

## Primary Immune Deficiency Treatment Consortium

### Journal Articles

1. Griffith LM, Cowan MJ, Kohn DB, et al. Allogeneic hematopoietic cell transplantation for primary immune deficiency diseases: current status and critical needs. *J. Allergy Clin. Immunol.* Dec 2008;122(6):1087-1096. PMID: 18992926, PMCID: PMC3357108
2. Patel NC, Chinen J, Rosenblatt HM, et al. Long-term outcomes of nonconditioned patients with severe combined immunodeficiency transplanted with HLA-identical or haploidentical bone marrow depleted of T cells with anti-CD6 mAb. *J. Allergy Clin. Immunol.* Dec 2008;122(6):1185-1193. PMID: 19084111. Pending PMCID
3. Shearer WT, Notarangelo LD, Griffith LM. Treatment of immunodeficiency: long-term outcome and quality of life. *J. Allergy Clin. Immunol.* Dec 2008;122(6):1065-1068. PMID: 19084107. Pending PMCID
4. Chinen J, Shearer WT. Advances in basic and clinical immunology in 2008. *J. Allergy Clin. Immunol.* Feb 2009;123(2):328-332. PMID: 19203657. Pending PMCID
5. Sarzotti-Kelsoe M, Win CM, Parrott RE, et al. Thymic output, T-cell diversity, and T-cell function in long-term human SCID chimeras. *Blood.* Aug 13 2009;114(7):1445-1453. PMID: 19433858, PMCID: PMC2727406
6. Patel NC, Chinen J, Rosenblatt HM, et al. Outcomes of patients with severe combined immunodeficiency treated with hematopoietic stem cell transplantation with and without preconditioning. *J. Allergy Clin. Immunol.* Nov 2009;124(5):1062-1069 e1061-1064. PMID: 19895994, PMCID: PMC3271026
7. Griffith LM, Cowan MJ, Notarangelo LD, et al. Improving cellular therapy for primary immune deficiency diseases: recognition, diagnosis, and management. *J. Allergy Clin. Immunol.* Dec 2009;124(6):1152-1160 e1112. PMID: 20004776, PMCID: PMC2831471
8. Railey MD, Lokhnygina Y, Buckley RH. Long-term clinical outcome of patients with severe combined immunodeficiency who received related donor bone marrow transplants without pretransplant chemotherapy or post-transplant GVHD prophylaxis. *J. Pediatr.* Dec 2009;155(6):834-840 e831. PMID: 19818451, PMCID: PMC2784223
9. Dvorak CC, Cowan MJ. Radiosensitive severe combined immunodeficiency disease. *Immunol. Allergy Clin. North Am.* Feb 2010;30(1):125-142. PMID: 20113890, PMCID: PMC2818388
10. Chinen J, Shearer WT. Secondary immunodeficiencies, including HIV infection. *J Allergy Clin Immunol.* 2010 Feb; 125(2 Suppl 2):S195-203. PMID: 20042227. Pending PMCID
11. Chinen J, Shearer WT. Advances in basic and clinical immunology in 2009. *J. Allergy Clin. Immunol.* Mar 2010;125(3):563-568. PMID: 20226292, PMCID: PMC2841291
12. Buckley RH. B-cell function in severe combined immunodeficiency after stem cell or gene therapy: a review. *J. Allergy Clin. Immunol.* Apr 2010;125(4):790-797. PMID: 20371393, PMCID: PMC2857969

