

# Unique finding in congenital muscular torticollis

## Clinic screening on the neck of one day old neonate and ultrasonographic imaging from birth through 3 years of follow-up

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### Abstract

Congenital muscular torticollis (CMT) is a common musculoskeletal abnormality in children, which has been characterized by unclarified pathological changes in the sternocleidomastoid muscle (SCM) and various hypothetical etiologies. There are 2 main hypothetical etiologies for CMT in the literature: 1 infers that CMT may represent the sequela of an intrauterine or perinatal compartment syndrome, and the other regard CMT as a maldevelopment of the fetal SCM.

To better understand the etiopathogenesis of CMT, we screened the necks of 1-day-old newborns that may potentially have CMT for evidence of SCM trauma or tumor.

A convenience sample of 2564 full-term (>37 weeks) Chinese neonates were included in this study. All neonates were screened for CMT by physical examination at birth. If CMT was suspected, further ultrasonic and physical examinations were performed. When CMT was confirmed, we provided appropriate interventions and follow-up. The progress and changes in patients with CMT were recorded.

Following physical examination, 44 of 2564 neonates were diagnosed with suspected CMT based on obvious facial asymmetry or palpable swelling or mass in the SCM. Among these, ultrasound examination showed 81.8% (36/44) had asymmetry in the thickness of the bilateral SCM. The 36 neonates were followed-up for 6 months; among them, 1 infant developed CMT and 35 showed normal development in bilateral SCM. The 1 patient with CMT underwent regular physiotherapy and recovered with no evidence of recurrence after the final 3 years of follow-up. No neonates suffered from signs of neck trauma, such as hematoma or subcutaneous ecchymosis.

There was no evidence of neck trauma in this 1 day old newborn. The pseudotumor of SCM that developed after birth underwent differentiation, maturation, and disappeared as the baby grew. The SCM asymmetry did exist in some of the newborn babies, and became symmetric with the baby's growth. Data from this clinical study and our previous ultra-structural pathological studies suggested that both prenatal and postnatal factors play important roles in CMT. We hypothesized that CMT might be a developmental disease.

**Abbreviations:** CMT = congenital muscular torticollis, SCM = sternocleidomastoid muscle, SCMPOI = SCM pseudotumor of infants, sCMT = suspected CMT.

**Keywords:** congenital muscular torticollis, long-term follow-up, screening, ultrasonography

Editor: Johannes Mayr.

The research was supported by Shenzhen Innovation Science and Technology Committee (JCYJ20130401114111461).

The authors have no conflicts of interest to disclose.

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Medicine (2019) 98:11(e14794)

Received: 22 May 2018 / Received in final form: 10 February 2019 / Accepted: 14 February 2019

<http://dx.doi.org/10.1097/MD.0000000000014794>

## 1. Introduction

Congenital muscular torticollis (CMT) is a common disorder characterized by unilateral shortening of the sternocleidomastoid muscle (SCM). CMT usually develops by 2 to 4 weeks of age and occurs with a reported prevalence of 0.3% to 2%.<sup>[1–4]</sup> Pathological features of CMT include proliferation of fibrotic and adipose tissue in the SCM.<sup>[5,6]</sup>

CMT was first described in 1912, but its etiology remains unknown.<sup>[1,7,8]</sup> Several theories have been proposed, including intrauterine malposition or crowding,<sup>[2,9,10]</sup> muscle trauma due to a complicated delivery,<sup>[2,9–11]</sup> compression of soft-tissue resulting in compartment syndrome, and impaired soft-tissue differentiation in the SCM.<sup>[11,12]</sup> One theory for the compartment syndrome is that the SCM is selectively injured in utero by the head position, and the CMT may represent the sequela of an intrauterine or perinatal compartment syndrome.<sup>[12]</sup> To date, these theories have not been substantiated since a hemorrhagic inflammatory reaction or myofiber disruption is often not observed in the SCM of infants with CMT.<sup>[11]</sup> In previous

ultra-structural and pathological studies, we showed that the interstitium of the mass in patients with SCM pseudotumor of infants (SCMPOI) contained mesenchyme-like cells, myoblasts, fibroblasts, and myofibroblasts; the myoblasts were at different stages of differentiation and degeneration depending on the age of the infant and the duration of the condition.<sup>[5]</sup> We hypothesized that the myoblasts in the SCM originated from remnant mesenchyme-like cells that remain static during embryogenesis. After birth, these mesenchyme-like cells differentiate to muscle, fibrous, and adipose tissues. Then pseudotumor emerges and develops into CMT. An imbalance of fibrogenesis, adipogenesis, and myogenesis can cause the mass to develop into CMT.<sup>[5,6]</sup>

