

Mapping and Screening in Families With Multiple Endocrine Neoplasia Type 2A: Four Reports

Recently, multiple endocrine neoplasias type 2A have been mapped to chromosome 10. A number of polymorphic DNA markers around the gene allow prediction in most families of those individuals who are carriers of the gene. In addition, prospective screening annually for manifestations of the disease appears to be effective in prevention of morbidity and mortality. For example, provocative tests to guarantee the release of calcitonin can be used to monitor whether or not "medullary" thyroid carcinoma is present, and 24-hour urine screening for both epinephrine excretion and the ratio of urinary epinephrine to norepinephrine allows detection of proliferation of the adrenal medulla before life-threatening manifestations occur.

An 18-year follow-up study of a large family by Gagel et al suggests that total thyroidectomy, when done at the first appearance of increased calcitonin secretion, is curative since there were no recurrences or metastatic diseases in their patients. Parathyroid dis-

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ease seemed to occur only in those patients with well-established medullary thyroid carcinoma or pheochromocytoma. Because more than 50% of affected individuals within the family eventually developed adrenal medullary abnormalities, screening in such families is mandatory.

Sobol H, Salvetti A, Bonnardel C, et al. *Lancet* 1988;i:62.

Gagel RF, Tashjian AH, Cummings T, et al. *N Engl J Med* 1988; 318:478-484.

Mathew CGP, Chin KS, Easton DF, et al. *Nature* 1987;328:527-528.

Simpson NE, Kidd KK, Goodfellow PJ, et al. *Nature* 1987;328: 528-530.

Editor's comment—*The potential for malignancy of multiple endocrine neoplasia type 2A is frightening, but these new chromosomal and metabolic screening techniques allow us to recognize family members at risk. The screening techniques also suggest clear and reliable methods to be used in following at-risk individuals.*

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