13. Walter JE, Rucci F, Patrizi L, Recher M, Regenass S, Paganini T, Keszei M, Pessach I, Lang PA, Poliani PL, Giliani S, Al-Herz W, Cowan MJ, Puck JM, Bleesing J, Niehues T, Schuetz C, Malech H, DeRavin SS, Facchetti F, Gennery AR, Andersson E, Kamani NR, Sekiguchi J, Alenezi HM, Chinen J, Dbaibo G, ElGhazali G, Fontana A, Pasic S, Detre C, Terhorst C, Alt FW, Notarangelo LD. Expansion of immunoglobulin-secreting cells and defects in B cell tolerance in Rag-dependent immunodeficiency. *J. Exp. Med.* Jul 5 2010;207(7):1541-1554. PMID: 20547827, PMCID: PMC2901061
14. Zemble R, Luning Prak E, McDonald K, McDonald-McGinn D, Zackai E, Sullivan K. Secondary immunologic consequences in chromosome 22q11.2 deletion syndrome (DiGeorge syndrome/velocardiofacial syndrome). *Clin. Immunol.* Sep 2010;136(3):409-418. PMID: 20472505, PMCID: PMC2917481
15. Buckley RH. Transplantation of hematopoietic stem cells in human severe combined immunodeficiency: longterm outcomes. *Immunol. Res.* Apr 2011;49(1-3):25-43. PMID: 21116871, PMCID: PMC3798033
16. Nicholas S, Krance RA, Hanson IC, et al. Early versus delayed diagnosis of SCID: triumph versus tragedy. *Clin. Immunol.* Jun 2011;139(3):360-362. PMID: 21497138. Pending PMCID
17. Moratto D, Giliani S, Bonfim C, Mazzolari E, Fischer A, Ochs HD, Cant AJ, Thrasher AJ, Cowan MJ, Albert MH, Small T, Pai SY, Haddad E, Lisa A, Hambleton S, Slatter M, Cavazzana-Calvo M, Mahlaoui N, Picard C, Torgerson TR, Burroughs L, Koliski A, Neto JZ, Porta F, Qasim W, Veys P, Kavanau K, Honig M, Schulz A, Friedrich W, Notarangelo LD. Long-term outcome and lineage-specific chimerism in 194 patients with Wiskott-Aldrich syndrome treated by hematopoietic cell transplantation in the period 1980-2009: an international collaborative study. *Blood.* Aug 11 2011;118(6):1675-1684. PMID: 21659547, PMCID: PMC3156052
18. Becker-Herman S, Meyer-Bahlburg A, Schwartz MA, Jackson SW, Hudkins KL, Liu C, Sather BD, Khim S, Liggitt D, Song W, Silverman GJ, Alpers CE, Rawlings DJ. WASp-deficient B cells play a critical, cell-intrinsic role in triggering autoimmunity. *J. Exp. Med.* Sep 26 2011;208(10):2033-2042. PMID: 21875954, PMCID: PMC3182055
19. Marcus N, Takada H, Law J, et al. Hematopoietic stem cell transplantation for CD3delta deficiency. *J. Allergy Clin. Immunol.* Nov 2011;128(5):1050-1057. PMID: 21757226, PMCID: PMC4490832
20. Tison BE, Nicholas SK, Abramson SL, et al. Autoimmunity in a cohort of 130 pediatric patients with partial DiGeorge syndrome. *J. Allergy Clin. Immunol.* Nov 2011;128(5):1115-1117 e1111-1113. PMID: 21835443. Pending PMCID
21. Yu GP, Nadeau KC, Berk DR, de Saint Basile G, Lambert N, Knapnougel P, Roberts J, Kavanau K, Dunn E, Stiehm ER, Lewis DB, Umetsu DT, Puck JM, Cowan MJ. Genotype, phenotype, and outcomes of nine patients with T-B+NK+ SCID. *Pediatr. Transplant.* Nov 2011;15(7):733-741. PMID: 21883749, PMCID: PMC3196791
22. Puck JM. The case for newborn screening for severe combined immunodeficiency and related disorders. *Ann. N. Y. Acad. Sci.* Dec 2011;1246:108-117. PMID: 22236435, PMCID: PMC4474477

23. Puck JM. Neonatal screening for severe combined immunodeficiency. *Curr. Opin. Pediatr.* Dec 2011;23(6):667-673. PMID: 22001765, PMCID: PMC3299571
24. Recher M, Berglund LJ, Avery DT, Cowan MJ, Gennery AR, Smart J, Peake J, Wong M, Pai SY, Baxi S, Walter JE, Palendira U, Tangye GA, Rice M, Brothers S, Al-Herz W, Oettgen H, Eibel H, Puck JM, Cattaneo F, Ziegler JB, Giliani S, Tangye SG, Notarangelo LD. IL-21 is the primary common gamma chain-binding cytokine required for human B-cell differentiation in vivo. *Blood.* Dec 22 2011;118(26):6824-6835. PMID: 22039266, PMCID: PMC3338166
25. Martinez CA, Shah S, Shearer WT, et al. Excellent survival after sibling or unrelated donor stem cell transplantation for chronic granulomatous disease. *J. Allergy Clin. Immunol.* Jan 2012;129(1):176-183. PMID: 22078471. Pending PMCID
26. Chinen J, Shearer WT. Advances in basic and clinical immunology in 2011. *J. Allergy Clin. Immunol.* Feb 2012;129(2):342-348. PMID: 22206779, PMCID: PMC3279946
27. Siberry GK, Leister E, Jacobson DL, et al. Increased risk of asthma and atopic dermatitis in perinatally HIV-infected children and adolescents. *Clin. Immunol.* Feb 2012;142(2):201-208. PMID: 22094294, PMCID: PMC3273595
28. Buckley RH. The long quest for neonatal screening for severe combined immunodeficiency. *J. Allergy Clin. Immunol.* Mar 2012;129(3):597-604; quiz 605-596. PMID: 22277203, PMCID: PMC3294102
29. Hanson IC, Shearer WT. Ruling out HIV infection when testing for severe combined immunodeficiency and other T-cell deficiencies. *J. Allergy Clin. Immunol.* Mar 2012;129(3):875-876 e875. PMID: 22386446. Pending PMCID
30. Puck JM. Laboratory technology for population-based screening for severe combined immunodeficiency in neonates: the winner is T-cell receptor excision circles. *J. Allergy Clin. Immunol.* Mar 2012;129(3):607-616. PMID: 22285280, PMCID: PMC3294074
31. Roberts JL, Buckley RH, Luo B, et al. CD45-deficient severe combined immunodeficiency caused by uniparental disomy. *Proc. Natl. Acad. Sci. U. S. A.* Jun 26 2012;109(26):10456-10461. PMID: 22689986, PMCID: PMC3387083
32. Leechawengwongs E, Shearer WT. Lymphoma complicating primary immunodeficiency syndromes. *Curr. Opin. Hematol.* Jul 2012;19(4):305-312. PMID: 22525579. Pending PMCID
33. Taylor PA, Kelly RM, Bade ND, Smith MJ, Stefanski HE, Blazar BR. FTY720 markedly increases alloengraftment but does not eliminate host anti-donor T cells that cause graft rejection on its withdrawal. *Biol. Blood Marrow Transplant.* Sep 2012;18(9):1341-1352. PMID: 22728248, PMCID: PMC3520609
34. Chan SK, Shearer WT. HCT survival in ADA-SCID: what's the buzz? *Blood.* Oct 25 2012;120 (17):3392-3393. PMID: 23100302. Pending PMCID
35. Punwani D, Gonzalez-Espinosa D, Comeau AM, Dutra A, Pak E, Puck J. Cellular calibrators to quantitate T-cell receptor excision circles (TRECs) in clinical samples. *Mol. Genet. Metab.* Nov 2012;107(3):586-591. PMID: 23062576, PMCID: PMC3483425