Clinically, if the neck was screened by physical examination in 1-day-old newborns, the pseudotumor of the SCM can be detected if it was present in the uterus. However, if compartment syndrome was responsible for the etiology,<sup>[11,12]</sup> the SCM injury can also be found in the neck by screening. The aim of the current study was to better understand the etiopathogenesis of CMT by applying our previous ultra-structural and pathological findings<sup>[5,6]</sup> to the clinical setting. Furthermore, screening the newborn's neck is a practical and effective approach for the early detection of SCM injury or tumor. Therefore, we screened one-day old Chinese neonates for CMT, and then followed-up the patients diagnosed with CMT for 3 years to investigate the clinical manifestations of CMT and ultrasonic features of the SCM.

## 2. Materials and methods

### 2.1. Study population

This study included a convenience sample of full-term (>37 weeks) 1-day-old neonates who were born in the Peking University Shenzhen Hospital between October 2013 and November 2013 and in the Shenzhen Maternity and Child Healthcare Hospital between December 2013 and May 2014.

Oral informed consent was obtained from all legal guardians of each neonate. The study protocol conformed to the ethical guidelines of the 1975 *Declaration of Helsinki* and was approved by the Ethics Committee on Human Research of the Peking University Shenzhen Hospital, the Shenzhen Maternity and Child Healthcare Hospital and the Shenzhen Children's Hospital (2013013, August 20, 2015).

### 2.2. CMT diagnostic criteria

The criteria for the suspected CMT (sCMT) in 1-day-old neonates included the following:

1. unilateral neck flexion and head tilt at inspection,
2. minimal restriction in range of neck movement in the physical exam, and/or
3. severe plagiocephaly.

The criteria for establishing a diagnosis of CMT in 1-day-old neonates included all of the following:

1. obvious unilateral neck flexion and head tilt,
2. limited range of neck movement;
3. palpable tumor on SCM; and
4. abnormal echotexture of SCM in ultrasonography.

### 2.3. Screening

Medical records were reviewed. The mother's name, admission number, gravidity, parity, length of gestation, method of delivery,

volume of amniotic fluid, length of labor, and the neonate's name, gender, birth weight, and Apgar score were recorded. All neonates at both centers were physically examined by one attending doctor. If CMT was suspected, ultrasonography was performed to confirm the diagnosis. If a diagnosis of CMT was confirmed, appropriate interventions were provided. Otherwise, parents were advised to attend regular check-ups with a healthcare professional until the child was 1 year old.

### 2.4. Physical and ultrasonic examination

Each neonate was examined in the supine position in a room maintained at 25°C. By providing no support to the head and neck, the examiner first observed and palpated the neck, then performed a passive rotation of the neck to check the range of cervical movement.

The ultrasound examination was performed by an experienced sonographer. Using a 5 to 12 MHz linear array ultrasound transducer (General Electric LOGIQ-E9, USA), the examiner scanned the longitudinal and transverse views to directly measure the diameter of the SCM. The examiner observed the morphology and echogenic characteristics and measured the size of the SCM tumor. Sonograms and data of the affected and unaffected SCM were compared.

## 3. Results

### 3.1. Screening results

This study included 2564 neonates. Of these, 44 neonates were diagnosed with sCMT. One neonate showed limited passive range of motion in the neck. No neonates suffered from signs of neck trauma, such as hematoma or subcutaneous ecchymosis. Among the 44 neonates with sCMT, there were 22 males and 22 females with a mean body weight of 3.05 kg (range, 2.52–4.10 kg) and a mean body length of 50.3 cm (range, 49–53 cm); 18 neonates were born normally and 26 neonates were born by cesarean section. All sCMT were detected by ultrasonography one day after birth. Ultrasound showed that 81.8% of (36/44) these neonates had asymmetry in the thickness of the left and right SCM, and there was no difference in the thickness of the right and left SCM in 18.2% (8/44) of the neonates. The former 36 neonates were followed-up for 6 months; among them, 1 infant developed CMT and 35 showed normal developments of both SCM. Therefore, asymmetry in the thickness of the left and right SCM disappeared during the 6 months of follow-up in 35 infants, and none of these infants developed clinical manifestations of CMT through 1 year of follow-up (Fig. 1).

### 3.2. Follow-up results

Nevertheless, 1 infant finally developed CMT. This infant was born by cesarean section to a gravidity 1 and parity 1 mother. At the time of birth, the amniotic fluid was clear, the neonate had a weight of 3.78 kg, and the 1, 5, and 10-minute Apgar scores were 10.

