

## Immunodeficiency

Abolhassani, H., Sadaghiani, M. S., Aghamohammadi, A., Ochs, H. D., & Rezaei, N. (2012). Home-based subcutaneous immunoglobulin versus hospital-based intravenous immunoglobulin in treatment of primary antibody deficiencies: systematic review and meta analysis. *Journal of clinical immunology*, 32, 1180-1192. <https://doi.org/10.1007/s10875-012-9720-1>

Adrian YS Lee, P. E. G. (2014). Evaluating for immunodeficiency in children with recurrent infection. *Australian Journal for General Practitioners*, 43, 629-632.

<https://www.racgp.org.au/afp/2014/september/evaluating-for-immunodeficiency-in-children-with-I>

Agarwal, S., Ferreira, V. P., Cortes, C., Pangburn, M. K., Rice, P. A., & Ram, S. (2010). An evaluation of the role of properdin in alternative pathway activation on *Neisseria meningitidis* and *Neisseria gonorrhoeae*. *J Immunol*, 185(1), 507-516. <https://doi.org/10.4049/jimmunol.0903598>

Åhlin, A., Fugeläng, J., de Boer, M., Ringden, O., Fasth, A., & Winiarski, J. (2013). Chronic granulomatous disease-haematopoietic stem cell transplantation versus conventional treatment. *Acta Paediatr*, 102(11), 1087-1094. <https://doi.org/10.1111/apa.12384>

Akalu, Y. T., & Bogunovic, D. (2024). Inborn errors of immunity: an expanding universe of disease and genetic architecture. *Nature Reviews Genetics*, 25(3), 184-195. <https://doi.org/10.1038/s41576-023-00656-z>

Al-Herz, W., Bousfiha, A., Casanova, J.-L., Chatila, T., Conley, M. E., Cunningham-Rundles, C., Etzioni, A., Franco, J. L., Gaspar, H. B., Holland, S. M., Klein, C., Nonoyama, S., Ochs, H. D., Oksenhendler, E., Picard, C., Puck, J. M., Sullivan, K., & Tang, M. L. K. (2014). Primary Immunodeficiency Diseases: An Update on the Classification from the International Union of Immunological Societies Expert Committee for Primary Immunodeficiency [Classification]. *Frontiers in Immunology*, Volume 5 - 2014. <https://doi.org/10.3389/fimmu.2014.00162>

Ameratunga, R., Ahn, Y., Steele, R., & Woon, S. T. (2019). Transient hypogammaglobulinaemia of infancy: many patients recover in adolescence and adulthood. *Clinical and Experimental Immunology*, 198(2), 224-232. <https://doi.org/10.1111/cei.13345>

---

## ASCIA Immunodeficiency References

---

- Ameratunga, R., Woon, S.-T., Brewerton, M., Koopmans, W., Jordan, A., Brothers, S., & Singh, R. (2011). Primary immune deficiency disorders in the South Pacific: the clinical utility of a customized genetic testing program in New Zealand. *Annals of the New York Academy of Sciences*, 1238(1), 53-64. <https://doi.org/10.1111/j.1749-6632.2011.06238.x>
- Ameratunga, R., Woon, S.-T., Gillis, D., Koopmans, W., & Steele, R. (2013). New diagnostic criteria for common variable immune deficiency (CVID), which may assist with decisions to treat with intravenous or subcutaneous immunoglobulin. *Clinical and Experimental Immunology*, 174(2), 203-211. <https://doi.org/10.1111/cei.12178>
- Anderson, J. T., Cowan, J., Condino-Neto, A., Levy, D., & Prusty, S. (2022). Health-related quality of life in primary immunodeficiencies: Impact of delayed diagnosis and treatment burden. *Clinical Immunology*, 236, 108931. <https://doi.org/10.1016/j.clim.2022.108931>
- APIIEG. (2009). Recommendations for the use of Immunoglobulin Replacement Therapy in Immune Deficiency. . [http://www.korektorzdrowia.pl/wp-content/uploads/apiieg\\_2009.pdf](http://www.korektorzdrowia.pl/wp-content/uploads/apiieg_2009.pdf)
- Aranda, C. S., Gouveia-Pereira, M. P., da Silva, C. J. M., Rizzo, M. C. F. V., Ishizuka, E., de Oliveira, E. B., & Condino-Neto, A. (2024). Severe combined immunodeficiency diagnosis and genetic defects. *Immunological Reviews*, 322(1), 138-147. <https://doi.org/10.1111/imr.13310>
- Arkwright, P. D., & Gennery, A. R. (2011). Ten warning signs of primary immunodeficiency: a new paradigm is needed for the 21st century. *Annals of the New York Academy of Sciences*, 1238(1), 7-14. <https://doi.org/10.1111/j.1749-6632.2011.06206.x>
- ASCIA-TAPID. (2019). Consensus Guideline: Diagnosis, management and transplantation of severe combined immunodeficiency in Australia and New Zealand. .  
<https://www.allergy.org.au/hp/papers/ascia-guidelines-scid-tapid>
- Australian Technical Advisory Group on Immunisation (ATAGI). Australian Immunisation Handbook, Australian Government Department of Health and Aged Care, Canberra, 2022,  
<https://immunisationhandbook.health.gov.au>
- Ballow, M., Sánchez-Ramón, S., & Walter, J. E. (2022). Secondary Immune Deficiency and Primary Immune Deficiency Crossovers: Hematological Malignancies and Autoimmune Diseases. *Front Immunol*, 13, 928062. <https://doi.org/10.3389/fimmu.2022.928062>

Berger, M. (2011). Incidence of Infection is Inversely Related to Steady-State (Trough) Serum IgG Level in Studies of Subcutaneous IgG in PIDD. *Journal of clinical immunology*, 31(5), 924-926.  
<https://doi.org/10.1007/s10875-011-9546-2>

Bezrodnik, L., Gómez Raccio, A., Belardinelli, G., Regairaz, L., Díaz Ballve, D., Seminario, G., Moreira, I., Riganti, C., Cantisano, C., Díaz, H., & Di Giovanni, D. (2013). Comparative Study of Subcutaneous Versus Intravenous IgG Replacement Therapy in Pediatric Patients with Primary Immunodeficiency Diseases: A Multicenter Study in Argentina. *Journal of clinical immunology*, 33(7), 1216-1222. <https://doi.org/10.1007/s10875-013-9916-z>

Bonilla, F. A., Barlan, I., Chapel, H., Costa-Carvalho, B. T., Cunningham-Rundles, C., de la Morena, M. T., Espinosa-Rosales, F. J., Hammarström, L., Nonoyama, S., Quinti, I., Routes, J. M., Tang, M. L. K., & Warnatz, K. (2016). International Consensus Document (ICON): Common Variable Immunodeficiency Disorders. *The Journal of Allergy and Clinical Immunology: In Practice*, 4(1), 38-59. <https://doi.org/10.1016/j.jaip.2015.07.025>

Bonilla, F. A., Khan, D. A., Ballas, Z. K., Chinen, J., Frank, M. M., Hsu, J. T., Keller, M., Kobrynski, L. J., Komarow, H. D., Mazer, B., Nelson, R. P., Jr., Orange, J. S., Routes, J. M., Shearer, W. T., Sorensen, R. U., Verbsky, J. W., Bernstein, D. I., Blessing-Moore, J., Lang, D.,...Verbsky, J. W. (2015). Practice parameter for the diagnosis and management of primary immunodeficiency. *Journal of Allergy and Clinical Immunology*, 136(5), 1186-1205.e1178.  
<https://doi.org/10.1016/j.jaci.2015.04.049>

Bousfiha, A., Moundir, A., Tangye, S. G., Picard, C., Jeddane, L., Al-Herz, W., Rundles, C. C., Franco, J. L., Holland, S. M., Klein, C., Morio, T., Oksenhendler, E., Puel, A., Puck, J., Seppänen, M. R. J., Somech, R., Su, H. C., Sullivan, K. E., Torgerson, T. R., & Meyts, I. (2022). The 2022 Update of IUIS Phenotypical Classification for Human Inborn Errors of Immunity. *Journal of clinical immunology*, 42(7), 1508-1520. <https://doi.org/10.1007/s10875-022-01352-z>

