Youngest Known Case of Autoimmune Thyroiditis Causing Hyperthyroidism in a Down's Syndrome Toddler



Parissa Salemi D.O.
Assistant Professor
Division of Pediatric Endocrinology
Department of Pediatrics
Cohen Children's Medical Center of Northwell Health
New Hyde Park, New York

Background: The frequency of thyroid disease is elevated in patients with Down's syndrome starting in the newborn period where it is 0.7% (28x more frequent than in the general population). Most commonly, thyroid dysfunction in Downs Syndrome includes primary hypothyroidism, pituitary-hypothalamic hypothyroidism, TBG deficiency and chronic lymphocytic thyroiditis. The incidence of Grave's disease is believed to be between 0.1 and 3 per 100,000 children in the general population. Autoimmune thyroid disease is uncommon in young children with Down's syndrome less than 8 years of age with hyperthyroidism being quite rare.

Objective and hypotheses: To determine if yearly screening labs done on a 15 month old female with Down's syndrome who has a strong family history of autoimmune thyroid disease with initial lab values of TSH=5.77 and FT4=1.77 is due to Hashimoto's thyrotoxicosis, Grave's hyperthyroidism or transient. My hypothesis was that this was due to autoimmune thyroiditis given the elevated FT4 and strong family history.

Method: To obtain thyroid stimulating immunoglobulins, thyroglobulin antibodies and thyroid peroxidase antibodies and to observe tsh and ft4 over time for progression.

Results: Lost to follow up until age 21 months when labs obtained: TSH=<0.001uU/ml, FT4=2.25ng/dl(nl <1.6), TT3=215ng/dl(High), TT4=16.4ug/dl(4.5-12.0), TGAB<20iu/ml, TPO AB=167iu/ml(nl<9), TSI=284(nl<140). Patient started on methimazole 2.5 mg qd with labs 2 weeks later showing a TSH<0.001 and FT4 of 2.52. Methimazole increased to 5 mg qd.

Conclusion: Given progression of hyperthyroidism over a 6.5 month time course even after starting on methimazole, in a patient with highly positive TSI antibodies, this is very likely to be autoimmune hyperthyroidism caused by Grave's disease. To my knowledge, this is the youngest known patient with DS to have autoimmune hyperthyroidism to date. Family history of autoimmune disease may make DS patients even more susceptible to development of autoimmune thyroiditis at an earlier age.

References

Bauer, Andrew J., Approach to the Pediatric Patient with Grave's Disease: When definitive therapy is warranted?, JCEM July 2013











