Pediatric Head and Neck Manifestations Associated with Multiple Endocrine Neoplasia Syndromes

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INTRODUCTION: Multiple endocrine neoplasia (MEN) syndromes are a group of hereditary cancer syndromes that can predispose children to endocrine neoplasms developing within the head and neck.
OBJECTIVE: To examine the neoplastic manifestations of MEN1 and MEN2 in the pediatric head and neck.
METHODS: Single-institution, retrospective review of pediatric MEN between 2005 and 2022.
RESULTS: Fifty-three children were genetically confirmed with MEN (15 MEN1, 34 MEN2A, and 4 MEN2B), while three patients received clinical diagnoses of MEN1. The male to female ratio was essentially equal (1.15:1), and a family history of cancer was present in 98% (50/51). After evaluation, a familial MEN diagnosis was confirmed in 91% (51/56). The mean ages of initial presentation and surgical intervention were 8.9 years (SD 5) and 9.8 years (SD 4.8), respectively. Although MEN2 patients received surgery earlier than MEN1 patients (8.7 vs 12.7 years), MEN2 surgical patients in this cohort were older relative to current American Thyroid Association (ATA) guidelines primarily due to late presentation. Thyroid malignancies were identified in 36% (9/25) of thyroidectomy specimens (21 MEN2A, 4 MEN2B), with medullary thyroid carcinoma (MTC) present in five MEN2A patients and three MEN2B patients (89%), and papillary thyroid carcinoma (PTC) present in one MEN2A patient (11%). Nearly 90% (8/9) of thyroid malignancies were occult, with some occurring earlier than predicted by current guidelines (ATA-MOD and ATA-H). Neck dissections were performed in 24% (2 MEN1, 2 MEN2A, and 4 MEN2B), with two MEN2B (50%) demonstrating cervical lymph node (LN) metastases. Additional histopathologic findings included C-cell hyperplasia in 57% (12/21) of MEN2A thyroidectomy patients. Of the eight MEN1 parathyroidectomy patients, four demonstrated parathyroid hyperplasia, and four presented with parathyroid adenoma.
CONCLUSION: MEN syndromes predispose children to early neoplasia within the pediatric head and neck, with our review finding pediatric patients still presenting late relative to current ATA guidelines. MEN2 children in lower ATA risk categories have significant risk for developing early MTC, suggesting current surgical guidelines for these patients may be too conservative.