Bousfiha, A. A., Jeddane, L., Moundir, A., Poli, M. C., Aksentijevich, I., Cunningham-Rundles, C., Hambleton, S., Klein, C., Morio, T., Picard, C., Puel, A., Rezaei, N., Seppänen, M. R. J., Somech, R., Su, H. C., Sullivan, K. E., Torgerson, T. R., Tangye, S. G., & Meyts, I. (2025). The 2024 update of IUIS phenotypic classification of human inborn errors of immunity. *Journal of Human Immunity*, 1(1), e20250002. <https://doi.org/10.70962/jhi.20250002>

## ASClA Immunodeficiency References

---

- Bucciol, G., Delafontaine, S., Meyts, I., & Poli, C. (2024). Inborn errors of immunity: A field without frontiers. *Immunological Reviews*, 322(1), 15-27. <https://doi.org/10.1111/imr.13297>
- Buchbinder, D., Nugent, D. J., & Fillipovich, A. H. (2014). Wiskott-Aldrich syndrome: diagnosis, current management, and emerging treatments. *Appl Clin Genet*, 7, 55-66.  
<https://doi.org/10.2147/tacg.S58444>
- Buckley, R. H. (2002). Primary cellular immunodeficiencies. *J Allergy Clin Immunol*, 109(5), 747-757.  
<https://doi.org/10.1067/mai.2002.123617>
- Cardenas-Morales, M., & Hernandez-Trujillo, V. P. (2022). Agammaglobulinemia: from X-linked to Autosomal Forms of Disease. *Clin Rev Allergy Immunol*, 63(1), 22-35.  
<https://doi.org/10.1007/s12016-021-08870-5>
- Casanova, J.-L., & Abel, L. (2007). Primary Immunodeficiencies: A Field in Its Infancy. *Science*, 317(5838), 617-619. <https://doi.org/doi:10.1126/science.1142963>
- Casanova, J.-L., & Abel, L. (2022). From rare disorders of immunity to common determinants of infection: Following the mechanistic thread. *Cell*, 185(17), 3086-3103.  
<https://doi.org/10.1016/j.cell.2022.07.004>
- Casanova, J.-L., & Anderson, M. S. (2023). Unlocking life-threatening COVID-19 through two types of inborn errors of type I IFNs. *The Journal of Clinical Investigation*, 133(3).  
<https://doi.org/10.1172/JCI166283>
- Casanova, J.-L., Conley, M. E., Seligman, S. J., Abel, L., & Notarangelo, L. D. (2014). Guidelines for genetic studies in single patients: lessons from primary immunodeficiencies. *Journal of Experimental Medicine*, 211(11), 2137-2149. <https://doi.org/10.1084/jem.20140520>
- Castagnoli, R., Delmonte, O. M., Calzoni, E., & Notarangelo, L. D. (2019). Hematopoietic Stem Cell Transplantation in Primary Immunodeficiency Diseases: Current Status and Future Perspectives. *Front Pediatr*, 7, 295. <https://doi.org/10.3389/fped.2019.00295>
- Cavazzana-Calvo, M., Hacein-Bey, S., Basile, G. d. S., Gross, F., Yvon, E., Nusbaum, P., Selz, F., Hue, C., Certain, S., Casanova, J.-L., Bousso, P., Deist, F. L., & Fischer, A. (2000). Gene therapy of human severe combined immunodeficiency (SCID)-X1 disease. *Science*, 288(5466), 669-672. <https://doi.org/doi:10.1126/science.288.5466.669>
- Chan, A., Scalchunes, C., Boyle, M., & Puck, J. M. (2011). Early vs. delayed diagnosis of severe combined immunodeficiency: a family perspective survey. *Clin Immunol*, 138(1), 3-8.  
<https://doi.org/10.1016/j.clim.2010.09.010>

---

## ASClA Immunodeficiency References

---

Chapel, H., Prevot, J., Gaspar, H. B., Español, T., Bonilla, F. A., Solis, L., & Drabwell, J. (2014). Primary Immune Deficiencies – Principles of Care [Hypothesis and Theory]. *Frontiers in Immunology*, Volume 5 - 2014. <https://doi.org/10.3389/fimmu.2014.00627>

Chen, R., Lukianova, E., van der Loeff, I. S., Spegarova, J. S., Willet, J. D. P., James, K. D., Ryder, E. J., Griffin, H., IJspeert, H., Gajbhiye, A., Lamoliatte, F., Marin-Rubio, J. L., Woodbine, L., Lemos, H., Swan, D. J., Pintar, V., Sayes, K., Ruiz-Morales, E. R., Eastham, S.,...Hambleton, S. (2024). NUDCD3 deficiency disrupts V(D)J recombination to cause SCID and Omenn syndrome. *Science Immunology*, 9(95), eade5705.

<https://doi.org/doi:10.1126/sciimmunol.ade5705>

Chinnici, A., Beneforti, L., Pegoraro, F., Trambusti, I., Tondo, A., Favre, C., Coniglio, M. L., & Sieni, E. (2023). Approaching hemophagocytic lymphohistiocytosis [Review]. *Frontiers in Immunology*, Volume 14 - 2023. <https://doi.org/10.3389/fimmu.2023.1210041>

Chong-Neto, H. J., Radwan, N., Condino-Neto, A., Rosário Filho, N. A., Ortega-Martell, J. A., & El-Sayed, Z. A. (2024). Newborn screening for inborn errors of immunity: The status worldwide. *World Allergy Organ J*, 17(6), 100920. <https://doi.org/10.1016/j.waojou.2024.100920>

Cinetto, F., Neri, R., Vianello, F., Visentin, A., Barilà, G., Gianese, S., Lanciarotta, A., Milito, C., Rattazzi, M., Piazza, F., Trentin, L., Zambello, R., Agostini, C., & Scarpa, R. (2021). Subcutaneous immunoglobulins replacement therapy in secondary antibody deficiencies: Real life evidence as compared to primary antibody deficiencies. *PLoS One*, 16(3), e0247717. <https://doi.org/10.1371/journal.pone.0247717>

Cole, T., McLean-Tooke, A., Loh, R., Quinn, P., Peake, J., Sinclair, J., & Smart, J. . (2018). P48: ASCIA TRANSPLANTATION AND PRIMARY IMMUNODEFICIENCY (TAPID) PROJECT. *Internal Medicine Journal*, 48(S6), 18-19. [https://doi.org/10.1111/imj.48\\_14077](https://doi.org/10.1111/imj.48_14077)

Cole, T., Pearce, M. S., Cant, A. J., Cale, C. M., Goldblatt, D., & Gennery, A. R. (2013). Clinical outcome in children with chronic granulomatous disease managed conservatively or with hematopoietic stem cell transplantation. *Journal of Allergy and Clinical Immunology*, 132(5), 1150-1155. <https://doi.org/10.1016/j.jaci.2013.05.311> ng the Lives of People With Primary Immunodeficiencies (PI) With Early Testing and Diagnosis. *Front Immunol*, 9, 1439. <https://doi.org/10.3389/fimmu.2018.01439>

