Autoimmune polyendocrinopathy candidiasis ectodermal dystrophy (APECED) is a recessive autoimmune disease caused by a loss of function mutation in the Autoimmune Regulator (AIRE) transcription factor.[[1]](#endnote-1) Patients with APECED suffer from endocrine disorders, such as hypoparathyroidism and adrenocortical insufficiency, along with ectodermal conditions, most notably chronic mucocutaneous candidiasis.[[2]](#endnote-2) The many autoimmune symptoms found in APECED patients have been linked to an overall loss of central tolerance.[[3]](#endnote-3) Tolerance is lost because AIRE’s primary role is in the thymus, where it promotes transcription of self-antigens in medullary thymic epithelial cells. These cells then kill self-reactive T-cells in a process called negative selection.[[4]](#endnote-4) However, recent studies have also identified extrathymic AIRE expressing cells (eTACs) responsible for maintaining peripheral tolerance through expression of self-antigen. Notably, the self-antigens expressed by eTACs differ from those in medullary thymic epithelial cells.[[5]](#endnote-5) *Despite these findings, the role of AIRE in promoting transcription of these unique tissue specific antigens in eTACs remains unclear.*

1. Finnish-German APECED Consortium. 1997. An autoimmune disease, APECED, caused by mutations in a novel gene featuring two PHD-type zinc-finger domains. Nature. https://www.ncbi.nlm.nih.gov/pubmed/9398840/​ [↑](#endnote-ref-1)
2. Kisand, Kai and Peterson, Pärt. 2015. Autoimmune Polyendocrinopathy Candidiasis Ectodermal Dystrophy. Journal of Clinical Immunology. pp 463-478. [↑](#endnote-ref-2)
3. Peterson et al. 2004. Autoimmune polyendocrinopathy candidiasis ectodermal dystrophy (APECED): a model disease to study molecular aspects of endocrine autoimmunity. Clinical and Experimental Immunology. pp 348–357 [↑](#endnote-ref-3)
4. Owen et al. 2013. Kuby Immunology. 7th edition. [↑](#endnote-ref-4)
5. Gardner et al. 2008. Deletional Tolerance Mediated by Extrathymic Aire-Expressing Cells. Science. Vol 231. pp. 843-847. [↑](#endnote-ref-5)