A physical examination on the first day after birth showed obvious facial asymmetry due to the smaller right hemiface. There were no signs of ecchymosis of the neck. The left SCM was taut compared to the right side, and no palpable lump or mass existed. Furthermore, the range of neck motion was normal.

Ultrasonography showed that the thickness of the right and left sides of the SCM were 0.48 cm and 0.68 cm, respectively, at 1 day after birth, and 0.62 cm and 0.65 cm, respectively, at 7 days after



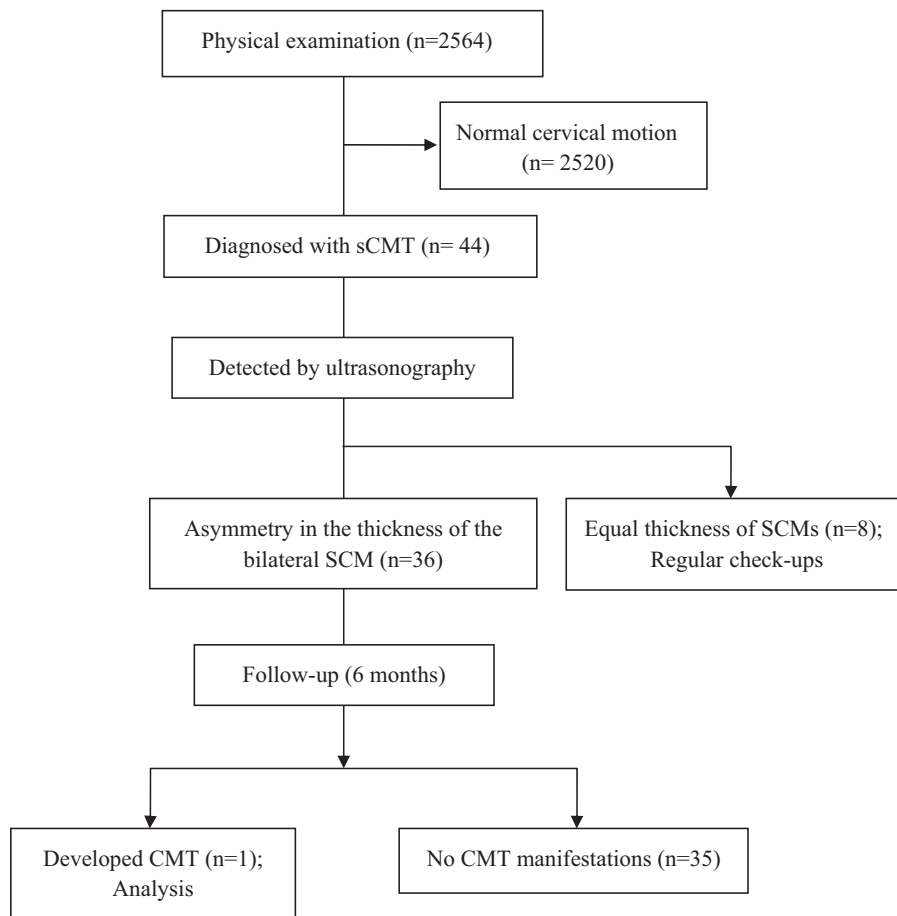


Figure 1. Screening process.

birth (Figs. 2 and 3). Likewise, there were no signs of a SCM mass or tumor in the SCM at these times.

However, the physical examination at 15 days after birth revealed obvious facial asymmetry due to the smaller right hemiface, and the head was tilted to the right side. Besides, there

was a palpable tumor (3.0 × 2.0 × 1.0 cm) in the middle of the right SCM, and right rotation of the neck was decreased by 30° compared to the unaffected side. Ultrasound examination of the middle aspect of the right SCM showed hyperechoic muscle tissue thickening (0.64 cm). The contralateral SCM presented a uniform