---

## ASClA Immunodeficiency References

---

- Cooper, M. A., Pommering, T. L., & Korányi, K. (2003). Primary immunodeficiencies. *Am Fam Physician*, 68(10), 2001-2008.
- Coss, S. L., Zhou, D., Chua, G. T., Aziz, R. A., Hoffman, R. P., Wu, Y. L., Ardoine, S. P., Atkinson, J. P., & Yu, C.-Y. (2023). The complement system and human autoimmune diseases. *Journal of Autoimmunity*, 137, 102979. <https://doi.org/10.1016/j.jaut.2022.102979>
- Cunningham-Rundles, C. (2012). The many faces of common variable immunodeficiency. *Hematology Am Soc Hematol Educ Program*, 2012, 301-305. <https://doi.org/10.1182/asheducation-2012.1.301>
- Dahl, C., Petersen, I., Ilkjær, F. V., Westh, L., Katzenstein, T. L., Hansen, A.-B. E., Nielsen, T. L., Larsen, C. S., Johansen, I. S., & Rasmussen, L. D. (2023). Missed Opportunities to Diagnose Common Variable Immunodeficiency: a Population-Based Case–Control Study Identifying Indicator Diseases for Common Variable Immunodeficiency. *Journal of clinical immunology*, 43(8), 2104-2114. <https://doi.org/10.1007/s10875-023-01590-9>
- De Decker, H. P., & Lawrenson, J. B. (2001). The 22q11.2 deletion: From diversity to a single gene theory. *Genetics in Medicine*, 3(1), 2-5. <https://doi.org/10.1097/00125817-200101000-00002>
- Dvorak, C. C., Haddad, E., Heimall, J., Dunn, E., Buckley, R. H., Kohn, D. B., Cowan, M. J., Pai, S. Y., Griffith, L. M., Cuvelier, G. D. E., Eissa, H., Shah, A. J., O'Reilly, R. J., Pulsipher, M. A., Wright, N. A. M., Abraham, R. S., Satter, L. F., Notarangelo, L. D., & Puck, J. M. (2023). The diagnosis of severe combined immunodeficiency (SCID): The Primary Immune Deficiency Treatment Consortium (PIDTC) 2022 Definitions. *J Allergy Clin Immunol*, 151(2), 539-546. <https://doi.org/10.1016/j.jaci.2022.10.022>
- EBMT/ESID. (2017). EBMT/ESID Guidelines for haematopoietic stem cell transplantation (HSCT) for primary immunodeficiencies HSCT Guidelines. <https://www.ebmt.org/ebmt/documents/esid-ebmt-hsct-guidelines-2017>
- Fernández-Cruz, E., Alecsandru, D., & Ramón, S. S. (2009). Mechanisms of action of immune globulin. *Clinical and Experimental Immunology*, 157(Supplement\_1), 1-2. <https://doi.org/10.1111/j.1365-2249.2009.03955.x>
- Firatoglu, H., Aytekin, C., Dogu, F., Bal, S. K., Haskologlu, S., Boztug, K., & Ikinciogullari, A. (2025). Evaluation of Patients with Combined Immunodeficiency: A Single Center Experience. *Iran J Immunol*, 22(1), 89-99. <https://doi.org/10.22034/iji.2025.103499.2844>

---

## ASClA Immunodeficiency References

---

- Fischer, A. (2000). Severe combined immunodeficiencies (SCID). *Clin Exp Immunol*, 122(2), 143-149.  
<https://doi.org/10.1046/j.1365-2249.2000.01359.x>
- Fischer, A., & Rausell, A. (2018). What do primary immunodeficiencies tell us about the essentiality/redundancy of immune responses? *Seminars in Immunology*, 36, 13-16.  
<https://doi.org/10.1016/j.smim.2017.12.001>
- Fomin, A. B., Pastorino, A. C., Kim, C. A., Pereira, C. A., Carneiro-Sampaio, M., & Abe-Jacob, C. M. (2010). DiGeorge Syndrome: a not so rare disease. *Clinics (Sao Paulo)*, 65(9), 865-869.  
<https://doi.org/10.1590/s1807-59322010000900009>
- Fornes, O., Jia, A., Kuehn, H. S., Min, Q., Pannicke, U., Schleussner, N., Thouenon, R., Yu, Z., de los Angeles Astbury, M., Biggs, C. M., Galicchio, M., Garcia-Campos, J. A., Gismondi, S., Gonzalez Villarreal, G., Hildebrand, K. J., Höning, M., Hou, J., Moshous, D., Pittaluga, S.,...Wang, J.-Y. (2023). A multimorphic mutation in IRF4 causes human autosomal dominant combined immunodeficiency. *Science Immunology*, 8(79), eade7953.  
<https://doi.org/doi:10.1126/sciimmunol.adc7953>
- Frank, M. M. (2010). Complement disorders and hereditary angioedema. *J Allergy Clin Immunol*, 125(2 Suppl 2), S262-271. <https://doi.org/10.1016/j.jaci.2009.10.063>
- Gardulf, A. (2007). Immunoglobulin treatment for primary antibody deficiencies: advantages of the subcutaneous route. *BioDrugs*, 21(2), 105-116. <https://doi.org/10.2165/00063030-200721020-00005>
- Gaspar, J., Gerritsen, B., & Jones, A. (1998). Immunoglobulin replacement treatment by rapid subcutaneous infusion. *Archives of Disease in Childhood*, 79(1), 48.  
<https://doi.org/10.1136/adc.79.1.48>
- George, M. R. (2014). Hemophagocytic lymphohistiocytosis: review of etiologies and management. *J Blood Med*, 5, 69-86. <https://doi.org/10.2147/jbm.S46255>
- Gray, P. E. A., Namasivayam, M., & Ziegler, J. B. (2012). Recurrent infection in children: When and how to investigate for primary immunodeficiency? *Journal of Paediatrics and Child Health*, 48(3), 202-209. <https://doi.org/10.1111/j.1440-1754.2011.02080.x>
- Grosserichter-Wagener, C., Franco-Gallego, A., Ahmadi, F., Moncada-Vélez, M., Dalm, V. A. S. H., Rojas, J. L., Orrego, J. C., Correa Vargas, N., Hammarström, L., Schreurs, M. W. J., Dik, W. A., van Hagen, P. M., Boon, L., van Dongen, J. J. M., van der Burg, M., Pan-Hammarström, Q., Franco, J. L., & van Zelm, M. C. (2020). Defective formation of IgA memory B cells, Th1 and

- Th17 cells in symptomatic patients with selective IgA deficiency. Clinical & Translational Immunology, 9(5), e1130. <https://doi.org/10.1002/cti2.1130>
- Gruber, C., & Bogunovic, D. (2020). Incomplete penetrance in primary immunodeficiency: a skeleton in the closet. Hum Genet, 139(6-7), 745-757. <https://doi.org/10.1007/s00439-020-02131-9>
- Guérin, A., Kerner, G., Marr, N., Markle, J. G., Fenollar, F., Wong, N., Boughorbel, S., Avery, D. T., Ma, C. S., Bougarn, S., Bouaziz, M., Béziat, V., Della Mina, E., Oleaga-Quintas, C., Lazarov, T., Worley, L., Nguyen, T., Patin, E., Deswarte, C.,...Casanova, J.-L. (2018). IRF4 haploinsufficiency in a family with Whipple's disease. eLife, 7, e32340. <https://doi.org/10.7554/eLife.32340>
- Guo, Y., Tian, X., Wang, X., & Xiao, Z. (2018). Adverse Effects of Immunoglobulin Therapy. Front Immunol, 9, 1299. <https://doi.org/10.3389/fimmu.2018.01299>
- Heather, N., de Hora, M., Brothers, S., Grainger, P., Knoll, D., & Webster, D. (2022). Introducing Newborn Screening for Severe Combined Immunodeficiency-The New Zealand Experience. Int J Neonatal Screen, 8(2). <https://doi.org/10.3390/ijns8020033>
- Heimall, J., & Cowan, M. J. (2017). Long term outcomes of severe combined immunodeficiency: therapy implications. Expert Rev Clin Immunol, 13(11), 1029-1040. <https://doi.org/10.1080/1744666x.2017.1381558>
- Henter, J. I., Sieni, E., Eriksson, J., Bergsten, E., Hed Myrberg, I., Canna, S. W., Coniglio, M. L., Cron, R. Q., Kernan, K. F., Kumar, A. R., Lehmberg, K., Minoia, F., Naqvi, A., Ravelli, A., Tang, Y. M., Bottai, M., Bryceson, Y. T., Horne, A., & Jordan, M. B. (2024). Diagnostic guidelines for familial hemophagocytic lymphohistiocytosis revisited. Blood, 144(22), 2308-2318. <https://doi.org/10.1182/blood.2024025077>
- Holland, S. M. (2013). Chronic granulomatous disease. Hematol Oncol Clin North Am, 27(1), 89-99, viii. <https://doi.org/10.1016/j.hoc.2012.11.002>
- Houghton, B. C., & Booth, C. (2021). Gene Therapy for Primary Immunodeficiency. Hemosphere, 5(1), e509. <https://doi.org/10.1097/hs9.0000000000000509>
- Inwald, D. P., Peters, M. J., Walshe, D., Jones, A., Davies, E. G., & Klein, N. J. (2000). Absence of platelet CD40L identifies patients with X-linked hyper IgM syndrome. Clin Exp Immunol, 120(3), 499-502. <https://doi.org/10.1046/j.1365-2249.2000.01235.x>
- Israel, L., Wang, Y., Bulek, K., Della Mina, E., Zhang, Z., Pedergnana, V., Chrabieh, M., Lemmens, N. A., Sancho-Shimizu, V., Descatoire, M., Lasseau, T., Israelsson, E., Lorenzo, L., Yun, L.,