36. Mangurian C, Cowan MJ. The missing vital sign. *BMJ*. 2013;347:f4163. PMID: 23833077, PMCID: PMC4688548
37. Buckley RH, Win CM, Moser BK, Parrott RE, Sajaroff E, Sarzotti-Kelsoe M. Post-transplantation B cell function in different molecular types of SCID. *J. Clin. Immunol.* Jan 2013;33(1):96-110. PMID: 23001410, PMCID: PMC3549311
38. Chinen J, Notarangelo LD, Shearer WT. Advances in basic and clinical immunology in 2012. *J. Allergy Clin. Immunol.* Mar 2013;131(3):675-682. PMID: 23374612. Pending PMCID
39. Cattaneo F, Recher M, Masneri S, et al. Hypomorphic Janus kinase 3 mutations result in a spectrum of immune defects, including partial maternal T-cell engraftment. *J. Allergy Clin. Immunol.* Apr 2013;131(4):1136-1145. PMID: 23384681, PMCID: PMC6141242
40. Gelfand EW, Ochs HD, Shearer WT. Controversies in IgG replacement therapy in patients with antibody deficiency diseases. *J. Allergy Clin. Immunol.* Apr 2013;131(4):1001-1005. PMID: 23540617. Pending PMCID
41. Haddad E, Leroy S, Buckley RH. B-cell reconstitution for SCID: Should a conditioning regimen be used in SCID treatment? *J. Allergy Clin. Immunol.* Apr 2013;131(4):994-1000. PMID: 23465660, PMCID: PMC3615028
42. Henderson LA, Frugoni F, Hopkins G, et al. First reported case of Omenn syndrome in a patient with reticular dysgenesis. *J. Allergy Clin. Immunol.* Apr 2013;131(4):1227-1230, 1230 e1221-1223. PMID: 23014587, PMCID: PMC3894621
43. Selleri S, Dieng MM, Nicoletti S, et al. Cord-blood-derived mesenchymal stromal cells downmodulate CD4+ T-cell activation by inducing IL-10-producing Th1 cells. *Stem Cells Dev.* Apr 1 2013;22(7):1063-1075. PMID: 23167734, PMCID: PMC3608091
44. Horn B, Cowan MJ. Unresolved issues in hematopoietic stem cell transplantation for severe combined immunodeficiency: need for safer conditioning and reduced late effects. *J. Allergy Clin. Immunol.* May 2013;131(5):1306-1311. PMID: 23622119, PMCID: PMC5575916
45. Kwan A, Church JA, Cowan MJ, et al. Newborn screening for severe combined immunodeficiency and T-cell lymphopenia in California: results of the first 2 years. *J. Allergy Clin. Immunol.* Jul 2013;132(1):140-150. PMID: 23810098, PMCID: PMC3759317
46. Teigland CL, Parrott RE, Buckley RH. Long-term outcome of non-ablative booster BMT in patients with SCID. *Bone Marrow Transplant.* Aug 2013;48(8):1050-1055. PMID: 23396406, PMCID: PMC3737279
47. Chen R, Giliani S, Lanzi G, et al. Whole-exome sequencing identifies tetratricopeptide repeat domain 7A (TTC7A) mutations for combined immunodeficiency with intestinal atresias. *J. Allergy Clin. Immunol.* Sep 2013;132(3):656-664 e617. PMID: 23830146, PMCID: PMC3759618
48. Dvorak CC, Cowan MJ, Logan BR, et al. The natural history of children with severe combined immunodeficiency: baseline features of the first fifty patients of the primary