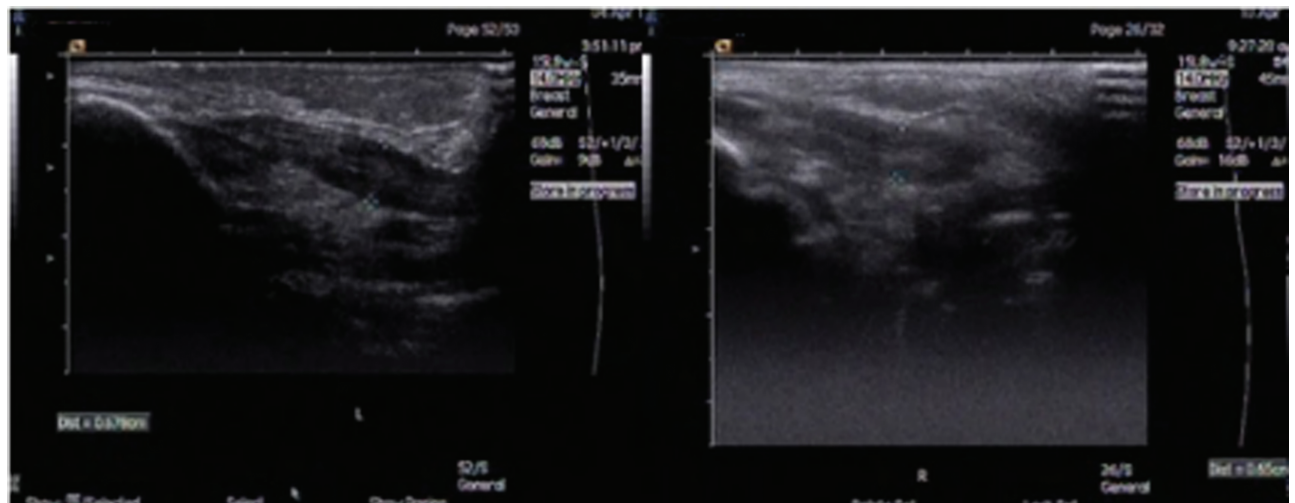
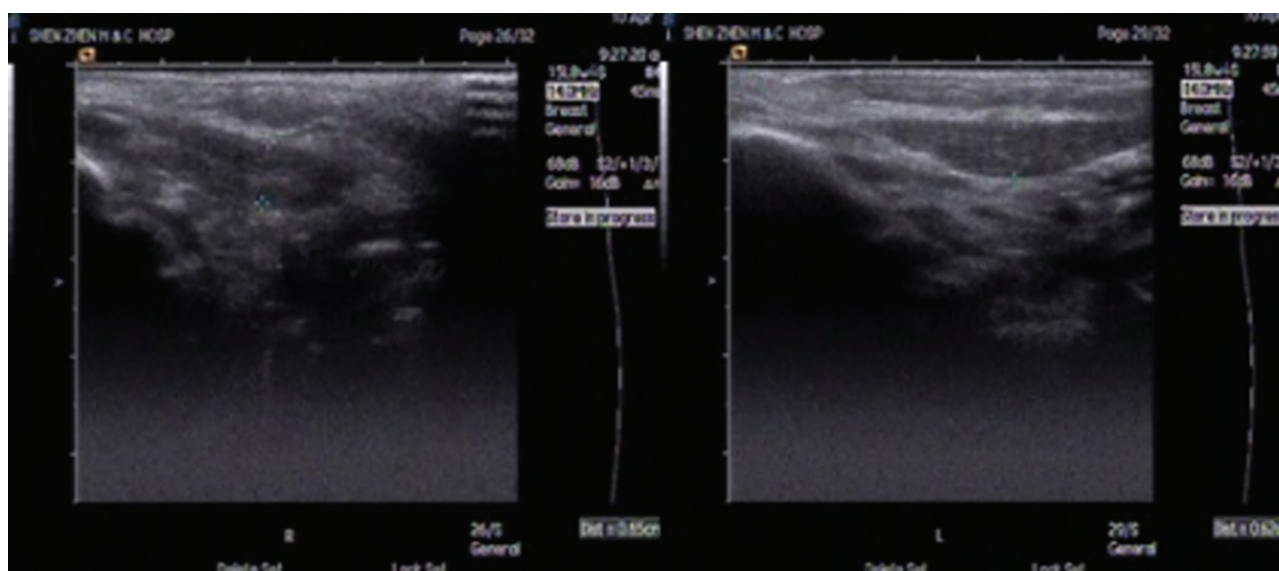


Figure 2. One day after birth: ultrasound of the right (a) and left (b) sternocleidomastoid muscles.