- Belkadi, A., Moran, A., Weisman, L. E., Vandenesch, F., Batteux, F.,...Puel, A. (2017). Human Adaptive Immunity Rescues an Inborn Error of Innate Immunity. *Cell*, 168(5), 789-800.e710. <https://doi.org/10.1016/j.cell.2017.01.039>
- Jin, Y., Mazza, C., Christie, J. R., Giliani, S., Fiorini, M., Mella, P., Gandellini, F., Stewart, D. M., Zhu, Q., Nelson, D. L., Notarangelo, L. D., & Ochs, H. D. (2004). Mutations of the Wiskott-Aldrich Syndrome Protein (WASP): hotspots, effect on transcription, and translation and phenotype/genotype correlation. *Blood*, 104(13), 4010-4019. <https://doi.org/10.1182/blood-2003-05-1592>
- Johnson, M. B., Ogishi, M., Domingo-Vila, C., De Franco, E., Wakeling, M. N., Imane, Z., Resnick, B., Williams, E., Galão, R. P., Caswell, R., Russ-Silsby, J., Seeleuthner, Y., Rinchai, D., Fagniez, I., Benson, B., Dufort, M. J., Speake, C., Smithmyer, M. E., Hudson, M.,...Oram, R. A. (2024). Human inherited PD-L1 deficiency is clinically and immunologically less severe than PD-1 deficiency. *J Exp Med*, 221(6). <https://doi.org/10.1084/jem.20231704>
- Jolles, S., Orange, J. S., Gardulf, A., Stein, M. R., Shapiro, R., Borte, M., & Berger, M. (2015). Current treatment options with immunoglobulin G for the individualization of care in patients with primary immunodeficiency disease. *Clinical and Experimental Immunology*, 179(2), 146-160. <https://doi.org/10.1111/cei.12485>
- Jolles, S., Sewell, W. A., & Misbah, S. A. (2005). Clinical uses of intravenous immunoglobulin. *Clin Exp Immunol*, 142(1), 1-11. <https://doi.org/10.1111/j.1365-2249.2005.02834.x>
- Jolles, S., & Sleasman, J. W. (2011). Subcutaneous immunoglobulin replacement therapy with Hizentra®, the first 20% SCIG preparation: a Practical approach. *Advances in Therapy*, 28(7), 521-533. <https://doi.org/10.1007/s12325-011-0036-y>
- Justiz-Vaillant, A. A., Hoyte, T., Davis, N., Deonarinesingh, C., De Silva, A., Dhanpaul, D., Dookhoo, C., Doorpat, J., Dopson, A., Durgaparsad, J., Palmer, C., Asin-Milan, O., Williams-Persad, A. F.-A., & Arozarena-Fundora, R. (2023). A Systematic Review of the Clinical Diagnosis of Transient Hypogammaglobulinemia of Infancy. *Children*, 10(8), 1358. <https://www.mdpi.com/2227-9067/10/8/1358>
- Kaçar, A. G., & Celkan, T. T. (2022). Hemophagocytic Lymphohistiocytosis. *Balkan Med J*, 39(5), 309-317. <https://doi.org/10.4274/balkanmedj.galenos.2022.2022-4-83>
- Kang, E. M., Marciano, B. E., DeRavin, S., Zaremba, K. A., Holland, S. M., & Malech, H. L. (2011). Chronic granulomatous disease: Overview and hematopoietic stem cell transplantation. *Journal*

- of Allergy and Clinical Immunology, 127(6), 1319-1326.  
<https://doi.org/10.1016/j.jaci.2011.03.028>
- Karhan, A. N., Esenboğa, S., Gümüş, E., Karaatmaca, B., Cagdas, D., Demir, H., Saltik Temizel İ, N., Özen, H., Yüce, A., & Tezcan, İ. (2022). Nutritional status of children with primary immunodeficiency: A single center experience. Pediatr Int, 64(1), e14996.  
<https://doi.org/10.1111/ped.14996>
- Kavanagh, D., Barratt, J., Schubart, A., Webb, N. J. A., Meier, M., & Fakhouri, F. (2025). Factor B as a therapeutic target for the treatment of complement-mediated diseases. Front Immunol, 16, 1537974. <https://doi.org/10.3389/fimmu.2025.1537974>
- Kirkpatrick, P., & Riminton, S. (2007). Primary Immunodeficiency Diseases in Australia and New Zealand. Journal of clinical immunology, 27(5), 517-524. <https://doi.org/10.1007/s10875-007-9105-z>
- Kobrynski, L. (2012). Subcutaneous immunoglobulin therapy: a new option for patients with primary immunodeficiency diseases. Biologics, 6, 277-287. <https://doi.org/10.2147/btt.S25188>
- Kohn, D. B., Hershfield, M. S., Puck, J. M., Aiuti, A., Blincoe, A., Gaspar, H. B., Notarangelo, L. D., & Grunebaum, E. (2019). Consensus approach for the management of severe combined immune deficiency caused by adenosine deaminase deficiency. Journal of Allergy and Clinical Immunology, 143(3), 852-863. <https://doi.org/10.1016/j.jaci.2018.08.024>
- Kostel Bal, S., Giuliani, S., Block, J., Repiscak, P., Hafemeister, C., Shahin, T., Kasap, N., Ransmayer, B., Miao, Y., van de Wetering, C., Frohne, A., Jimenez Heredia, R., Schuster, M., Zoghi, S., Hertlein, V., Thian, M., Bykov, A., Babayeva, R., Bilgic Eltan, S.,...Boztug, K. (2023). Biallelic NFATC1 mutations cause an inborn error of immunity with impaired CD8+ T-cell function and perturbed glycolysis. Blood, 142(9), 827-845. <https://doi.org/10.1182/blood.2022018303>
- Kuo, C. Y., Garabedian, E., Puck, J., Cowan, M. J., Sullivan, K. E., Buckley, R. H., Cunningham-Rundles, C., Marsh, R., Candotti, F., & Kohn, D. B. (2020). Adenosine Deaminase (ADA)-Deficient Severe Combined Immune Deficiency (SCID) in the US Immunodeficiency Network (USIDNet) Registry. J Clin Immunol, 40(8), 1124-1131. <https://doi.org/10.1007/s10875-020-00857-9>
- Larson, R. S., & Butler, M. G. (1995). Use of fluorescence in situ hybridization (FISH) in the diagnosis of DiGeorge sequence and related diseases. Diagn Mol Pathol, 4(4), 274-278.  
<https://doi.org/10.1097/00019606-199512000-00008>

