Infection and autoimmune disease in 22q11.2 deletion syndrome

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Infection was one of the defining features in DiGeorge’s description. He realized that the central problem was a small thymus gland, the gland that is responsible for the education of developing T cells. In the half century since his description and the recognition of 22q11.2 deletions as the most common etiology for DiGeorge syndrome, we have come to appreciate the tremendous variability in the features in terms of organ system involvement and severity of the effects. The immune system is similarly variable in 22q11.2 deletion syndrome. Laboratory evidence of immune deficiency is seen in the majority of infants but standard evaluations are often normal in adulthood, belying the functional deficits. Infections as a result of the immune deficiency are typical sinopulmonary infections. Opportunistic infections are rare and limited to those infants with thymic aplasia or near aplasia. Risk from live viral vaccines are minimal unless the CD4 T cell count is <400 cells/mm3. Infections are usually treated with standard anti-microbial approaches. Some people benefit from prophylactic antibiotics but some people have an anatomical conformation that is unfavorable and few strategies are proven to work in those cases. Atopy and autoimmunity are downstream consequences of a poor immune system and these clinical findings are increased in people with 22q11.2 deletion syndrome.