Ectopic cervical thymoma (ECT) is a rare tumor that is frequently misdiagnosed as a thyroid tumor or other malignancy. A 34-year-old male with a right palpable neck mass had been mistakenly diagnosed with T-cell lymphoblastic lymphoma even after an open biopsy. The atypical clinical course, including hypogammaglobulinemia, led us to the correct diagnosis: ECT accompanied by Good’s syndrome (GS). After the intravenous infusion of gammaglobulin, tumor resection and a subsequent video-assisted thoracoscopic extended thymectomy were performed. The final diagnosis was type AB thymoma, Masaoka stage I. This report is, to the best of our knowledge, the first description of this extremely rare combination.

**Keywords:** thymic epithelial tumor, paraneoplastic autoimmunity, immunodeficiency

**Introduction**

Ectopic cervical thymoma (ECT) is a rare tumor which is thought to result from ectopic thymic tissue. ECT is difficult to diagnose due to its rarity and location, and it is sometimes accompanied by parathyroid syndromes. Good’s syndrome (GS), characterized by hypogammaglobulinemia, is a rare adult-onset immunodeficiency associated with thymoma. Patients with GS are vulnerable to infections resulting from defects in both humoral and cell-mediated immunity, and have significant mortality. We herein describe a case of ECT accompanied by GS with a diagnostic process that should be of interest to clinicians.

**Case Report**

A previously healthy 34-year-old male was admitted to another hospital because of a palpable right anterior cervical mass, which had been incidentally found during a medical check-up. An open biopsy showed lymphocytes of various sizes that were positive for CD3, CD5, CD10, and TdT by immunohistochemical staining. Under a diagnosis of T-cell lymphoblastic lymphoma (T-LBL), 2 cycles of hyper-CVAD (cyclophosphamide, vincristine, doxorubicin, and dexamethasone)/MA (methotrexate and cytosine arabinoside) chemotherapy were administered; however, the mass did not decrease in size. Unusual clinical manifestations, including hypogammaglobulinemia, led us to redefine the diagnosis; ECT accompanied by Good’s syndrome (GS). After the intravenous infusion of gammaglobulin, tumor resection and a subsequent video-assisted thoracoscopic extended thymectomy were performed. The final diagnosis was type AB thymoma, Masaoka stage I. This report is, to the best of our knowledge, the first description of this extremely rare combination.
At presentation, the patient had normal findings on a physical examination, except that he was suffering from a superficial tinea infection. Complete blood cells counts and a serum chemistry panel showed a decreased leukocyte count of 2,660/μl, serum total protein of 6.1 g/dl, with normal albumin of 4.6 g/dl. The low serum levels of immunoglobulin indicated the existence of hypogammaglobulinemia, with IgG 472 mg/dl, IgA 20 mg/dl, and IgM 20 mg/dl. The CD4/CD8 ratio (1.4) and peripheral B cell count (5%). The titer of the anti-acetylcholine receptor antibody (AchR-Ab) in the serum was elevated at 2.20 nmol/l, but he had no symptoms of myasthenia gravis (MG). No serum tumor markers were elevated. Computed tomography (CT) demonstrated a homogeneous soft tissue density, that was a 27 × 35 × 40 mm oval-shaped mass with sharp borders in the right cervix (arrow). The mass was not invasive to the neighboring structures. The thymus gland was slightly hypertrophic. The operation time was 362 minutes, and the blood loss was 110 ml. Microscopically, the tumor contained a combination of both lymphocyte-poor spindle cell areas and lymphocyte-rich areas; and was finally diagnosed as a type AB thymoma, Masaoka stage I (Fig. 2). The patient was discharged 19 days after the operation, although he experienced transient right recurrent laryngeal nerve palsy. There has been no sign of recurrence nor infections except for his skin lesions, with periodical intravenous infusion of gammaglobulin as of the 4-month postoperative follow-up examination.

Discussion

GS, first described by Good in 1955, is a rare association of thymoma and immunodeficiency. Although there remains some controversy, GS is currently defined as immunodeficiency with thymoma, most commonly characterized by hypogammaglobulinemia.1 The incidence of hypogammaglobulinemia is reported as 6%–11% in patients with thymoma in English literature,2 while that is reported as 0.2% in Japanese patients.3 This difference might be explained by the racial difference in incidence and/or the definition of the disease. Patients with GS are usually in their fourth to seventh decades, and develop either thymoma or infectious complications. Their clinical presentation is similar to patients with X-linked agammaglobulinemia and common variable immunodeficiency; GS patients have an increased susceptibility to
Ectopic Cervical Thymoma

infections related to both humoral and cell-mediated immune deficiencies. Fortunately, the postoperative course of the present patient was uneventful, without any infection. The prognosis of GS is reported to be poor, with a significant overall mortality of 44.5%. The surgical indications were considered to be the malignant mediastinal tumor and stable general status of the patient in the present case.

ECT, first described by Boman in 1941, is thought to result from ectopic thymic tissue that develops due to defective migration along the path of descent in the embryonic stage. ECT is a rare tumor, and only 41 cases have been reported in English literature so far. The clinical features of ECT are quite different from those of mediastinal thymoma. Most patients are females presenting with palpable masses in the neck, which are often misdiagnosed as either thyroid tumors or other malignancies. Eleven out of 13 cases (84.6%) in the literature were not correctly diagnosed by biopsy, including the diagnosis of T-LBL in our present patient. Chang provided another example of ECT mistaken for T-LBL in 2003, which had a similar diagnostic process to the present case.

The complication rate of parathymic syndromes with ECT seems to be lower than that of mediastinal thymoma; there were 5 cases of MG (12.2%), 1 case of hyperparathyroidism (2.4%), and 1 case of hypoparathyroidism (2.4%) out of the 41 cases reported. We herein reported a case of ECT accompanied by GS. This is, to the best of our knowledge, the first report of this extremely rare combination. It is worth mentioning that the patient’s atypical clinical course, where the tumor neither grew rapidly nor responded to the chemotherapy, and the presence of hypogammaglobulinemia made us suspect thymoma in an ECT patient. An immunohistochemical study of epithelial markers should be added for patients with lymphocyte-rich cervical tumors, considering the possibility of detecting a thymoma.

ECT is reported to be mostly non-invasive (35 cases; 85.4%) and complete resection of the tumor is generally considered to be a sufficient treatment. Adjuvant radiotherapy or chemotherapy is an option for invasive cases. In the present case, resection of the tumor through a cervical incision and subsequent video-assisted thoracoscopic extended thymectomy with the sternum-lifting method were performed because the patient had elevated AchR-Ab, although he lacked symptoms of MG. The outcomes of video-assisted thoracoscopic extended thymectomy have been reported to be comparable to those of trans-sternal extended thymectomy in patients with MG. On the other hand, resection of the thymoma and extended thymectomy does not usually reverse the immunological abnormalities of GS. Therefore, regular follow-up with close attention to the development of infections is necessary. For patients with GS, periodic intravenous infusions of gammaglobulin has been reported to reduce the incidence of infections, and is recommended. With the preoperative administration of gammaglobulin, our patient did not develop any perioperative infections.

Conclusion

In conclusion, we herein reported the first case of ECT accompanied by GS. Thoracic surgeons should therefore be aware of this rare tumor when a cervical tumor with parathymic syndromes is encountered.

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