## ASCIA Immunodeficiency References

---

- Lee, T. K., Gereige, J. D., & Maglione, P. J. (2021). State-of-the-art diagnostic evaluation of common variable immunodeficiency. *Ann Allergy Asthma Immunol*, 127(1), 19-27.  
<https://doi.org/10.1016/j.anai.2021.03.005>
- Lehmberg, K., Moshous, D., & Booth, C. (2019). Haematopoietic Stem Cell Transplantation for Primary Haemophagocytic Lymphohistiocytosis. *Front Pediatr*, 7, 435.  
<https://doi.org/10.3389/fped.2019.00435>
- Leiding, J. W., & Forbes, L. R. (2019). Mechanism-Based Precision Therapy for the Treatment of Primary Immunodeficiency and Primary Immunodysregulatory Diseases. *The Journal of Allergy and Clinical Immunology: In Practice*, 7(3), 761-773. <https://doi.org/10.1016/j.jaip.2018.12.017>
- Li, Y., Yu, Z., Schenk, M., Lagovsky, I., Illig, D., Walz, C., Rohlf, M., Conca, R., Muise, A. M., Snapper, S. B., Uhlig, H. H., Garty, B. Z., Klein, C., & Kotlarz, D. (2023). Human MD2 deficiency—an inborn error of immunity with pleiotropic features. *J Allergy Clin Immunol*, 151(3), 791-796.e797.  
<https://doi.org/10.1016/j.jaci.2022.09.033>
- Li, Z., & Mahmood, I. (2024). Immunoglobulin therapies for primary immunodeficiency diseases (part 2): considerations for dosing strategies. *Immunotherapy*, 16(13), 895-905.  
<https://doi.org/10.1080/1750743x.2024.2382074>
- Lingman-Framme, J., & Fasth, A. (2013). Subcutaneous Immunoglobulin for Primary and Secondary Immunodeficiencies: an Evidence-Based Review. *Drugs*, 73(12), 1307-1319.  
<https://doi.org/10.1007/s40265-013-0094-3>
- Loh, R. K., Vale, S., & McLean-Tooke, A. (2013). Quantitative serum immunoglobulin tests. *Aust Fam Physician*, 42(4), 195-198.
- Lucas, C. L. (2024). Human genetic errors of immunity illuminate an adaptive arsenal model of rapid defenses. *Trends in Immunology*, 45(2), 113-126. <https://doi.org/10.1016/j.it.2023.12.006>
- Ma, C. S., & Tangye, S. G. (2019). Flow Cytometric-Based Analysis of Defects in Lymphocyte Differentiation and Function Due to Inborn Errors of Immunity [Review]. *Frontiers in Immunology*, Volume 10 - 2019. <https://doi.org/10.3389/fimmu.2019.02108>
- Mallick, R., Hahn, N., & Scalchunes, C. (2025). Immunoglobulin replacement therapy in patients with primary and secondary immunodeficiencies: impact of infusion method on immunoglobulin-specific perceptions of quality of life and treatment satisfaction. *Allergy, Asthma & Clinical Immunology*, 21(1), 2. <https://doi.org/10.1186/s13223-024-00939-y>

## ASClA Immunodeficiency References

---

- Marsh, R. A., Vaughn, G., Kim, M.-O., Li, D., Jodele, S., Joshi, S., Mehta, P. A., Davies, S. M., Jordan, M. B., Bleesing, J. J., & Filipovich, A. H. (2010). Reduced-intensity conditioning significantly improves survival of patients with hemophagocytic lymphohistiocytosis undergoing allogeneic hematopoietic cell transplantation. *Blood*, 116(26), 5824-5831. <https://doi.org/10.1182/blood-2010-04-282392>
- Marshall, J. S., Warrington, R., Watson, W., & Kim, H. L. (2018). An introduction to immunology and immunopathology. *Allergy, Asthma & Clinical Immunology*, 14(2), 49. <https://doi.org/10.1186/s13223-018-0278-1>
- McCusker, C., Upton, J., & Warrington, R. (2018). Primary immunodeficiency. *Allergy, Asthma & Clinical Immunology*, 14(2), 61. <https://doi.org/10.1186/s13223-018-0290-5>
- McCusker, C., & Warrington, R. (2011). Primary immunodeficiency. *Allergy, Asthma & Clinical Immunology*, 7(1), S11. <https://doi.org/10.1186/1710-1492-7-S1-S11>
- McLean-Tooke, A., Spickett, G. P., & Gennery, A. R. (2007). Immunodeficiency and Autoimmunity in 22q11.2 Deletion Syndrome. *Scandinavian Journal of Immunology*, 66(1), 1-7. <https://doi.org/https://doi.org/10.1111/j.1365-3083.2007.01949.x>
- McMurray, J. C., Schornack, B. J., Weskamp, A. L., Park, K. J., Pollock, J. D., Day, W. G., Brockshus, A. T., Beakes, D. E., Schwartz, D. J., Mikita, C. P., & Pittman, L. M. (2024). Immunodeficiency: Complement disorders. *Allergy Asthma Proc*, 45(5), 305-309. <https://doi.org/10.2500/aap.2024.45.240050>
- Mickey, D., Camacho, J. V., Khan, A., & Kaufman, D. (2024). Immunodeficiency: Quantitative and qualitative phagocytic cell defects. *Allergy Asthma Proc*, 45(5), 299-304. <https://doi.org/10.2500/aap.2024.45.240049>
- Mitchell, R., Nivison-Smith, I., Anazodo, A., Tiedemann, K., Shaw, P., Teague, L., Fraser, C., Carter, T., Tapp, H., Alvaro, F., & O'Brien, T. A. (2013). Outcomes of hematopoietic stem cell transplantation in primary immunodeficiency: a report from the Australian and New Zealand Children's Haematology Oncology Group and the Australasian Bone Marrow Transplant Recipient Registry. *Biology of Blood and Marrow Transplantation*, 19(3), 338-343. <https://doi.org/10.1016/j.bbmt.2012.11.619>
- Mizoguchi, Y., & Okada, S. (2021). Inborn errors of STAT1 immunity. *Current Opinion in Immunology*, 72, 59-64. <https://doi.org/10.1016/j.co.2021.02.009>

- Mustillo, P. J., Sullivan, K. E., Chinn, I. K., Notarangelo, L. D., Haddad, E., Davies, E. G., de la Morena, M. T., Hartog, N., Yu, J. E., Hernandez-Trujillo, V. P., Ip, W., Franco, J., Gambineri, E., Hickey, S. E., Varga, E., & Markert, M. L. (2023). Clinical Practice Guidelines for the Immunological Management of Chromosome 22q11.2 Deletion Syndrome and Other Defects in Thymic Development. *J Clin Immunol*, 43(2), 247-270. <https://doi.org/10.1007/s10875-022-01418-y>
- Neven, B., & Ferrua, F. (2020). Hematopoietic Stem Cell Transplantation for Combined Immunodeficiencies, on Behalf of IEWP-EBMT [Mini Review]. *Frontiers in Pediatrics*, Volume 7 - 2019. <https://doi.org/10.3389/fped.2019.00552>
- Nicolay, U., Haag, S., Eichmann, F., Herget, S., Spruck, D., & Gardulf, A. (2005). Measuring treatment satisfaction in patients with primary immunodeficiency diseases receiving lifelong immunoglobulin replacement therapy. *Quality of Life Research*, 14(7), 1683-1691. <https://doi.org/10.1007/s11136-005-1746-x>
- Notarangelo, L. D., Bacchetta, R., Casanova, J.-L., & Su, H. C. (2020). Human inborn errors of immunity: An expanding universe. *Science Immunology*, 5(49), eabb1662. <https://doi.org/10.1126/scimmunol.abb1662>
- Ochs, H. D., Smith, C. E., & Puck, J. M. (2013). Primary Immunodeficiency Diseases: A Molecular and Genetic Approach. Oxford University Press. <https://doi.org/10.1093/med/9780195389838.001.0001>
- Ochs, H. D., Gupta, S., Kiessling, P., Nicolay, U., & Berger, M. (2006). Safety and Efficacy of Self-Administered Subcutaneous Immunoglobulin in Patients with Primary Immunodeficiency Diseases. *Journal of clinical immunology*, 26(3), 265-273. <https://doi.org/10.1007/s10875-006-9021-7>
- Oikonomopoulou, Z., Shulman, S., Mets, M., & Katz, B. (2022). Chronic Granulomatous Disease: an Updated Experience, with Emphasis on Newly Recognized Features. *J Clin Immunol*, 42(7), 1411-1419. <https://doi.org/10.1007/s10875-022-01294-6>
- O'Keefe, A. W., Halbrich, M., Ben-Shoshan, M., & McCusker, C. (2016). Primary immunodeficiency for the primary care provider. *Paediatrics & Child Health*, 21(2), e10-e14. <https://doi.org/10.1093/pch/21.2.e10>
- Oliveira, J. B., & Fleisher, T. A. (2010). Laboratory evaluation of primary immunodeficiencies. *J Allergy Clin Immunol*, 125(2 Suppl 2), S297-305. <https://doi.org/10.1016/j.jaci.2009.08.043>

