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Case report

Ectopic cervical thymus associated with infant death: 2 case reports and literature review

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ABSTRACT

An ectopic cervical thymus is a rare congenital anomaly that can be located anywhere along the developmental pathway of thymic descent. Most lesions manifest as a cystic mass and have an indolent course. Two fatal cases associated with ectopic cervical thymus in the form of a solid mass are presented in conjunction with a review of the clinicopathological characteristics of the solid form. This report emphasizes the importance of considering a diagnosis of ectopic cervical thymus in infants with neck masses, with or without obstructive symptoms, to prevent possibly fatal outcomes.

1. Introduction

Ectopic cervical thymus (ECT) is a rare congenital anomaly found in 1% of pediatric autopsies [1]. Most cases follow an indolent course, but several cases have shown life-threatening symptoms such as respiratory distress due to tracheal compression [2–5]. Only 1 fatal case, caused by mechanical asphyxia, has been previously reported [6]. We report 2 sudden, unexpected deaths of infants with ECT and review 95 cases of solid ECT to clarify the clinicopathological features of these lesions.

2. Case reports

2.1. Case 1

A 4-month-old boy was found dead in a prone position, approximately 5 h after falling asleep in a supine position. His mother, who had previously borne 3 other children, did not have a significant family medical history; the pregnancy was unremarkable and the baby, who was bottle-fed, was in good health until death. The mother stated that the infant had recently started rolling over, but was not yet proficient. He had received a second combination vaccine against diphtheria, pertussis, and tetanus 2 days before his death.

Microbiological tests for rotavirus, respiratory syncytial virus, and influenza virus were negative. Tandem mass spectrometry screening showed no metabolic disorders. Horizontal sections of a computed tomography (CT) scan, performed 1-h postmortem, showed 2 isodense nodules immediately inferior to the thyroid gland (Fig. 1).

A complete postmortem examination, performed 13 h after death, revealed a well-developed and well-nourished boy (height, 66.0 cm; weight, 8.5 kg). External examination showed no apparent anomalies or injuries. An examination of his cervical organs revealed 2 solid nodules (maximum diameters, 1.5 and 1.9 cm), with intact capsules, lying anterolateral to the cervical trachea and immediately inferior to the thyroid gland (Fig. 2A). Histological examination of the nodules showed normal thymic structure, indicating a diagnosis of ECT (Fig. 2B–D). An intrathoracic examination confirmed a normally positioned thymus weighing 50 g (expected weight, 38.1 g [7]). The other findings were characteristic of a rapidly fatal course caused by mechanical asphyxia. Other remarkable physical or toxicological findings were not found.

The postmortem findings led to the conclusion that the cause of death was mechanical asphyxia, likely due to compression of the cervical trachea by the 2 ECTs while the infant was prone. The ECTs were considered to play a crucial role in the mechanical asphyxia because they were located such that they could easily compress the cervical trachea and were large enough to significantly compress it.

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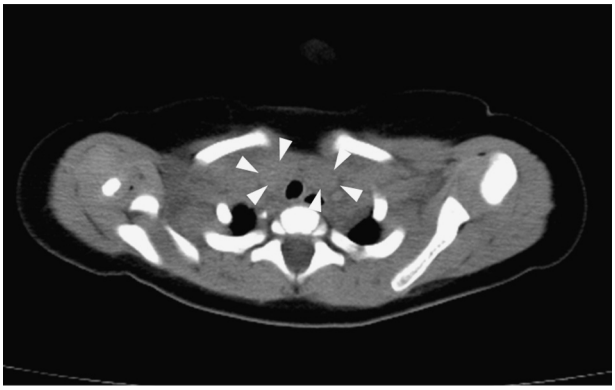

Fig. 1. Postmortem computed tomography image of the neck. A horizontal image shows bilateral, solid nodules (surrounded by white arrowheads) anterolateral to the cervical trachea and immediately inferior to the thyroid gland.

2.2. Case 2

A 2-month-old boy was found dead in a supine position, 6 h after falling asleep. He was his mother's third child, was breast-fed, and did not have a significant family history or any birth or growth abnormalities.

A complete postmortem examination, performed 30 h after death, revealed a well-developed and well-nourished boy (height, 58.0 cm; weight, 5.6 kg). External examination failed to reveal any apparent anomalies or injuries. An internal examination, followed by a histological examination, revealed a right-sided ECT (diameter, 1.2 cm), anterior to the cervical trachea and immediately inferior to the thyroid gland (Fig. 3). A normally positioned thymus weighing 37 g (expected weight, 30.4 g [7]) was also found. No other remarkable physical, toxicological, or microbiological findings were found. Vitreous chemistry was not performed.

