

Commentary

The Use of TREC Analysis as a Newborn Screening Test for Jacobsen Syndrome and T cell Deficiency

Amrita Dosanjh

Pediatric Respiratory, San Diego, California, USA

Among the advances in medical detection of newborn disease, is the development of the T cell excision receptor circle or TREC assay. The use of the assay in the detection of Severe Combined Immunodeficiency (SCID) and T cell lymphopenia in newborn screening facilitates the detection of SCID early in life. TRECs may be used as surrogate markers for the production of newly formed T cells. To date, as of 2015, 26 states have implemented newborn screening utilizing TREC analysis. Cases of SCID have been identified with TREC analysis, and as a result the revised population based estimate of the incidence of SCID has been increased to an estimated 1 in every 58,000 newborn infants [1]. With the advancement in molecular diagnostics, other conditions and syndromes associated with T cell deficiency may be detected and treated earlier in life.

Syndromes which may be associated with SCID, or other T cell lymphopenias, include Jacobsen Syndrome (JS). JS is a congenital disorder associated with terminal deletion of chromosome 11. The features of this syndrome include cardiac defects (congenital heart disease), intellectual disability, growth retardation, low platelets and variable immune defects [2]. A defect in the ETS-1 gene is a candidate gene for contributing to the immune disorders in JS. ETS-1 plays a critical role in the development of lymphocytes [3]. Among JS cases identified in the literature, there have been reports of combined variable immunodeficiency (CVID), or T cell lymphopenia, among patients with recurrent infections. The JS patients described were treated with IVIG and antibiotics. Some JS patients will exhibit selective T helper cell deficiency, while others have lymphopenia and T cell lymphopenia with functional defects in response to mitogens [4-6]. To date in the California program, there have been no cases of TREC detected immune disease among JS patients (email communication, CA). Among infants identified with T cell lymphopenia, it is not known if JS was considered in the evaluation. The application of TREC analysis in the detection of immune defects in JS is an area of future analysis. A multi-center approach to analyze stored dried blood spots may yield further insight regarding immune defects in this rare disease.

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Corresponding Author: pulmd@aol.com

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DISCLOSURE

The author declares no conflicts of interest.

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