- Ozdemir, O. (2021). Primary immunodeficiency diseases in the newborn. *North Clin Istanb*, 8(4), 405-413. <https://doi.org/10.14744/nci.2020.43420>
- Palmeira, P., Quinello, C., Silveira-Lessa, A. L., Zago, C. A., & Carneiro-Sampaio, M. (2012). IgG placental transfer in healthy and pathological pregnancies. *Clin Dev Immunol*, 2012, 985646. <https://doi.org/10.1155/2012/985646>
- Panch, S. R., Szymanski, J., Savani, B. N., & Stroncek, D. F. (2017). Sources of Hematopoietic Stem and Progenitor Cells and Methods to Optimize Yields for Clinical Cell Therapy. *Biology of Blood and Marrow Transplantation*, 23(8), 1241-1249. <https://doi.org/10.1016/j.bbmt.2017.05.003>
- Perez, E., Bonilla, F. A., Orange, J. S., & Ballow, M. (2017). Specific Antibody Deficiency: Controversies in Diagnosis and Management. *Front Immunol*, 8, 586. <https://doi.org/10.3389/fimmu.2017.00586>
- Picard, C., Bobby Gaspar, H., Al-Herz, W., Bousfiha, A., Casanova, J.-L., Chatila, T., Crow, Y. J., Cunningham-Rundles, C., Etzioni, A., Franco, J. L., Holland, S. M., Klein, C., Morio, T., Ochs, H. D., Oksenhendler, E., Puck, J., Tang, M. L. K., Tangye, S. G., Torgerson, T. R., & Sullivan, K. E. (2018). International Union of Immunological Societies: 2017 Primary Immunodeficiency Diseases Committee Report on Inborn Errors of Immunity. *Journal of clinical immunology*, 38(1), 96-128. <https://doi.org/10.1007/s10875-017-0464-9>
- Picard, C., & Fischer, A. (2014). Contribution of high-throughput DNA sequencing to the study of primary immunodeficiencies. *European Journal of Immunology*, 44(10), 2854-2861. <https://doi.org/10.1002/eji.201444669>
- Poli, M. C., Aksentijevich, I., Bousfiha, A. A., Cunningham-Rundles, C., Hambleton, S., Klein, C., Morio, T., Picard, C., Puel, A., Rezaei, N., Seppänen, M. R. J., Somech, R., Su, H. C., Sullivan, K. E., Torgerson, T. R., Meyts, I., & Tangye, S. G. (2025). Human inborn errors of immunity: 2024 update on the classification from the International Union of Immunological Societies Expert Committee. *Journal of Human Immunity*, 1(1), e20250003. <https://doi.org/10.70962/jhi.20250003>
- Radinsky, S., & Bonagura, V. R. (2003). Subcutaneous immunoglobulin infusion as an alternative to intravenous immunoglobulin. *Journal of Allergy and Clinical Immunology*, 112(3), 630-633. [https://doi.org/10.1016/S0091-6749\(03\)01781-0](https://doi.org/10.1016/S0091-6749(03)01781-0)

- Rae, W., Sowerby, J. M., Verhoeven, D., Youssef, M., Kotagiri, P., Savinykh, N., Coomber, E. L., Boneparth, A., Chan, A., Gong, C., Jansen, M. H., du Long, R., Santilli, G., Simeoni, I., Stephens, J., Wu, K., Zinicola, M., Allen, H. L., Baxendale, H.,...Smith, K. G. C. (2022). Immunodeficiency, autoimmunity, and increased risk of B cell malignancy in humans with TRAF3 mutations. *Science Immunology*, 7(74), eabn3800.  
<https://doi.org/doi:10.1126/sciimmunol.abn3800>
- Raje, N., & Dinakar, C. (2015). Overview of Immunodeficiency Disorders. *Immunol Allergy Clin North Am*, 35(4), 599-623. <https://doi.org/10.1016/j.iac.2015.07.001>
- Refaat, M., Oujane, C., Kholaiq, H., Aadam, Z., Errami, A., Baghad, B., Boussetta, S., El Kettani, A., Benhsaïen, I., Ailal, F., Bourhanbour, A. D., El Bakkouri, J., & Bousfiha, A. A. (2024). Innate immunodeficiencies: a group of primary immunodeficiencies predisposing exclusively to common diseases. *Egyptian Journal of Medical Human Genetics*, 25(1), 134.  
<https://doi.org/10.1186/s43042-024-00604-4>
- Richards, S., Gennery, A. R., Davies, E. G., Wong, M., Shaw, P. J., Peake, J., Fraser, C., Gray, P., Brothers, S., Sinclair, J., Prestidge, T., Preece, K., Quinn, P., Ramachandran, S., Loh, R., McLean-Tooke, A., Mitchell, R., & Cole, T. (2020). Diagnosis and management of severe combined immunodeficiency in Australia and New Zealand. *J Paediatr Child Health*, 56(10), 1508-1513. <https://doi.org/10.1111/jpc.15158>
- Rider, N. L., Truxton, A., Ohrt, T., Margolin-Katz, I., Horan, M., Shin, H., Davila, R., Tenembaum, V., Quinn, J., Modell, V., Modell, F., Orange, J. S., Branner, A., & Senerchia, C. (2024). Validating inborn error of immunity prevalence and risk with nationally representative electronic health record data. *Journal of Allergy and Clinical Immunology*, 153(6), 1704-1710.  
<https://doi.org/10.1016/j.jaci.2024.01.011>
- Riminton, D. S., Hartung, H.-P., & Reddel, S. W. (2011). Managing the risks of immunosuppression. *Current Opinion in Neurology*, 24(3). <https://doi.org/10.1097/WCO.0b013e328346d47d>
- Roos, D. (2016). Chronic granulomatous disease. *Br Med Bull*, 118(1), 50-63.  
<https://doi.org/10.1093/bmb/lbw009>
- Rosen, F. S., Wedgwood, R. J., Aiuti, F., Cooper, M. D., Good, R. A., Hanson, L. A., Hitzig, W. H., Matsumoto, S., Seligmann, M., Soothill, J. F., & Waldmann, T. A. (1983). Primary immunodeficiency diseases: Report prepared for the WHO by a scientific group on immunodeficiency. *Clinical Immunology and Immunopathology*, 28(3), 450-475.  
[https://doi.org/10.1016/0090-1229\(83\)90112-5](https://doi.org/10.1016/0090-1229(83)90112-5)

