Tumor Biology

ENDOCRINE NEOPLASIA CASE REPORTS

Calcitonin Changed During Pregnancy in MEN2A With MTC Patient: A Case Report.

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Background: The diagnosis of MTC during pregnancy was challenging. No definite calcitonin (Ct) cut-off level during pregnancy was defined. Moreover, cytology analysis accuracy in MTC was lower than other thyroid cancers. Stevenson et al. reported that the plasma Ct levels in pregnant women were much higher than normal women. The change in Ct levels during pregnancy was not well described. In this report, we present a case of a pregnant woman with recurrent MTC. The change in Ct levels during pregnancy, diagnosis methods, and proper management were presented here.
Clinical Case: A 31-year Thai woman, gravida 2, para 0 at 17 weeks of pregnancy, previously diagnosed with MTC and treated with total thyroidectomy and neck dissection seven years ago. During antenatal care at 14 weeks of pregnancy, the Ct level was 126 pg/ml (raised from 34 pg/ml seven years ago). The neck's USG demonstrates vascularized hypoechoic lesion at the right thyroid bed with multiple nodules. The Ct wash-out test from this lesion was 200,000 pg/ml. The cytology study showed several clusters of atypical cells possessing monotonous round eccentric nuclei, and the IHC for Ct was positive. Genetic analysis revealed germline mutation in exon 10 of the RET proto-oncogene (NM_020975.4: c.1858T>G, p.C620G). Finally, MEN 2A with recurrent MTC was diagnoses during pregnancy. The other associate diseases were not found (normal urine metanephrines and plasma calcium levels). The Ct level raised highest to 324 pg/ml at 28 weeks of pregnancy and declined to 236 pg/mL a few days after delivery. Within six weeks, the Ct levels raised again (335 and 362 pg/ml at six weeks and three months after delivery).
After giving birth, she underwent neck dissection; The Ct level at one month after surgery was still elevated (85 pg/ml). The CT of the chest, abdomen, and bone scan revealed multiple LN and bone metastasis. She regularly followed up in the oncology clinic and did well until nowadays (two years after delivery). After birth, her son could not pass meconium. Causing underwent exploratory laparotomy with double end ileostomy at day nine of life. The histopathology study from serial bowel biopsy demonstrated aganglionosis of the entire colon, which compatible with Hirschsprung's disease. Genetic study from colonic tissue and blood confirmed the diagnoses of MEN 2A like his mother. Total thyroidectomy before five years of age and investigate for other associated diseases were planned.
Conclusion: The Ct levels were elevated throughout the pregnancy. Transient declined of Ct levels during delivery and raised within six weeks suggest that pregnancy did not affect the Ct levels. In this report, we want to present the change of Ct levels during pregnancy, emphasize the diagnosis of MTC and MEN2A in pregnant women, which may change the patient's prognosis. Not only in the patients but also in their families. Clinicians should be aware of the management of these patients.

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Carney Complex With Endocrine Involvement Isolated to the Thyroid Gland

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Introduction: Carney complex is a rare autosomal dominant disorder characterized by pigmented lesions of skin and mucosae, endocrine neoplasms or overactivity, and myxomas of the heart, skin, and other organs. Most patients have at least two affected endocrine organs at time of diagnosis. We present a case of Carney complex with endocrine involvement limited to the thyroid gland.
Case: A 48-year-old female was referred for evaluation of thyroid nodules incidentally discovered on imaging for submandibular salivary gland swelling. Ultrasound evaluation of the thyroid revealed numerous, bilateral nodules that were mostly cystic or spongiform, though some had irregular borders and microcalcifications. A brother with Carney complex had been diagnosed with papillary carcinoma, and the patient decided to undergo thyroidectomy. Fortunately, no thyroid carcinoma was found on postsurgical histopathology. Initial biochemical evaluation showed no evidence of hypercortisolemia (8 AM cortisol 1.7 mcg/dL on 1 mg dexamethasone suppression test; 24-h urine cortisol 26.1 mcg [**reference:** 4-50]), growth hormone excess (IGF-1 190 ng/mL [**reference:** 52-328]), or hyperprolactinemia (prolactin 10 ng/mL [**reference:** 2.74-26.72]). Imaging showed no pituitary or adrenal masses. The patient underwent total hysterectomy with bilateral salpingo-oophorectomy for endometrial cancer prior to referral. She was diagnosed with Carney complex at age 19 years, and her manifestations included atrial and ventricular myxomas, intraductal adenoma of the breast, multiple skin lesions (lentigines, blue nevi, and cutaneous myxomas), and myxomas of the external auditory canals. She is in a Carney complex kindred that includes her mother, two brothers, and a niece. **Discussion:** Carney complex is usually caused by inactivating mutations or large deletions in the protein kinase A type I alpha regulatory subunit (*PRKAR1A*) gene located on chromosome 17q22-24. Most mutations are inherited in an autosomal dominant manner, though approximately 30% of cases are due to *de novo* mutations. In a review of 365 cases, the median age at diagnosis was 20 years. Growth hormone adenomas or somatomammotroph hyperplasia occurs in approximately 75% of patients, and most have at least one additional endocrine abnormality including multinodular goiter, primary pigmented nodular adrenocortical hyperplasia, and testicular or ovarian masses. There is an approximately 10% chance of developing well differentiated