Indications for Haemopoietic Stem Cell Transplantation

This is a list of Primary Immunodeficiency conditions which can benefit from Haemopoietic stem cell Transplantation compiled by:

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A. Profound T cell defects
    1. Severe Combined Immunodeficiency (SCID)
        - ARTEMIS
        - CD3ζ
        - CD3δ
        - CD3ε
        - CD45
        - IL2Rγ
        - IL2Rα[b, d]
        - IL7Rα
        - Jak-3
        - Orai-1
        - RAG1/RAG2
        - Zap-70
    2. SCID with unknown genotype
    3. SCID with residual dysfunctional T cell (CID) [b, c]
    4. Omenn syndrome

B. Profound T cell defects associated with metabolic or multi-organ syndromes
    5. Adenosine Deaminase (ADA) deficiency [a, b]
    6. Cartilage hair hypoplasia (CHH) [b, c]
    7. DNA ligase 4 deficiency, Cernunnos deficiency [b, c, d]
    8. Purine Nucleoside Phosphorylase (PNP) deficiency
    9. Wiskott Aldrich syndrome (WAS) [b, c]

C. Other conditions

10. Autoimmune Lymphoproliferative Syndrome (ALPS) [b, c]
11. CD40L deficiency [a, b, c]
12. CD40 deficiency [a, b, c]
13. Chediak-Higashi syndrome
14. Chronic granulomatous disease [b, c]
15. DiGeorge syndrome [a, c, d]
16. Dyskeratosis congenita (Hoyerall-Hreidarsson syndrome) [d]
17. Griscelli syndrome
18. Hemophagocytic lymphohistiocytosis (HLH) [b]
19. Hyper-IgE syndrome [a, b, d]
20. IFN-γR deficiency [b, d]
21. Immune dysregulation, Polyendocrinopathy, Enteropathy, and X-linked inheritance (IPEX) [b, c]
22. Leukocyte Adhesion Defect (LAD)
23. Mevalonate deficiency
24. Major Histocompatibility Antigens (MHC) class II deficiency
25. NF-Kappa-B Essential Modulator (NEMO) deficiency [d]
26. Reticular dysgenesis
27. Schimke syndrome [b, c, d]
28. Severe congenital neutropenia [d]
29. X-Linked Hypogammaglobulinemia (XLP)

Limitations:

[a]: MRD-BMT only

[b]: Limited to infancy and childhood/severe form, if evidence on adults is lacking.

[c]: Profound immunodeficiency

[d]: Selected cases