---

## ASClA Immunodeficiency References

---

- Salzer, U., Warnatz, K., & Peter, H. H. (2012). Common variable immunodeficiency: an update. *Arthritis Res Ther*, 14(5), 223. <https://doi.org/10.1186/ar4032>
- Segundo, G. R. S., & Condino-Neto, A. (2021). Treatment of patients with immunodeficiency: Medication, gene therapy, and transplantation. *J Pediatr (Rio J)*, 97 Suppl 1(Suppl 1), S17-s23. <https://doi.org/10.1016/j.jped.2020.10.005>
- Shearer, W. T., Fleisher, T. A., Buckley, R. H., Ballas, Z., Ballow, M., Blaese, R. M., Bonilla, F. A., Conley, M. E., Cunningham-Rundles, C., Filipovich, A. H., Fuleihan, R., Gelfand, E. W., Hernandez-Trujillo, V., Holland, S. M., Hong, R., Lederman, H. M., Malech, H. L., Miles, S., Notarangelo, L. D.,...Winkelstein, J. (2014). Recommendations for live viral and bacterial vaccines in immunodeficient patients and their close contacts. *J Allergy Clin Immunol*, 133(4), 961-966. <https://doi.org/10.1016/j.jaci.2013.11.043>
- Sjöholm, A. G., Jönsson, G., Braconier, J. H., Sturfelt, G., & Truedsson, L. (2006). Complement deficiency and disease: an update. *Mol Immunol*, 43(1-2), 78-85. <https://doi.org/10.1016/j.molimm.2005.06.025>
- Slade, C. A., Bosco, J. J., Binh Giang, T., Kruse, E., Stirling, R. G., Cameron, P. U., Hore-Lacy, F., Sutherland, M. F., Barnes, S. L., Holdsworth, S., Ojaimi, S., Unglik, G. A., De Luca, J., Patel, M., McComish, J., Spriggs, K., Tran, Y., Auyeung, P., Nicholls, K.,...van Zelm, M. C. (2018). Delayed Diagnosis and Complications of Predominantly Antibody Deficiencies in a Cohort of Australian Adults. *Front Immunol*, 9, 694. <https://doi.org/10.3389/fimmu.2018.00694>
- Sobh, A., & Bonilla, F. A. (2016). Vaccination in Primary Immunodeficiency Disorders. *J Allergy Clin Immunol Pract*, 4(6), 1066-1075. <https://doi.org/10.1016/j.jaip.2016.09.012>
- Soncini, E., Slatter, M. A., Jones, L. B. K. R., Hughes, S., Hodges, S., Flood, T. J., Barge, D., Spickett, G. P., Jackson, G. H., Collin, M. P., Abinun, M., Cant, A. J., & Gennery, A. R. (2009). Unrelated donor and HLA-identical sibling haematopoietic stem cell transplantation cure chronic granulomatous disease with good long-term outcome and growth. *British Journal of Haematology*, 145(1), 73-83. <https://doi.org/10.1111/j.1365-2141.2009.07614.x>

---

## ASCIA Immunodeficiency References

---

Sriaroon, P., & Ballow, M. (2015). Immunoglobulin Replacement Therapy for Primary Immunodeficiency. *Immunol Allergy Clin North Am*, 35(4), 713-730.  
<https://doi.org/10.1016/j.iac.2015.07.006>

Staudacher, O., & von Bernuth, H. (2024). Clinical presentation, diagnosis, and treatment of chronic granulomatous disease [Review]. *Frontiers in Pediatrics*, Volume 12 - 2024.  
<https://doi.org/10.3389/fped.2024.1384550>

Stewart, O. J., Gruber, C., Randolph, H. E., Patel, R., Ramba, M., Calzoni, E., Huang, L. H., Levy, J., Buta, S., Lee, A., Sazeides, C., Prue, Z., Hoytema van Konijnenburg, D. P., Chinn, I. K., Pedroza, L. A., Lupski, J. R., Schmitt, E. G., Cooper, M. A., Puel, A.,...Bogunovic, D. (2025). Monoallelic expression can govern penetrance of inborn errors of immunity. *Nature*, 637(8048), 1186-1197. <https://doi.org/10.1038/s41586-024-08346-4>

Stiehm, E. R., Casillas, A. M., Finkelstein, J. Z., Gallagher, K. T., Groncy, P. M., Kobayashi, R. H., Oleske, J. M., Roberts, R. L., Sandberg, E. T., & Wakim, M. E. (1998). Slow subcutaneous human intravenous immunoglobulin in the treatment of antibody immunodeficiency: Use of an old method with a new product. *Journal of Allergy and Clinical Immunology*, 101(6), 848-849.  
[https://doi.org/10.1016/S0091-6749\(98\)70314-8](https://doi.org/10.1016/S0091-6749(98)70314-8)

Sudhakar, M., Rikhi, R., Loganathan, S. K., Suri, D., & Singh, S. (2021). Autoimmunity in Wiskott-Aldrich Syndrome: Updated Perspectives. *Appl Clin Genet*, 14, 363-388.  
<https://doi.org/10.2147/tacg.S213920>

Sullivan, K. E. (2022). The yin and the yang of early classical pathway complement disorders. *Clin Exp Immunol*, 209(2), 151-160. <https://doi.org/10.1093/cei/uxac056>

Swain, S., Selmi, C., Gershwin, M. E., & Teuber, S. S. (2019). The clinical implications of selective IgA deficiency. *J Transl Autoimmun*, 2, 100025. <https://doi.org/10.1016/j.jtauto.2019.100025>

Takahashi, K., Ip, W. E., Michelow, I. C., & Ezekowitz, R. A. (2006). The mannose-binding lectin: a prototypic pattern recognition molecule. *Curr Opin Immunol*, 18(1), 16-23.  
<https://doi.org/10.1016/j.co.2005.11.014>

Tam, J. S., & Routes, J. M. (2013). Common variable immunodeficiency. *Am J Rhinol Allergy*, 27(4), 260-265. <https://doi.org/10.2500/ajra.2013.27.3899>

- Tangye, S. G., Al-Herz, W., Bousfiha, A., Chatila, T., Cunningham-Rundles, C., Etzioni, A., Franco, J. L., Holland, S. M., Klein, C., Morio, T., Ochs, H. D., Oksenhendler, E., Picard, C., Puck, J., Torgerson, T. R., Casanova, J.-L., & Sullivan, K. E. (2020). Human Inborn Errors of Immunity: 2019 Update on the Classification from the International Union of Immunological Societies Expert Committee. *Journal of clinical immunology*, 40(1), 24-64. <https://doi.org/10.1007/s10875-019-00737-x>
- Tangye, S. G., Nguyen, T., Deenick, E. K., Bryant, V. L., & Ma, C. S. (2023). Inborn errors of human B cell development, differentiation, and function. *Journal of Experimental Medicine*, 220(7), e20221105. <https://doi.org/10.1084/jem.20221105>
- Thakar, M. S., Logan, B. R., Puck, J. M., Dunn, E. A., Buckley, R. H., Cowan, M. J., O'Reilly, R. J., Kapoor, N., Satter, L. F., Pai, S.-Y., Heimall, J., Chandra, S., Ebens, C. L., Chellapandian, D., Williams, O., Burroughs, L. M., Saldana, B. D., Rayes, A., Madden, L. M.,...Notarangelo, L. D. (2023). Measuring the effect of newborn screening on survival after haematopoietic cell transplantation for severe combined immunodeficiency: a 36-year longitudinal study from the Primary Immune Deficiency Treatment Consortium. *The Lancet*, 402(10396), 129-140. [https://doi.org/10.1016/S0140-6736\(23\)00731-6](https://doi.org/10.1016/S0140-6736(23)00731-6)
- Thong, B. Y., Pawankar, R., Park, H. S., & Abdul Latiff, A. H. (2023). Evaluating immune responses to pneumococcal vaccines. *Asia Pac Allergy*, 13(3), 127-131. <https://doi.org/10.5415/apallergy.000000000000114>
- Thouenon, R., Chentout, L., Moreno-Corona, N., Poggi, L., Lombardi, E. P., Hoareau, B., Schmitt, Y., Lagresle-Peyrou, C., Bustamante, J., André, I., Cavazzana, M., Durandy, A., Casanova, J.-L., Galicier, L., Fadlallah, J., Fischer, A., & Kracker, S. (2023). A neomorphic mutation in the interferon activation domain of IRF4 causes a dominant primary immunodeficiency. *Journal of Experimental Medicine*, 220(6), e20221292. <https://doi.org/10.1084/jem.20221292>
- Tiri, A., Masetti, R., Conti, F., Tignanelli, A., Turrini, E., Bertolini, P., Esposito, S., & Pession, A. (2021). Inborn Errors of Immunity and Cancer. *Biology*, 10(4), 313. <https://doi.org/10.3390/biology10040313>
- Toskov, V., Kaiser-Labusch, P., Lee-Kirsch, M. A., Wolf, C., Speckmann, C., Ehl, S., Wegehaupt, O., & group, P. s. (2024). Variable Syndromic Immunodeficiency in Patients with Biallelic PRIM1 Mutations. *Journal of clinical immunology*, 44(6), 129. <https://doi.org/10.1007/s10875-024-01733-6>

