Metamorphic thyroid autoimmunity in Down Syndrome: from Hashimoto’s thyroiditis to Graves’ disease and beyond

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Disclosure Statement
Nothing to disclose

Purpose: To shed further light on the specific relationships between Down syndrome (DS) and metamorphic thyroid autoimmunity.

Design: We have reconstructed the conversion process from Hashimoto’s thyroiditis (HT) to Graves’ disease (GD) in a selected population consisting of 12 DS individuals aged between 3.0 and 13.5 years at HT diagnosis and between 4.1 and 19.9 years at GD diagnosis. All the patients underwent a treatment with methimazole (MMI), at a dose that was periodically adjusted on the basis of clinical findings and thyroid function tests.

Results: After MMI treatment onset all patients exhibited, at varying time intervals, a prolonged clinical and biochemical remission of hyperthyroidism. In 8/12 patients this treatment is still being continued 2-7 years after its initiation. The mean MMI dosage needed to maintain euthyroidism in these 8 patients was 0.12 ± 0.02 mg/kg/day. In the remaining 4 patients MMI was withdrawn from 1.9 to 7 years after its initiation and no relapses were recorded 2.0 – 2.1 years after its withdrawal. All these 4 patients developed, from 0.1 to 0.3 years after MMI withdrawal, a biochemical picture of overt hypothyroidism and needed treatment with L-T4, that is now being continued since 2.0 - 2.1 years.

No patients needed non-pharmacological therapies, such as surgery or radioiodine ablation.

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nts Age
(years)
TSH
(mU/l)
FT4°°
(pmol/l)
TPOAbs*
(IU/ml)
TGAbs**
(IU/ml)
Therapy duration
(years)
1 3.0 7.10 12.6 95 60 -
2 3.0 19.56 9.9 131 182 2.5
3 3.3 0.07 24.6 57 187 -
4 4.0 8.80 16.8 180 120 4.0
5 4.0 5.30 12.1 26 28 -
6 5.0 0.05 50.0 284 35 1.0
7 5.9 5.10 17.4 75 302 -
8 6.0 8.20 10.5 144 104 1.0
9 8.0 0.01 38.0 278 199 -
10 9.7 9.10 11.0 37 2 6.5
11 10.0 5.10 10.5 22 119 3.0
12 13.5 4.30 18.0 23 109 1.0

Table 1
Age, TSH, FT4 and thyroid autoantibody serum levels at Hashimoto’s thyroiditis diagnosis in the 12 patients of this series and duration of L-thyroxine treatment in the 7 patients who were treated.

Table 2

Conclusions

1) DS children may be incline to manifest over time a phenotypic metamorphosis from HT to GD.
2) A share of GD children with DS may subsequently fluctuate from hyperthyroidism to hypothyroidism.
3) In DS HT presentation is absolutely peculiar.
4) In DS GD is characterized by a mild biochemical and clinical course.