**Figure 3.** Seven days after birth: ultrasound of the right (a) and left (b) sternocleidomastoid muscles.

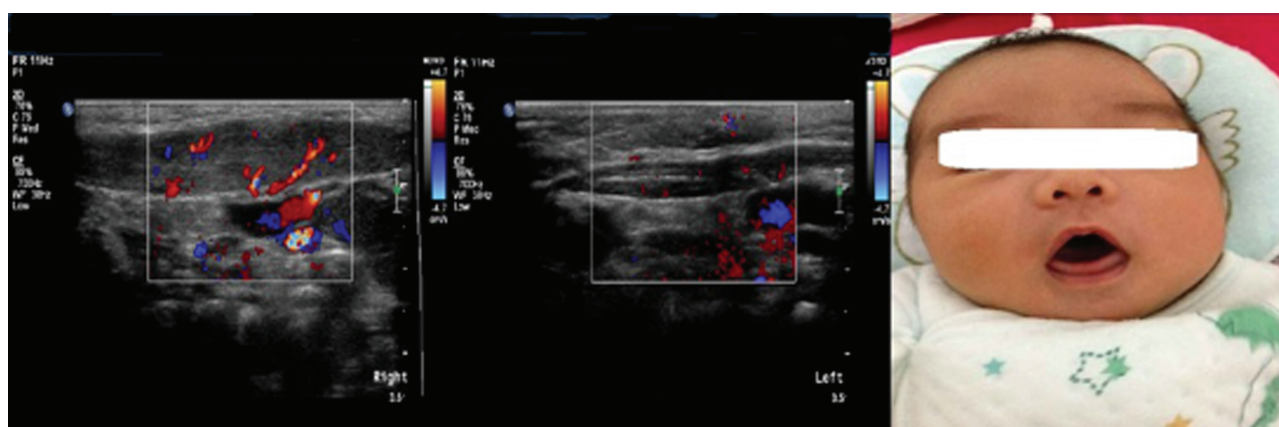
appearance, with diameters of 0.41, 0.43, and 0.42 cm for the upper, middle, and lower parts of the SCM, respectively (Fig. 4). Based on these findings and the clinical manifestations, the infant was diagnosed with CMT.

Physical examination at 22 days after birth revealed obvious facial asymmetry and limited rotational range of the neck. The tumor had increased in size ( $4.0 \times 3.0 \times 2.0$  cm), which encompassed the entire right SCM. The range of neck rotation was limited to  $45^\circ$  (Fig. 5a). Ultrasound examination revealed the affected SCM were hyperechoic with an abnormal thickness of upper, middle, and lower segments (0.81, 0.93, and 0.85 cm, respectively). While, the unaffected SCM still indicated a normal echotexture and thickness of corresponding segments (0.42, 0.43, and 0.42 cm, respectively) (Fig. 5b). Therefore, physiotherapy treatment was administered to the patient.

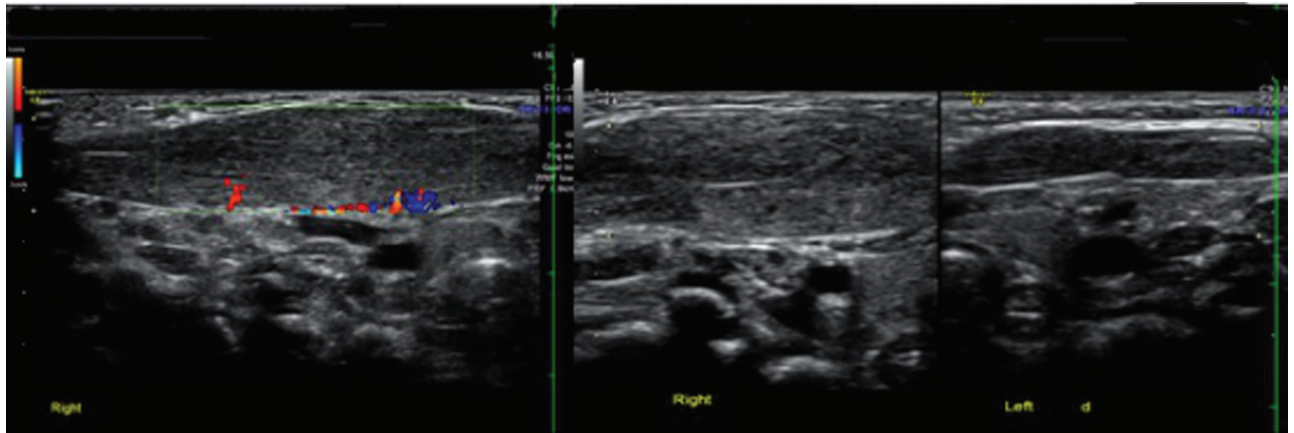
At 29 days after birth, the patient's right SCM was torn during physiotherapy, which resulted in the appearance of a large soft mass ( $5.00 \times 3.00 \times 2.00$  cm) in the middle-lower aspect of the right SCM (Fig. 6a). This allowed the neck to regain  $30^\circ$  of motion. Ultrasonography was immediately

performed, revealing that the right SCM was hyperechoic with multiple low level internal echos. We note abnormal SCM textures with abnormal diameters (max thickness of upper, middle, and lower aspects: 0.66, 1.24, and 1.00 cm, respectively). The muscle fibers in the upper aspect of the right SCM appeared disorganized, which were rounded by dark fluid-filled hypoechoic areas (max depth: 0.50 cm). In contrast, the left SCM showed a normal appearance and thickness of upper, middle, and lower segments (0.43, 0.44, and 0.43 cm, respectively) (Fig. 6b).