- Truedsson, L. (2015). Classical pathway deficiencies - A short analytical review. *Mol Immunol*, 68(1), 14-19. <https://doi.org/10.1016/j.molimm.2015.05.007>
- Tuovinen, E. A., Grönholm, J., Öhman, T., Pöysti, S., Toivonen, R., Kreutzman, A., Heiskanen, K., Trotta, L., Toiviainen-Salo, S., Routes, J. M., Verbsky, J., Mustjoki, S., Saarela, J., Kere, J., Varjosalo, M., Hänninen, A., & Seppänen, M. R. J. (2020). Novel Hemizygous IL2RG p.(Pro58Ser) Mutation Impairs IL-2 Receptor Complex Expression on Lymphocytes Causing X-Linked Combined Immunodeficiency. *Journal of clinical immunology*, 40(3), 503-514. <https://doi.org/10.1007/s10875-020-00745-2>
- Uzel, G., Orange, J. S., Poliak, N., Marciano, B. E., Heller, T., & Holland, S. M. (2010). Complications of tumor necrosis factor- $\alpha$  blockade in chronic granulomatous disease-related colitis. *Clinical Infectious Diseases*, 51(12), 1429-1434. <https://doi.org/10.1086/657308>
- Valiathan, R., Ashman, M., & Asthana, D. (2016). Effects of Ageing on the Immune System: Infants to Elderly. *Scand J Immunol*, 83(4), 255-266. <https://doi.org/10.1111/sji.12413>
- van der Made, C. I., Kersten, S., Chorin, O., Engelhardt, K. R., Ramakrishnan, G., Griffin, H., Schim van der Loeff, I., Venselaar, H., Rothschild, A. R., Segev, M., Schuurs-Hoeijmakers, J. H. M., Mantere, T., Essers, R., Esteki, M. Z., Avital, A. L., Loo, P. S., Simons, A., Pfundt, R., Warris, A.,...Hoischen, A. (2024). Expanding the PRAAS spectrum: De novo mutations of immunoproteasome subunit  $\beta$ -type 10 in six infants with SCID-Omenn syndrome. *The American Journal of Human Genetics*, 111(4), 791-804. <https://doi.org/10.1016/j.ajhg.2024.02.013>
- Vosughimotagh, A., Rasouli, S. E., Rafiemanesh, H., Safarirad, M., Sharifinejad, N., Madanipour, A., Dos Santos Vilela, M. M., Herropolitańska-Pliszka, E., & Azizi, G. (2023). Clinical manifestation for immunoglobulin A deficiency: a systematic review and meta-analysis. *Allergy, Asthma & Clinical Immunology*, 19(1), 75. <https://doi.org/10.1186/s13223-023-00826-y>
- Wadbudhe, A. M., Meshram, R. J., & Tidke, S. C. (2023). Severe Combined Immunodeficiency (SCID) and Its New Treatment Modalities. *Cureus*, 15(10), e47759. <https://doi.org/10.7759/cureus.47759>
- Wu, Y., Sun, X., Kang, K., Yang, Y., Li, H., Zhao, A., & Niu, T. (2024). Hemophagocytic lymphohistiocytosis: current treatment advances, emerging targeted therapy and underlying mechanisms. *Journal of Hematology & Oncology*, 17(1), 106. <https://doi.org/10.1186/s13045-024-01621-x>

## ASCIA Immunodeficiency References

---

- Xie, C. B., Jane-Wit, D., & Pober, J. S. (2020). Complement Membrane Attack Complex: New Roles, Mechanisms of Action, and Therapeutic Targets. *Am J Pathol*, 190(6), 1138-1150.  
<https://doi.org/10.1016/j.ajpath.2020.02.006>
- Yalcin Gungoren, E., Yorgun Altunbas, M., Dikici, U., Meric, Z., Eser Simsek, I., Kiykim, A., Can, S., Karabiber, E., Yakici, N., Orhan, F., Cokugras, H., Aydogan, M., Ozdemir, O., Bilgic Eltan, S., Baris, S., Ozen, A., & Karakoc-Aydiner, E. (2024). Insights into Patient Experiences with Facilitated Subcutaneous Immunoglobulin Therapy in Primary Immune Deficiency: A Prospective Observational Cohort. *J Clin Immunol*, 44(8), 169. <https://doi.org/10.1007/s10875-024-01771-0>
- Yazdani, R., Azizi, G., Abolhassani, H., & Aghamohammadi, A. (2017). Selective IgA Deficiency: Epidemiology, Pathogenesis, Clinical Phenotype, Diagnosis, Prognosis and Management. *Scandinavian Journal of Immunology*, 85(1), 3-12.  
<https://doi.org/https://doi.org/10.1111/sji.12499>
- Yazdani, R., Fekrvand, S., Shahkarami, S., Azizi, G., Moazzami, B., Abolhassani, H., & Aghamohammadi, A. (2019). The hyper IgM syndromes: Epidemiology, pathogenesis, clinical manifestations, diagnosis and management. *Clin Immunol*, 198, 19-30.  
<https://doi.org/10.1016/j.clim.2018.11.007>
- Yel, L. (2010). Selective IgA deficiency. *J Clin Immunol*, 30(1), 10-16. <https://doi.org/10.1007/s10875-009-9357-x>
- Younger, M. E. M., Aro, L., Blouin, W., Duff, C., Epland, K. B., Murphy, E., Sedlak, D., & Nurse Advisory Committee Immune Deficiency, F. (2013). Nursing Guidelines for Administration of Immunoglobulin Replacement Therapy. *Journal of Infusion Nursing*, 36(1).  
<https://doi.org/10.1097/NAN.0b013e3182798af8>
- Younger, M. E. M. e., IDF (2016). Guide for Nurses. *Immunoglobulin Therapy for Primary Immunodeficiency Diseases*. Immune Deficiency Foundation.  
<https://primaryimmune.org/resources/print-material/idf-guide-for-nurses-immunoglobulin-therapy-for-primary-immunodeficiency-diseases>
- Yu, J. E., Azar, A. E., Chong, H. J., Jongco, A. M., 3rd, & Prince, B. T. (2018). Considerations in the Diagnosis of Chronic Granulomatous Disease. *J Pediatric Infect Dis Soc*, 7(suppl\_1), S6-s11.  
<https://doi.org/10.1093/jpids/piy007>

## ASCIA Immunodeficiency References

---

Zimmerman, C., & Shenoy, S. (2020). Chimerism in the Realm of Hematopoietic Stem Cell Transplantation for Non-malignant Disorders—A Perspective [Perspective]. *Frontiers in Immunology*, Volume 11 - 2020. <https://doi.org/10.3389/fimmu.2020.01791>

NOTE: There is a separate ASCIA reference list for Hereditary Angioedema (HAE) publications on the ASCIA website [www.allergy.org.au/hp/papers#p4](http://www.allergy.org.au/hp/papers#p4)