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| **Disease** | **Cause** | **Effect** |
| Severe Combined Immunodeficiency Disease (SCID) | Mutation in signalling subunit of IL-7 receptor | Block T cell maturation 🡪 reduced B and T cell response |
| Deficiency in Adenosine Deaminase (ADA) | Accumulation of toxic purine metabolites in proliferating lymphocytes 🡪 reduced B and T cells |
| Deficiency in RAG ½ or ARTERMIS gene mutation | Block VDJ recombination 🡪 absence of T and B cells |
| X-linked agammaglobulinemia (XLA) | Defect in Bruton Tyrosine Kinase (BTK) gene | Blocks development of pre-B cell to immature B cell 🡪 low concentrations of serum Ig 🡪 reduction of lymphoid tissues, lack of plasma cells and germinal centers🡪 recurrent RTI by pyogenic bacteria and Ab-neutralized viruses |
| Autoimmune Polyendocrine Syndrome Type I (APS I)  [Others:  Autoimmune Polyendocrine Syndrome Type II (APS 2)  IPEX Syndrome] | Mutation in AIRE gene | Endocrine failure/hyperactivity;  2 of: Chronic Mucocutaneous candidiasis, Auotimmune adrenalitis, Autoimmune Hypoparathyroidism |
| Immune dysregulation, Polyendocrinopathy, Enteropathy, X-linked disease (IPEX Syndrome) | Mutation in FOXP3 gene | Absent/dysfunctional Treg cells 🡪 autoimmunity (Type 1a Diabetes) in neonates  \*BMT can prolong survival |
| Lepromatous and Tuberculoid Leprosy | Mounting a Th1 response 🡪 Tuberculoid leprosy  Mounting a Th2 response 🡪 Lepromatous Leprosy | Tuberculoid: low infectivity, localized infection, normal serum Ig, normal T cell response  Lepromatous: high infectivity, disseminated infection, hypergammaglobulinaemia |
| X-linked Hyper IgM Syndrome | Mutation in the CD40L gene | No CD40L-CD40 interaction 🡪 no class switching 🡪 no IgG, IgA, IgE, high IgM 🡪 infection with pyogenic bacteria and intracellular microbes |
| IgA Deficiency | Inability of naive B cells to differentiate into IgA | Low levels of serum and secretory IgA 🡪 increased resp, GI, and urogentical infections |
| Autoimmune Lymphoproliferative Syndrome | Mutations in Fas ligand gene | Defects in the Activated Induced Cell Death (AICD) – defective apoptosis of self-reactive T and B cells in the periphery |