Association between Alport's Syndrome and Familial Goiter

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Dear Sir,

Alport's syndrome [1] is a hereditary glomerular nephropathy which may lead to chronic renal insufficiency, especially in males; it has been found associated with many extrarenal abnormalities among which nerve deafness is the most common. An obvious thyroid abnormality must be rare since Gubler et al. [2] did not mention any in 58 cases of Alport's syndrome. Only Miyoshi et al. [3] have reported two families with antithyroid antibodies.

We report the presence of thyroid disorders in one family with Alport's syndrome. The family tree (fig. 1) shows 17 members, 8 of which had a nephropathy and 6 a thyroid disorder. Severity of the nephropathy was found variable in the 8 affected cases. Subjects 7, 8, 10 and 13 had chronic renal insufficiency from glomerulonephritis cause of 3 early deaths at an average age of 23, all 3 being males and suffering from deafness. Subjects 1, 5 and 12 had permanent albuminuria and macroscopic hematuria. Subject 9 had intermittent microscopic hematuria only. Thus, Alport's syndrome is confirmed in this family.

In the same kindred, 6 cases of thyroid disorder were found including 5 obvious goiters. 3 (subjects 1, 3 and 11) were euthyroid goiters although subject 11 developed hypo-thyroidism after excision of the goiter. In addition, evidence for 3 cases of primary thyroid insufficiency (subjects 13 and 15 with goiter and subject 16 without) was provided by the serum TSH levels: 100, 34 and 10 $\mu\text{U/ml}$ respectively (normal value $3.5 \pm 1.4 \mu\text{U/ml}$). 2 of these cases were clinically euthyroid siblings (subjects 15 and 16) and thyroid investigation disclosed low ^{123}I radioiodine uptake (14% at 1 h), negative perchlorate discharge test and normal T4 and T3 serum concentrations. The young age of these siblings precludes any further thyroid investigation, and the mechanism of the thyroid disorder remains uncertain. The discrepancy observed between the raised TSH and the low thyroid uptake suggests that some resistance to TSH takes place while the goitrogenic effect of TSH remains

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Fig.1. Family tree. Underlined numbers: number of the subject; other numbers: age of the subject at the time of the investigation or at death; solid squares or circles: nephropathy; double symbols: thyroid disorder; diagonal line: dead.

unimpaired. Autoimmunity plays no apparent role since antithyroid antibodies were found undetectable in subjects 11–17.

The occurrence of 2 patients of inherited diseases (renal and thyroidal) in one family with Alport's syndrome is reported here for the first time. Whether they are related or not remains to be seen.

References

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