- immune deficiency treatment consortium prospective study 6901. *J. Clin. Immunol.* Oct 2013;33(7):1156-1164. PMID: 23818196, PMCID: PMC3784642
49. Henderson LA, Frugoni F, Hopkins G, et al. Expanding the spectrum of recombination-activating gene 1 deficiency: a family with early-onset autoimmunity. *J. Allergy Clin. Immunol.* Oct 2013;132(4):969-971 e961-962. PMID: 23891352, PMCID: PMC3874115
  50. Savic RM, Cowan MJ, Dvorak CC, et al. Effect of weight and maturation on busulfan clearance in infants and small children undergoing hematopoietic cell transplantation. *Biol. Blood Marrow Transplant.* Nov 2013;19(11):1608-1614. PMID: 24029650, PMCID: PMC3848313
  51. Johnson TS, Terrell CE, Millen SH, Katz JD, Hildeman DA, Jordan MB. Etoposide selectively ablates activated T cells to control the immunoregulatory disorder hemophagocytic lymphohistiocytosis. *J. Immunol.* Jan 1 2014;192(1):84-91. PMID: 24259502, PMCID: PMC4177106
  52. Schuetz C, Neven B, Dvorak CC, et al. SCID patients with ARTEMIS vs RAG deficiencies following HCT: increased risk of late toxicity in ARTEMIS-deficient SCID. *Blood.* Jan 9 2014;123(2):281-289. PMID: 24144642, PMCID: PMC3953035
  53. Griffith LM, Cowan MJ, Notarangelo LD, et al. Primary Immune Deficiency Treatment Consortium (PIDTC) report. *J. Allergy Clin. Immunol.* Feb 2014;133(2):335-347. PMID: 24139498, PMCID: PMC3960312
  54. Haddad E, Allakhverdi Z, Griffith LM, Cowan MJ, Notarangelo LD. Survey on retransplantation criteria for patients with severe combined immunodeficiency. *J. Allergy Clin. Immunol.* Feb 2014;133(2):597-599. PMID: 24331379, PMCID: PMC3960313
  55. Chen K, Wu W, Mathew D, et al. Autoimmunity due to RAG deficiency and estimated disease incidence in RAG1/2 mutations. *J. Allergy Clin. Immunol.* Mar 2014;133(3):880-882 e810. PMID: 24472623, PMCID: PMC4107635
  56. Chinen J, Notarangelo LD, Shearer WT. Advances in basic and clinical immunology in 2013. *J. Allergy Clin. Immunol.* Apr 2014;133(4):967-976. PMID: 24589342, PMCID: PMC3988899
  57. Lee YN, Frugoni F, Dobbs K, et al. A systematic analysis of recombination activity and genotype-phenotype correlation in human recombination-activating gene 1 deficiency. *J. Allergy Clin. Immunol.* Apr 2014;133(4):1099-1108. PMID: 24290284, PMCID: PMC4005599
  58. Shearer WT, Dunn E, Notarangelo LD, et al. Establishing diagnostic criteria for severe combined immunodeficiency disease (SCID), leaky SCID, and Omenn syndrome: the Primary Immune Deficiency Treatment Consortium experience. *J. Allergy Clin. Immunol.* Apr 2014;133(4):1092-1098. PMID: 24290292, PMCID: PMC3972266
  59. Shearer WT, Fleisher TA, Buckley RH, et al. Recommendations for live viral and bacterial vaccines in immunodeficient patients and their close contacts. *J. Allergy Clin. Immunol.* Apr 2014;133(4):961-966. PMID: 24582311, PMCID: PMC4009347