The patient received regular physiotherapy twice a week in our clinic. Six months later, the patient showed no torticollis, and the tumor in the affected SCM had completely disappeared. Additionally, the range of neck motion recovered completely. However, the facial asymmetry remained (Fig. 7). After 2 years of further follow-up, the patient's symptoms disappeared gradually. At the last follow-up visit, there was no evidence of an SCM mass or tumor, with free neck rotation. A sonogram of the right SCM revealed a uniform echo, which was thinner than the unaffected SCM (Fig. 8a, b).



**Figure 4.** Fifteen days after birth: ultrasound of the right (a) and left (b) sternocleidomastoid muscles; photograph showing the infant having a smaller right hemiface (c).

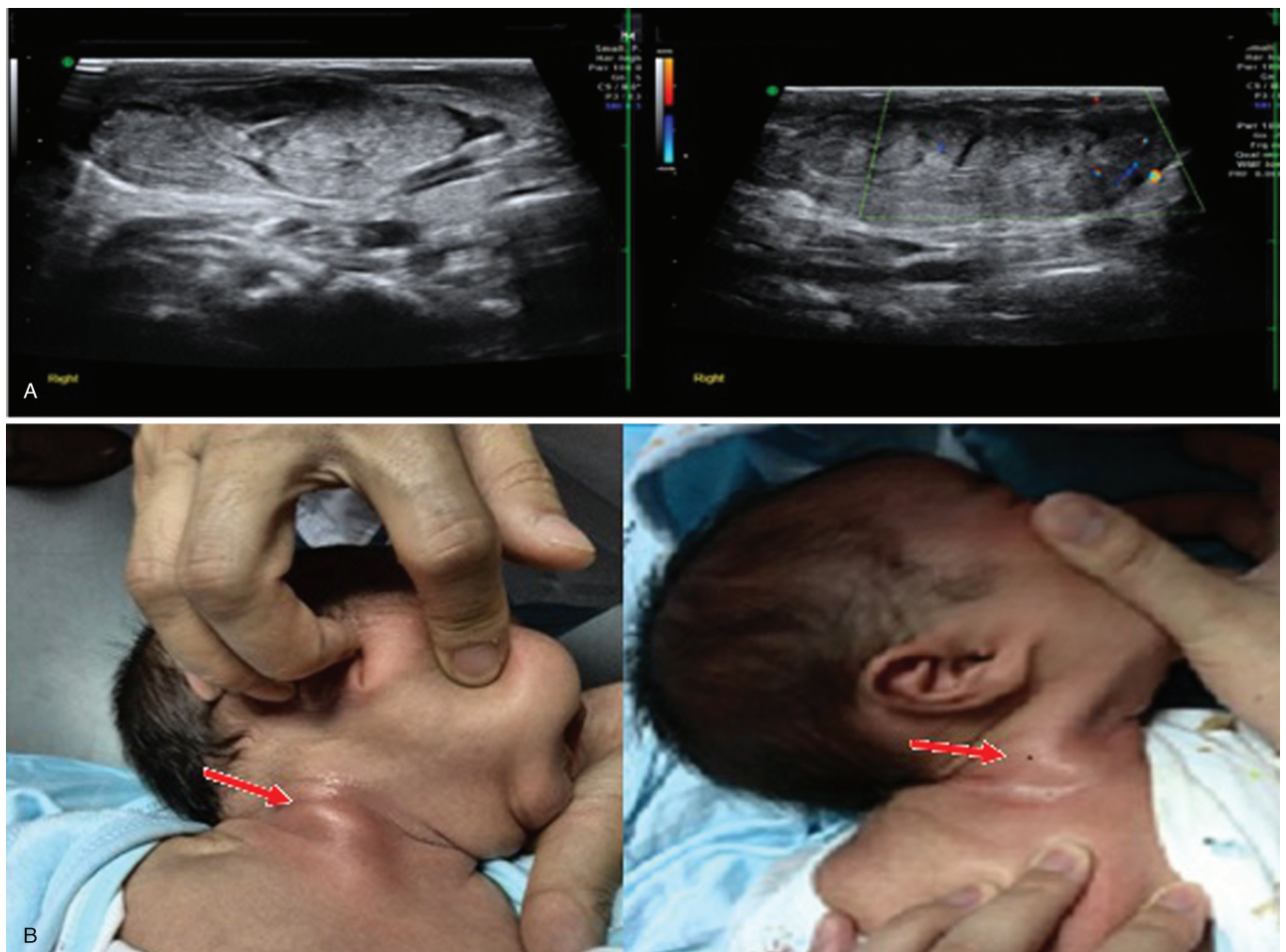


**Figure 5.** Twenty-two days after birth: ultrasound of the right (a) and left (b) sternocleidomastoid muscles (SCMs); photographs showing the infant has a smaller right hemiface and palpable mass in the right SCM (c).

#### 4. Discussion

CMT is a common musculoskeletal disorder in infants, which results from unilateral shortening or excessive contraction of the SCM. Infants with CMT usually present with an ipsilateral head tilt and the chin pointing contralaterally.

The range of motion in the neck is limited, and there may be an obvious facial asymmetry. Most children with CMT have been treated with conservative therapy;<sup>[13,14]</sup> if left untreated, progressive fibrosis and contracture of the SCM will occur.<sup>[1,15]</sup>



**Figure 6.** After 1 week of physiotherapy: ultrasound of the sternocleidomastoid muscle (SCM) before (left) and after (right) tearing (a); photographs of the SCM before (left) and after (right) tearing (b).





**Figure 7.** Photographs of the right (a) and left (b) sternocleidomastoid muscles after 6 months of physiotherapy.

Traditionally, the etiology of CMT has been explained by birth trauma and compartment syndrome theories, which suggested that the SCM was torn at birth, resulting in the formation of a hematoma and fibrous contracture.<sup>[3,12]</sup> However, the findings from the current study did not substantiate these theories. Among the 44 neonates with sCMT screened by ultrasonography, there were no signs of tearing of the SCM or hemorrhage.

In a previous study, we discovered undifferentiated mesenchyme-like cells in the ultra-microstructure of the CMT lesion tissue.<sup>[5]</sup> Myoblasts and fibroblasts at different stages of differentiation were also present,<sup>[6]</sup> suggesting that the pathogenesis of CMT was associated with maldevelopment of the fetal SCM. Likewise, the findings from the current study implied that maybe a postnatal factor was involved in the etiopathogenesis of SCM pseudotumor of infants (SCMPOI), which supports our theory stating that SCM differentiation and development may be responsible for the etiology of CMT.<sup>[5]</sup>

