

# 21-HYDROXYLASE AND INTERFERON OMEGA AUTOANTIBODIES IN TURNER SYNDROME

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## INTRODUCTION

An increased frequency of autoimmune diseases and an elevated incidence of autoantibodies have been observed in Turner syndrome (TS). Indirect immunofluorescence (IIF) has not been able to demonstrate autoantibodies against the adrenal cortex. We asked if the more sensitive radioimmunosorbant assay (RIA) employing recombinant human 21-hydroxylase was able to identify autoantibodies against 21-hydroxylase, (21OH-Ab) in TS patients; 21-hydroxylase is the major adrenal cortex autoantigen in patients with autoimmune Addison's disease. Moreover, TS patients were tested for antibodies against interferon omega (IFN $\omega$ -Ab), a marker for autoimmune polyendocrine syndrome 1 (APS 1) where autoimmune Addison's disease is one of the main components along with chronic mucocutaneous candidiasis and hypoparathyroidism.

## METHODS

Blood samples from 144 karyotyped TS (11-62 years) were assayed for 21OH-Ab and IFN $\omega$ -Ab using in vitro transcribed and translated autoantigen. An index was calculated [(cpm sample - cpm negative control / cpm positive control - cpm negative control) x 1000] with a cut-off point of 57 and 200 for 21OH-Ab and IFN $\omega$ -Ab, respectively. Pooled human serum from healthy donors was used as negative control, and serum from patients with known high antibody titers were used as positive controls.

## RESULTS

Autoantibodies against 21-hydroxylase with low indices (index range 60-535) were present in 6 TS patients (4.2%); none had interferon omega autoantibodies. Overall, the TS patients had a mean age of 31.6 years (range 11.2-62.2). 53% (n=77) had the karyotype 45,X, 4% (n=6) had the karyotype 46,Xi(X)(q), and 9% (n=13) had the karyotype 45,X/46,Xi(X)(q). Hypothyroidism was recorded in 9% (n=20), coeliac disease in 1.4% (n=2), and type 1 diabetes mellitus in 0.7% (n=1). The six TS patients with 21-hydroxylase antibodies had a mean age of 32.7 years (range 17.7-44.7). Two of the six patients had the karyotype 45,X. One patient had hypothyroidism, but none had clinical apparent Addison's disease.

## CONCLUSION

21-hydroxylase autoantibodies can be detected by using RIA in some patients with TS. These findings add to previous studies showing a high proportion of TS with an array of different autoimmune antibodies. Whether any of the autoantibody-positive TS patients will eventually develop Addison's disease remains to be seen.

