) prophylaxis, has emerged as a feasible option when matched related or unrelated donors are unavailable[7]. Bone marrow and cord blood are the preferred stem cell sources, with similar overall survival. Post-HSCT autoimmunity occurs in 14–20% of patients, sometimes associated with mixed chimerism, which is observed in 18–50% of cases and can affect platelet recovery, with studies correlating ≥ 50% donor myeloid engraftment to normal platelet counts[8].

FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS

Familial hemophagocytic lymphohistiocytosis (FHLH) is a hyper-inflammatory syndrome characterized by an uncontrolled immune response, leading to a cytokine storm resulting from a primary immune defect (Table 1). Several mutations have been identified as causative factors of FHLH, including PRF1, UNC13D, STX11, and STXBP2. Additionally, other genetic syndromes, such as Chediak-Higashi syndrome, Griscelli syndrome type II, and X-linked lymphoproliferative disease, can present clinically similarly to hemophagocytic lymphohistiocytosis (HLH).

Notably, up to 20% of primary HLH cases may not have a known genetic mutation. Initial HLH treatment follows the recommended HLH-2004 protocol, with salvage therapies including antibody-based treatments (*e.g.*, thymoglobulin and alemtuzumab), interleukin inhibitors, and JAK/STAT inhibitors, such as ruxolitinib[1,3]. HCT is the preferred treatment for primary HLH, ideally performed in patients in remission with a matched related (unaffected) or unrelated donor. The use of haploidentical donors may be an option in selected cases. The choice of conditioning regimen should consider donor type, disease status, and patient performance. Reduced-toxicity regimens—such as busulfan (with PK monitoring), fludarabine, and serotherapy, or fludarabine, melphalan, and thiotepa (Flu-Mel-TT)—are preferred. While Flu-Mel-TT is associated with lower rates of graft failure and VOD, limited access to thiotepa in Brazil makes busulfan-based regimens a practical alternative with similar survival.

In 2025, thiotepa was approved by the national health authority in Brazil. The availability of this drug may change this scenario. Serotherapy options include thymoglobulin or alemtuzumab. Alemtuzumab is preferred for its superior disease control, but its availability is also limited in our country. GVHD prophylaxis is usually managed with cyclosporine and mycophenolate mofetil[9–11]. The high incidence of graft failure in these patients, especially after reduced intensity conditioning conditioning with fludarabine and melphalan, may require further intervention with second transplant or CD34+ cells boost (strategy not easily available in our country). A stable mixed chimerism (some reports suggest as low as 30%) might be sufficient to protect against disease relapse[12]. In these situations, the evaluation of NK cell cytotoxic activity might be helpful to identify patients in need of further treatment. To date, this test is not commercially available in Brazil.

CHRONIC GRANULOMATOUS DISEASE

Chronic granulomatous disease (CGD) is a rare primary immunodeficiency disorder characterized by functional impairment of the phagocyte NADPH-oxidase complex. HCT can cure CGD and reverse organ dysfunction. The use of reduced toxicity regimens led to improved survival and reduced toxicities, expanding the indications for transplantation in this disease[14]. The control of pre-existing infections and inflammatory manifestations before HCT results in better outcomes. However, active infection should not prevent the patient from undergoing transplantation.

Preferred donors are matched sibling donor or a well-matched unrelated donor. Carrier family donors should be avoided[1,5,13,14]. Non-carrier haploidentical donors are increasingly being used, but graft failure is still an issue[15]. Reduced toxicity conditioning based on busulfan (with pharmacokinetics), fludarabine, and serotherapy (thymoglobulin or alemtuzumab) is the regimen of choice[15]. Alternatively, conditionings based on treosulfan show good results, but this drug is not available in Brazil. Stable mixed chimerism may be sufficient to protect against infections.

PRIMARY IMMUNE REGULATORY DISORDERS

Primary immune regulatory disorders (PIRDs) are inherited immune deficiencies characterized by excessive inflammation, autoimmunity that often affects various tissues, lymphoproliferation, and malignancy. These conditions arise from either a loss or gain of function in genes associated with the regulatory mechanism of the inflammatory or immune response. As many of these diseases were recently described, most lack robust (or any) data regarding transplantation. Thus, no definite recommendations can be currently made in respect of transplant indication or conditioning regimens. An exception is IPEX syndrome, for which a large multicenter study showed an advantage in overall survival and quality of life in transplanted patients compared to those treated with immunosuppression[16]. For PIRDs, disease management seems crucial for the success of HCT, as shown by the European Blood and Marrow Transplant Inborn Errors Working Party, indicating that using Janus Kinases inhibitors as a bridge to transplantation significantly improves overall survival to 91% for patients with JAK/STAT (signal transducer and activator of transcription proteins) signaling-related immune deficiencies, compared to historical data[17].

Another target agent, a mechanism-specific treatment called abatacept, can successfully control LRBA (LPS responsive beige-like anchor protein) deficiency and CTLA4 (cytotoxic T-lymphocyte associated protein 4) insufficiency, serving as a bridge to transplant or even dismiss the need for transplantation[18]. On the other hand, some diseases lack satisfying response to conventional immunosuppression and may be cured with HCT. Very early onset inflammatory bowel disease (VEOIBD) has been reported in patients with interleukin-(IL)-10 and IL-10 receptor (IL-10R) deficiencies, and despite steroids, immunomodulating agents, and surgical intervention, long-term management of inflammatory bowel disease is not satisfactory in these patients. HCT has been successful in several patients[19]. X-linked inhibitor of apoptosis (XIAP) deficiency is characterized by immune dysregulation and a broad spectrum of clinical manifestations, including: HLH and treatment-refractory inflammatory bowel disease. Arnold et al.[20] observed a two-year overall survival of 74% in XIAP deficient HSCT recipients, but with a high incidence of GVHD. PIRDs are a heterogeneous group of diseases, ranging from mild disease to neonatal life-threatening manifestations. Molecular diagnosis is mandatory to define the disease mechanism and benefit for HCT, and control of immune deregulation is desired and should be measured by an objective score such as the immune deficiency and dysregulation activity (IDDA) 2.1 score[21]. Regarding the decision of whether to proceed to transplant, we recommend that these patients be referred to specialized, experienced centers, and discussed by an expert panel.

CONFLICT OF INTEREST

Nothing to declare.

DATA AVAILABILITY STATEMENT

Data sharing is not applicable.

AUTHORS' CONTRIBUTIONS

Substantive scientific and intellectual contributions to the study: Fernandes JF, Muratori R, Loth G, Vieira AK, Klinger P, Franco S and Bonfim C. **Conception and design:** Fernandes JF and Bonfim C. **Manuscript writing:** Fernandes JF, Muratori R, Loth G, Vieira AK, Klinger P, Franco S and Bonfim C. **Final approval:** Fernandes JF, Muratori R, Loth G, Vieira AK, Klinger P, Franco S and Bonfim C.

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