In our patient, the ultrasound examination on first day after birth showed that the affected SCM was thinner than the other one, and without any mass on the bilateral SCM. After a week, another ultrasound examination revealed that the diameter of the right SCM was approximately equal to the left SCM, which suggested that progressive changes occurred in the right SCM.

Since we continued to follow-up the patient with regular examination 15 days after birth, we found a mass within the right SCM, which restricted neck activity. Furthermore, the muscle was continually thickening. At 22 days after birth, the thickening was becoming bigger, and further limited the rotation of the neck. Similarly, the ultrasound revealed coincident echotexture of the SCM.

In this study, there was no evidence of neck or SCM injury in all 2564 newborn babies in the screening by physical examination. Furthermore, no evidence of neck injury in 44 1-day-old babies with sCMT was detected by ultrasound examination. The clinic investigation and ultrasound follow-ups revealed that SCMPOI developed postnatally in 1 patient. This phenomenon raised questions about the origin of the SCM mass in SCMPOI. As no obvious abnormality was found in SCM on the first day after birth, the definition of CMT as a congenital defect may be

inaccurate. On the contrary, the affected SCM mass and SCMPOI developed during the first 2 weeks after birth. These findings indicated that the SCM tumor developed postnatally. In addition, it is not known why a tumor developed in the SCM, or why the SCMPOI disappeared, resulting in an almost normal appearance of the neck. Based on our current and previous studies, SCM differentiation or maturation during embryogenesis could be disrupted, which may lead to remnants of mesenchyme-like cells in the SCM of the fetus. After birth, as the environmental changes, the mesenchyme-like cells grow, undergo differentiation and maturation, and then the mass in the SCM emerges, resulting in CMT.<sup>[5,6]</sup> We hypothesized that mesenchyme-like cells that remain static during embryogenesis in the SCM undergo asymmetrical division, differentiation, and proliferation after birth, and during growth and development of the infant. The remnant immature cells in the affected side of the SCM proliferate excessively and cause SCMPOI.