60. Pai SY, Logan BR, Griffith LM, et al. Transplantation outcomes for severe combined immunodeficiency, 2000-2009. *N. Engl. J. Med.* Jul 31 2014;371(5):434-446. PMID: 25075835, PMCID: PMC4183064
61. Kwan A, Abraham RS, Currier R, et al. Newborn screening for severe combined immunodeficiency in 11 screening programs in the United States. *JAMA.* Aug 20 2014;312(7):729-738. PMID: 25138334, PMCID: PMC4492158
62. Mandala WL, Ananworanich J, Apornpong T, et al. Control lymphocyte subsets: can one country's values serve for another's? *J. Allergy Clin. Immunol.* Sep 2014;134(3):759-761 e758. PMID: 25171870, PMCID: PMC4150016
63. Dvorak CC, Hassan A, Slatter MA, et al. Comparison of outcomes of hematopoietic stem cell transplantation without chemotherapy conditioning by using matched sibling and unrelated donors for treatment of severe combined immunodeficiency. *J. Allergy Clin. Immunol.* Oct 2014;134(4):935-943 e915. PMID: 25109802, PMCID: PMC4186906
64. Abolhassani H, Wang N, Aghamohammadi A, et al. A hypomorphic recombination-activating gene 1 (RAG1) mutation resulting in a phenotype resembling common variable immunodeficiency. *J. Allergy Clin. Immunol.* Dec 2014;134(6):1375-1380. PMID: 24996264, PMCID: PMC4261008
65. Pai SY, Cowan MJ. Stem cell transplantation for primary immunodeficiency diseases: the North American experience. *Curr. Opin. Allergy Clin. Immunol.* Dec 2014;14(6):521-526. PMID: 25259542, PMCID: PMC4238389
66. Walter JE, Lo MS, Kis-Toth K, et al. Impaired receptor editing and heterozygous RAG2 mutation in a patient with systemic lupus erythematosus and erosive arthritis. *J. Allergy Clin. Immunol.* Jan 2015;135(1):272-273. PMID: 25312763, PMCID: PMC4289116
67. Buchbinder D, Baker R, Lee YN, et al. Identification of patients with RAG mutations previously diagnosed with common variable immunodeficiency disorders. *J. Clin. Immunol.* Feb 2015;35(2):119-124. PMID: 25516070, PMCID: PMC4479182
68. Punwani D, Wang H, Chan AY, et al. Combined immunodeficiency due to MALT1 mutations, treated by hematopoietic cell transplantation. *J. Clin. Immunol.* Feb 2015;35(2):135-146. PMID: 25627829, PMCID: PMC4352191
69. Punwani D, Pelz B, Yu J, et al. Coronin-1A: immune deficiency in humans and mice. *J. Clin. Immunol.* Feb 2015;35(2):100-107. PMID: 25666293, PMCID: PMC4489527
70. Bunupuradah T, Hansudewechakul R, Kosalaraksa P, et al. HLA-DRB1454 and predictors of new-onset asthma in HIV-infected Thai children. *Clin. Immunol.* Mar 2015;157(1):26-29. PMID: 25546395. Pending PMCID
71. Niss O, Sholl A, Bleesing JJ, Hildeman DA. IL-10/Janus kinase/signal transducer and activator of transcription 3 signaling dysregulates Bim expression in autoimmune lymphoproliferative syndrome. *J. Allergy Clin. Immunol.* Mar 2015;135(3):762-770. PMID: 25174872, PMCID: PMC4344440
72. Wahlstrom JT, Dvorak CC, Cowan MJ. Hematopoietic Stem Cell Transplantation for Severe Combined Immunodeficiency. *Curr Pediatr Rep.* Mar 2015;3(1):1-10. PMID: 25821657, PMCID: PMC4371740

73. Kwan A, Puck JM. History and current status of newborn screening for severe combined immunodeficiency. *Semin. Perinatol.* Apr 2015;39(3):194-205. PMID: 25937517, PMCID: PMC4433840
74. Long-Boyle JR, Savic R, Yan S, et al. Population pharmacokinetics of busulfan in pediatric and young adult patients undergoing hematopoietic cell transplant: a model-based dosing algorithm for personalized therapy and implementation into routine clinical use. *Ther. Drug Monit.* Apr 2015;37(2):236-245. PMID: 25162216, PMCID: PMC4342323
75. Chinen J, Notarangelo LD, Shearer WT. Advances in basic and clinical immunology in 2014. *J. Allergy Clin. Immunol.* May 2015;135(5):1132-1141. PMID: 25956014. Pending PMCID
76. Kwan A, Hu D, Song M, et al. Successful newborn screening for SCID in the Navajo Nation. *Clin. Immunol.* May 2015;158(1):29-34. PMID: 25762520, PMCID: PMC4420660
77. Zheng P, Noroski LM, Hanson IC, et al. Molecular mechanisms of functional natural killer deficiency in patients with partial DiGeorge syndrome. *J. Allergy Clin. Immunol.* May 2015;135(5):1293-1302. PMID: 25748067, PMCID: PMC5540306
78. Cowan MJ, Gennery AR. Radiation-sensitive severe combined immunodeficiency: The arguments for and against conditioning before hematopoietic cell transplantation-what to do? *J. Allergy Clin. Immunol.* Jun 2015. PMID: 26055221, PMCID: PMC4641002
79. Burbank AJ, Shah SN, Montgomery M, Peden D, Tarrant TK, Weimer ET. Clinically focused exome sequencing identifies an homozygous mutation that confers DOCK8 deficiency. *Pediatr Allergy Immunol.* Feb 2016;27(1):96-98. PMID: 26235511, PMCID: PMC4724217
80. De Ravin SS, Wu X, Moir S, et al. Lentiviral hematopoietic stem cell gene therapy for X-linked severe combined immunodeficiency. *Sci Transl Med.* Apr 2016;8(335):335ra357. PMID: 27099176, PMCID: PMC5557273
81. Merkel PA, Manion M, Gopal-Srivastava R, et al. The partnership of patient advocacy groups and clinical investigators in the rare diseases clinical research network. *Orphanet J. Rare Dis.* May 2016;11(1):66. PMID: 27194034, PMCID: PMC4870759
82. Selleri S, Bifsha P, Civini S, et al. Human mesenchymal stromal cell-secreted lactate induces M2-macrophage differentiation by metabolic reprogramming. *Oncotarget.* May 2016;7(21):30193-30210. PMID: 27070086, PMCID: PMC5058674
83. Brauer PM, Pessach IM, Clarke E, et al. Modeling altered T-cell development with induced pluripotent stem cells from patients with RAG1-dependent immune deficiencies. *Blood.* Aug 2016;128(6):783-793. PMID: 27301863, PMCID: PMC4982452
84. Griffith LM, Cowan MJ, Notarangelo LD, et al. Primary Immune Deficiency Treatment Consortium (PIDTC) update. *J. Allergy Clin. Immunol.* Aug 2016. PMID: 27262745, PMCID: PMC4986691

