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The inflatable thymus herniation of the normal mediastinal thymus: A case report and review of the literature

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ABSTRACT

Anterior neck masses in young children can be a diagnostic challenge for otolaryngologists and radiologists. We present a rare case of herniation of normal mediastinal thymus in a four-year-old girl. Additional medical features as an inguinal hernia and trochlear nerve paresis raised the question whether there is a causal relationship or an association. A connective tissue disorder could not be diagnosed as possible causal factor to the abnormal movement of the mediastinal thymus. Awareness and recognition of this benign phenomenon is important in order to avoid unnecessary biopsy or surgery. Diagnosis can be confirmed by ultrasonography. Magnetic Resonance Imaging might be valuable in order to obtain more information about the extension of the mass.

1. Introduction

Herniation of a normal mediastinal thymus is clinically visible as a suprasternal swelling in the neck that appears during an increase in intrathoracic pressure. This is an unusual cause of neck mass in children and it has been reported in literature in only a few cases [1–5]. In this case report, a four-year-old girl with an anterior neck mass and some additional medical features is presented. Clinical workup, differential diagnosis and an overview of all reported cases are presented.

2. Case report

A four-year-old girl presented with an asymptomatic suprasternal neck mass. Her parents noticed this mass a few weeks earlier and it was only visible when she cried or strained. She is the first child of non-consanguineous Dutch parents. The medical history of this girl mentions a surgical repair of a unilateral inguinal hernia and an idiopathic fourth cranial nerve paresis on the right side. The girl's growth parameters were normal. Psychomotor development was normal and she attends a regular school. During physical observation the following features were noticed: minimal heterochromia iridum, a vascular naevus on the forehead and in the neck and a small sacral dimple. There was no joint hyperlaxity or hypermobility. The Beighton Hypermobility Score was 0/7 [6]. Family history was negative for any congenital malformation.

Physical examination revealed a mass in the midline of the anterior neck, only visible during Valsalva maneuver (Fig. 1). The overlying skin appeared to be normal. The mass was tender and immobile during swallowing. A murmur or thrill could not be found. During normal breathing the mass was neither visible nor palpable.

Laryngoscopy demonstrated no structural abnormalities at rest or during Valsalva maneuver. Ultrasonography showed that almost the complete thymus protruded from the mediastinum into the suprasternal neck during forced expiration and Valsalva maneuver. The thymus had a normal appearance on ultrasonography, no vascular malformations or lymphadenopathy were seen (Fig. 2). Magnetic Resonance Imaging (MRI) of the chest and neck was performed. It confirmed an anterior mediastinal thymus that extended cervically more than two centimeters above the manubrium in rest, without any structural anomalies (Fig. 3).

These results in combination with patient specific medical history raised the question of a possible rare genetic syndrome. Therefore the pediatrician and the clinical geneticist were

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Fig. 1. The anterior neck mass is visible during Valsalva maneuver.

consulted. Both medical specialists could not confirm an underlying genetic condition. We saw no indication for a specific DNA test or for a whole exome sequencing. In conclusion, a diagnosis of herniation of a normal mediastinal thymus was made.

Follow-up after two years showed a slight decrease in size of the mobile mass. The patient could be discharged from further follow-up.

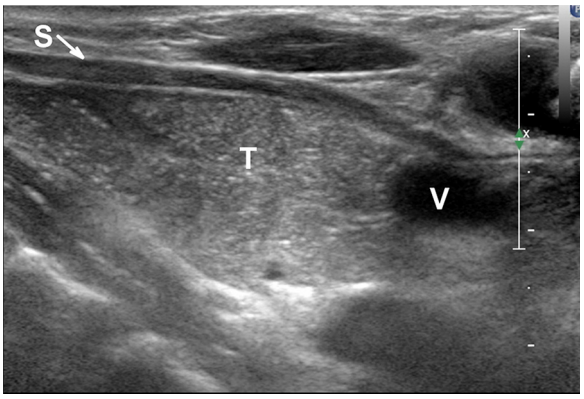

Fig. 2. Sagital ultrasonographic image of the suprasternal region of the neck during Valsalva maneuver. The whole thymus (T) protrudes in the neck. V = left brachiocephalic vein. S = sternothyroïd muscle.

3. Discussion

We report a case of a neck herniation of the non-pathological mediastinal thymus in a four-year-old girl. This condition is defined as "intermittent migration of the broadest part of the normal thymus out of the thorax in to the suprasternal region during increase in intrathoracic pressure"[7]. This is an extremely rare cause of a neck mass in children, with only five cases reported in literature (Table 1) [1–5]. However, the combination of this phenomenon with other medical features such as heterochromia iridum, a vascular naevus, a small sacral dimple, an inguinal hernia and a trochlear palsy, has not been reported before. Although thorough examination could not confirm any known hereditary disorder, the coincidence of all these features is remarkable.

Medical history, physical examination and ultrasonography normally lead to the diagnosis of herniation of a mediastinal thymus [1]. Typically, relatives of the patient notice a neck mass during crying or straining. Physical examination reveals an anterior neck mass, which appears or enlarges during Valsalva maneuver. Ultrasound is the imaging of first choice and typically

Table 1
Cases of herniation of the normal mediastinal thymus reported in the literature to date.

Author	Year of publication	Age at presentation	Sex	Symptoms	Medical history	Family history	Radiography	Ultrasound	MRI	CT	Airway fluoroscopy	Follow-up
Van Zwieten, Stuut et al.	2015 (Present case)	4 yr	F	Asymptomatic	Unilateral inguinal hernia, fourth cranial nerve paresis,	Negative	–	+	+	–	–	Persistent at age 6 yr
Ranga et al.	2015	6 yr	M	Asymptomatic	N/A	N/A	–	+	–	–	–	N/A
Su et al.	2014	7 mo	M	Intermittent stridor, mild dyspnoea	Bilateral congenital talipes equinovarus	Negative	+	+	–	–	–	N/A
Mc Dougall et al.	2012	8 mo	M	Asymptomatic	Intrauterine growth retardation, respiratory distress (viral infection)	Negative	+	+	–	–	+	Persistent at age 16 mo
Senel et al. [2]	2008	8 yr	M	Asymptomatic	Mild	Presented	+	+	–	+	–	N/A
		7 yr	M	Asymptomatic	Intellectual disabily N/A	cases are siblings	+	+	–	+	–	N/A
Wong et al.	2005	8 yr	M	Asymptomatic	N/A	N/A	–	+	+	–	–	N/A

(2) = two cases presented, F = female. M = male, N/A = not applicable, + = performed, – = not performed.