The cause of death was diagnosed as Category IB sudden infant death syndrome (SIDS), according to the San Diego definition [8]. The ECT was not considered to have been involved in the death because it could not compress the trachea while the boy was in a supine position.

3. Discussion

ECTs show 2 different macroscopic forms—cystic and solid. The cystic form is more frequently reported and accounts for 76–92% of ECTs [9–12]. This form includes 2 different entities: a thymic cyst, arising from a persistent thymopharyngeal duct, and a solid ECT, with cystic changes [11–13]. The pathogenesis of solid ECTs involves the failure of the thymus gland to descend or its sequestration and failure to involute [11–14]. All previous reviews included both forms in their analyses and documented their combined features [9–12]. Consequently, the previous reviews reflect mainly the features of cystic ECTs.

Thus far, a review focused on solid ECTs has not been reported. We searched the literature for solid ECT cases, confirmed by histological examination, and reviewed the cases to clarify their clinicopathological features. Overall, 93 cases of solid ECTs were found in 30 reports. The clinical and anatomical characteristics of the 95 solid ECT cases, including the present 2, are summarized in Tables 1 and 2, respectively [1,3–5,11,12,14–38].

Solid ECTs are most frequently seen in males, with patient age ranging from birth to 30 years (median age, 3 months). Most cases (70%) have been diagnosed at a patient age of <6 months. Approximately 36% of patients had a concomitant congenital anomaly/disorder, but none were strongly associated with ECTs. In most patients, surgical resection or autopsy was required for a correct diagnosis [14], except for a recent case diagnosed by fine needle aspiration (FNA) [14]. The majority of symptoms associated with solid ECTs are neck masses, as previously reported (Table 1) [9–12]. Respiratory symptoms and/or dysphasia are seen in approximately 12% of solid form cases, but the overall prevalence of these symptoms in ECT patients is approximately 5% [9–12], indicating that respiratory symptoms occur more frequently in cases involving solid ECTs. Therefore, this anomaly should be added to the list of differential diagnoses of such symptoms in infants.

The respiratory symptoms are induced by the lesion's compression of the cervical trachea [3,26,29]. Severe respiratory distress had developed in some serious cases [2–5,21], and mechanical asphyxia leading to death was reported in an additional instance [6]. Shackelford et al. also reported a case, similar to our first case, presenting with tracheal compression and

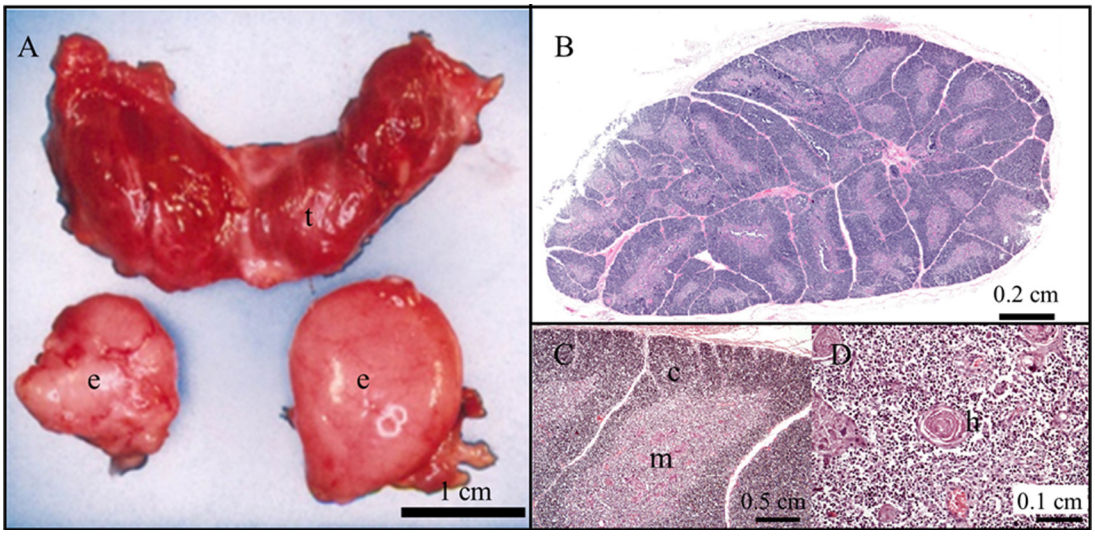

Fig. 2. Gross appearance of the resected specimens showing 2 solid nodules beneath the thyroid gland (A). Histological examination (hematoxylin and eosin stain) of the nodules revealing normal architecture (B) and microscopic components of the thymus tissue (C and D). c, cortex; e, ECT; h, Hassall's corpuscles; m, medulla; t, thyroid gland.