85. Cowan MJ. The Primary Immune Deficiency Treatment Consortium: how can it improve definitive therapy for PID?. *Expert Rev Clin Immunol*. Oct 2016;12(10):1007-1009. PMID: 27454438, PMCID: PMC5105591
86. Schussler E, Beasley MB, Maglione PJ. Lung Disease in Primary Antibody Deficiencies. *The journal of allergy and clinical immunology In practice*. Nov 2016;4(6):1039-1052. PMID: 27836055, PMCID: PMC5129846
87. Chinen J, Notarangelo LD, Shearer WT. Advances in clinical immunology in 2015. *J Allergy Clin Immunol*. Dec 2016;138(6):1531-1540. PMID: 27931534, PMCID: PMC5157931
88. Jackson SW, Scharping NE, Jacobs HM, Wang S, Chait A, Rawlings DJ. Cutting Edge: BAFF Overexpression Reduces Atherosclerosis via TACI-Dependent B Cell Activation. *J Immunol*. Dec 2016;197(12):4529-4534. PMID: 27837104, PMCID: PMC5147509
89. Lee YN, Frugoni F, Dobbs K, et al. Characterization of T and B cell repertoire diversity in patients with RAG deficiency. *Science immunology*. Dec 2016;1(6). PMID: 28783691, PMCID: PMC5586490
90. Punwani D, Zhang Y, Yu J, et al. Multisystem Anomalies in Severe Combined Immunodeficiency with Mutant BCL11B. *N Engl J Med*. Dec 2016;375(22):2165-2176. PMID: 27959755, PMCID: PMC5215776
91. Dietz AC, Duncan CN, Alter BP, et al. The Second Pediatric Blood and Marrow Transplant Consortium International Consensus Conference on Late Effects after Pediatric Hematopoietic Cell Transplantation: Defining the Unique Late Effects of Children Undergoing Hematopoietic Cell Transplantation for Immune Deficiencies, Inherited Marrow Failure Disorders, and Hemoglobinopathies. *Biol Blood Marrow Transplant*. Jan 2017;23(1):24-29. PMID: 27737772, PMCID: PMC5267609
92. Punwani D, Kawahara M, Yu J, et al. Lentivirus Mediated Correction of Artemis-Deficient Severe Combined Immunodeficiency. *Hum Gene Ther*. Jan 2017;28(1):112-124. PMID: 27611239, PMCID: PMC5278830
93. Wahlstrom J, Patel K, Eckhert E, et al. Transplacental maternal engraftment and posttransplantation graft-versus-host disease in children with severe combined immunodeficiency. *J Allergy Clin Immunol*. Feb 2017;139(2):628-633.e610. PMID: 27444177, PMCID: PMC5161721
94. Dorsey MJ, Dvorak CC, Cowan MJ, Puck JM. Treatment of infants identified as having severe combined immunodeficiency by means of newborn screening. *J Allergy Clin Immunol*. Mar 2017;139(3):733-742. PMID: 28270365, PMCID: PMC5385855
95. Heimall J, Puck J, Buckley R, et al. Current Knowledge and Priorities for Future Research in Late Effects after Hematopoietic Stem Cell Transplantation (HCT) for Severe Combined Immunodeficiency Patients: A Consensus Statement from the Second Pediatric Blood and Marrow Transplant Consortium International Conference on Late Effects after Pediatric HCT. *Biol Blood Marrow Transplant*. Mar 2017;23(3):379-387. PMID: 28068510, PMCID: PMC5659271
96. de la Morena MT, Leonard D, Torgerson TR, et al. Long-term outcomes of 176 patients with X-linked hyper-IgM syndrome treated with or without hematopoietic cell



- transplantation. *J Allergy Clin Immunol.* Apr 2017;139(4):1282-1292. PMID: 27697500, PMCID: PMC5374029
97. Hoenig M, Lagresle-Peyrou C, Pannicke U, et al. Reticular dysgenesis: international survey on clinical presentation, transplantation, and outcome. *Blood.* May 2017;129(21):2928-2938. PMID: 28331055, PMCID: PMC5445572
  98. Heimall J, Buckley RH, Puck J, et al. Recommendations for Screening and Management of Late Effects in Patients with Severe Combined Immunodeficiency after Allogeneic Hematopoietic Cell Transplantation: A Consensus Statement from the Second Pediatric Blood and Marrow Transplant Consortium International Conference on Late Effects after Pediatric HCT. *Biol Blood and Marrow Transplant.* Aug 2017;23(8):1229-1240. PMID: 28479164, PMCID: PMC6015789
  99. Dvorak CC, Puck J, Wahlstrom JT, Dorsey M, Melton A, Cowan MJ. Neurologic event-free survival demonstrates a benefit for SCID patients diagnosed by newborn screening. *Blood Advances.* Sep 2017;1(20):1694-1698. PMID: 29296816, PMCID: PMC5728344
  100. Heimall J, Logan BR, Cowan MJ, et al. Immune reconstitution and survival of 100 SCID patients post-hematopoietic cell transplant: a PIDTC natural history study. *Blood.* Dec 2017;130(25):2718-2727. PMID: 29021228, PMCID: PMC5746165
  101. Slack J, Albert MH, Balashov D, et al. Outcome of hematopoietic cell transplantation for DNA double-strand break repair disorders. *J of Allergy and Clin Immunol.* Jan 2018;141(1):322-328. PMID: 28392333. PMCID: PMC5632132
  102. Leiding JW, Okada S, Hagin D, et al. Hematopoietic stem cell transplantation in patients with gain-of-function signal transducer and activator of transcription 1 mutations. *J of Allergy and Clin Immunol.* Feb 2018;141(2): 704-717. PMID: 28601685. Pending PMCID
  103. Barzaghi F, Amaya Hernandez LC, Neven B, et al. Long-term follow-up of IPEX syndrome patients after different therapeutic strategies: An international multicenter retrospective study. *J of Allergy and Clin Immunol.* Mar 2018;141(3):1036-1049. PMID: 29241729, PMCID PMC6050203
  104. Kuo CY, Long JD, Campo-Fernandez B. Site-Specific Gene Editing of Human Hematopoietic Stem Cells for X-Linked Hyper-IgM Syndrome. *Cell Reports.* May 2018;23(9):2606-2616. PMID: 29847792. Pending PMCID
  105. Miggelbrink AM, Logan BR, Buckley RH, et al. B-cell differentiation and IL-21 response in IL2RG/JAK3 SCID patients after hematopoietic stem cell transplantation. *Blood.* Jun 2018;131(26):2967-2977. PMID 29728406, PMCID: PMC6024640
  106. Belderbos ME, Gennery AR, Dvorak CC, et al. Outcome of domino hematopoietic stem cell transplantation in human subjects: An international case series. *J of Allergy and Clin Immunol.* Jul 2018 Jul. PMID: 29981805. Pending PMCID
  107. Chinen J, Cowan M. Advances Series, Advances and Highlights In Primary Immunodeficiencies 2017. *J of Allergy and Clin Immunol.* Aug 2018. PMID 30170128. Pending PMCID

108. Haddad E, Logan BR, Griffith LM, et al. SCID genotype and 6-month post-transplant CD4 count predict survival and immune recovery: a PIDTC retrospective study. *Blood*. Aug 2018. PMID: 30154114. Pending PMCID
109. Dvorak CC, Haddad E, Buckley RH, et al. The genetic landscape of SCID in the US and Canada in the current era (2010-2018). Letter to the Editor. *J Allergy Clin Immunol*. Sep 2018. PMID: 30193840. Pending PMCID
110. Kohn DB, Hershfield MS, Puck JM, Aiuti A, Blincoe A, Gaspar HB, Notarangelo LD, Grunebaum E. Consensus approach for the management of severe combined immune deficiency caused by adenosine deaminase deficiency. *J Allergy Clin Immunol*. 2018 Sep 5. pii: S0091-6749(18)31268-5. doi: 10.1016/j.jaci.2018.08.024. [Epub ahead of print] PMID: 30194989
111. Mangurian C, Scalchunes C, Yoo J, Logan B, Henderson T, Iyengar S, Smith H, Cowan MJ. Psychosocial services for primary immunodeficiency disorder families during hematopoietic cell transplantation: A descriptive study. *Palliat Support Care*. 2018 Sep 18:1-6. doi: 10.1017/S1478951518000603. [Epub ahead of print] PubMed PMID: 30223912.
112. Yoo J, Halley M, Lown A, Yank V, Ort K, Cowan M, Dorsey M, Smith H, Iyengar S, Scalchunes C, Mangurian C. Supporting caregivers during hematopoietic cell transplantation for children with primary immunodeficiency disorders. *J Allergy Clin Immunol*. 2018 Oct 25. doi: 10.1016 /j.jaci.2018.10.017

## Selected Abstracts

### Primary Immune Deficiency Treatment Consortium

1. Dvorak CC, Cowan MJ, Logan BR, Griffith LM, Puck JM, Notarangelo LD, Kohn DB, Xiang Q, Eapen M, Shearer WT, Pulsipher MA, Buckley RH. The natural history of children with severe combined immunodeficiency disease (SCID): the first fifty patients of the primary immune deficiency treatment consortium (PIDTC) prospective study 6901. *Biol. Blood Marrow Transplant.* 2013; 19 Supplement 2: S161-S162. Abstract: 93. (Selected for oral presentation at “Best Pediatric Abstracts”, 2013 BMT Tandem Meetings).
2. Pai S-Y, Logan BR, O’Reilly RJ, et al Retrospective study of 240 patients with severe combined immunodeficiency transplanted from 2000-2009: a report from the Primary Immune Deficiency Treatment Consortium of North America. *Biol. Blood Marrow Transplant.* 2014; 20 Supplement 1: S24-S25. Abstract: 4. (Recipient of a “Best Abstracts Award” and oral presentation at the “Best Abstracts Plenary Session”, 2014 BMT Tandem Meetings).
3. Heimall J, Logan BR, Cowan MJ, et al. Early hematopoietic cell transplant outcomes of children with severe combined immunodeficiency (SCID): the first seventy-four patients of the Primary Immune Deficiency Treatment Consortium (PIDTC) prospective study 6901. *Biol. Blood Marrow Transplant.* 2015; 21 Supplement 1: S289-S291. Abstract: 413.
4. Heimall J, Logan BR, Cowan MJ, et al. Poor T cell reconstitution at 100 days after T cell-replete hematopoietic cell transplantation (HCT) for SCID is associated with later risk of death or need for 2<sup>nd</sup> transplant in the 6901 Prospective Study of the Primary Immune Deficiency Treatment Consortium (PIDTC). *Biol Blood Marrow Transplant* 2016; 22 Supplement 1: S101-S102. Abstract: 106. (Selected for oral presentation at “Best Pediatric Abstracts”, 2016 BMT Tandem Meetings).
5. Haddad E, O’Reilly R, PIDTC 6902 Study Investigators. Genotype, phenotype and T cell counts at one year predict survival and long term immune reconstitution after transplantation in severe combined immunodeficiency (SCID) – the Primary Immune Deficiency Treatment Consortium (PIDTC). Tandem American Society for Blood and Marrow Transplantation (ASBMT) and Center for International Blood and Marrow Transplant Research (CIBMTR) Annual Meetings, Orlando, FL, February 2017. (Selected for “Best Pediatric Abstracts”, 2017 BMT Tandem Meetings).
6. Leiding JW, Logan BR, Malech HW, Kang EM, et al. Resolution of CGD-related colitis after allogeneic hematopoietic stem cell transplantation in patients with Chronic Granulomatous Disease – early results from the 6903 study of the Primary Immune Deficiency Treatment Consortium (PIDTC). Tandem American Society for Blood and Marrow Transplantation (ASBMT) and Center for International Blood and Marrow Transplant Research (CIBMTR) Annual Meetings, Salt Lake City, UT, February 2018. (Selected for oral presentation, 2018 BMT Tandem Meetings).
7. Leiding JW, Torgerson TR, PIDTC 6906 PIRD Investigators. Hematopoietic stem cell transplantation in patients with primary immune regulatory disorders: a Primary Immune

- Deficiency Treatment Consortium (PIDTC) and Inborn Errors Working Party (IEWP) study. Clinical Immunology Society Annual Meeting, Toronto, Canada, April 2018.
8. Pai SY, Brazauskas R, Burroughs L, PIDTC 6904 WAS Study Investigators. Outcome of hematopoietic cell transplantation for Wiskott-Aldrich syndrome is excellent in all donor types in the modern era: a Primary Immune Deficiency Treatment Consortium (PIDTC) study. Clinical Immunology Society Annual Meeting, Toronto, Canada, April 2018.
  9. Kuo CY, Puck J, Zin Z, Logan B, Kohn DB, PIDTC 6901 and 6902 SCID Study Investigators. Adenosine deaminase (ADA)-deficient severe combined immune deficiency (SCID): the Primary Immune Deficiency Treatment Consortium (PIDTC) analysis. Clinical Immunology Society Annual Meeting, Toronto, Canada, April 2018.
  10. Berthe C, Bourgonnais S, Le Deist F, Decaluwe H, PIDTC 6901 and 6902 SCID Study Investigators. Aberrant T cell activation and exhaustion develops in poorly reconstituted SCID survivors after transplant and correlates with the absence of conditioning regimen: a Primary Immune Deficiency Treatment Consortium (PIDTC) study. Clinical Immunology Society Annual Meeting, Toronto, Canada, April 2018. (Selected for oral presentation).
  11. Heimall J, Dvorak C, PIDTC 6901 SCID Study Investigators. Diagnosis and pre-HCT management of SCID patients in the era of newborn screening: a survey of PIDTC center practices. Clinical Immunology Society Annual Meeting, Toronto, Canada, April 2018. (Late-Breaking Abstract, selected for oral presentation).
  12. Falcone EL, Kang E, PIDTC 6903 Study Investigators. Characterization of the intestinal microbiome in patients with chronic granulomatous disease after allogeneic hematopoietic stem cell transplantation: early results of the 6903 study of the Primary Immune Deficiency Treatment Consortium (PIDTC). ESID Biennial Meeting, Lisbon, Portugal, October 24-27, 2018.
  13. Haddad E, PIDTC 6902 Study Investigators. Genotype and CD4+ at six months predict survival and immune reconstitution after transplantation for SCID: a retrospective multicenter study of 662 patients from the PIDTC. ESID Biennial Meeting, Lisbon, Portugal, October 24-27, 2018.

## **Selected Chapters, Online and Other Publications**

### **Primary Immune Deficiency Treatment Consortium**

1. *Immune Deficiency Foundation Patient and Family Handbook for Primary Immunodeficiency Diseases, 5<sup>th</sup> Edition*. 2015.  
<https://primaryimmune.org/sites/default/files/publications/IDF-Patient-Family-Handbook-5th-Edition-2015-Reprint.pdf>