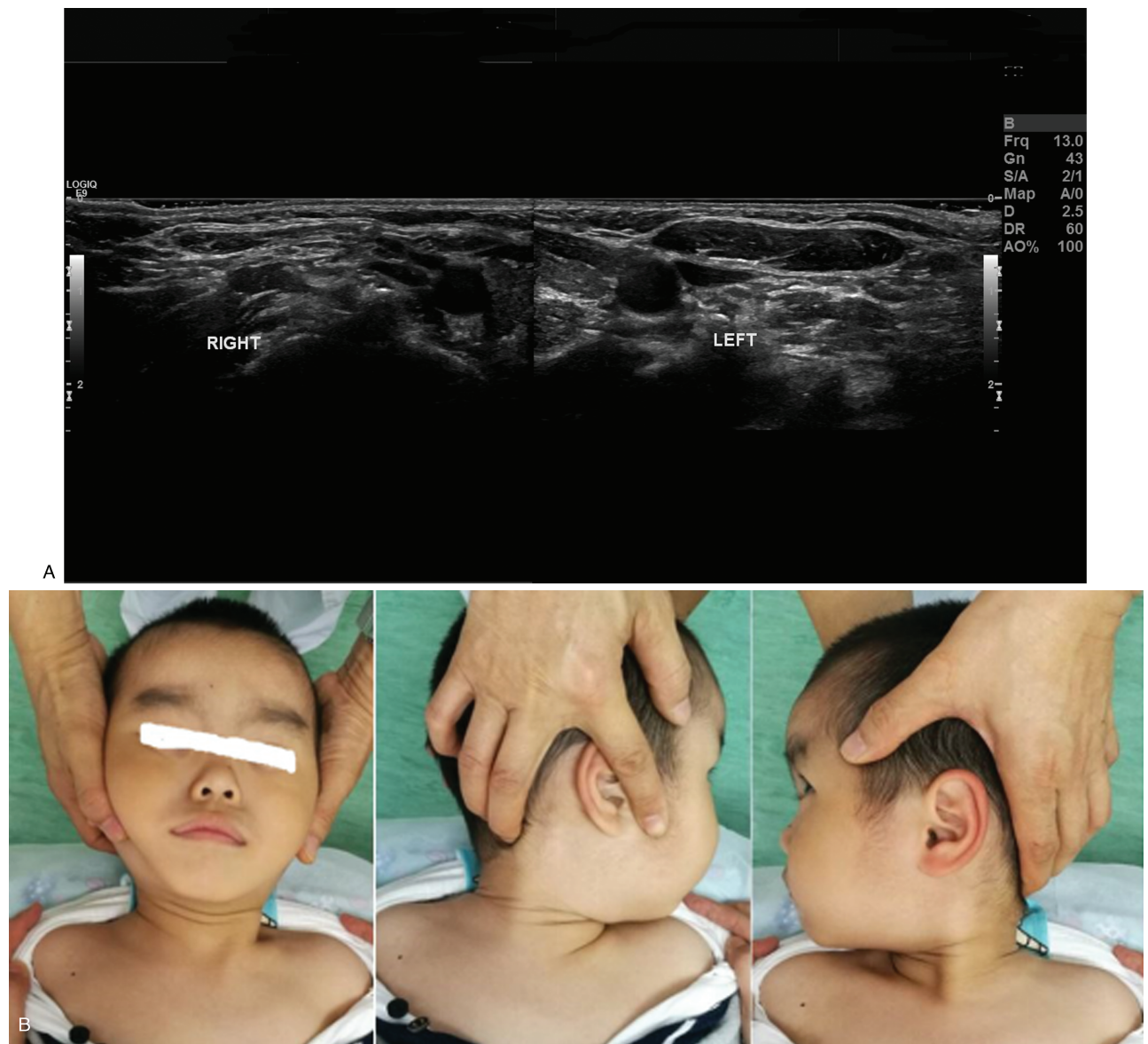
Although this case provided us with some new information to support the theory stating that the etiology of CMT involved a deficiency in the differentiation and development of the SCM, further studies are warranted to confirm these findings.

Notably, at each time point, the ultrasonography findings conformed to the clinical manifestations of CMT in our patient, indicating that ultrasound examination is an effective and safe method to diagnose CMT in infants. More importantly, it can be used to monitor recovery in infants with CMT who are undergoing rehabilitation. Hence, ultrasound can be used to efficiently assess therapeutic effects in CMT patients.<sup>[16]</sup>

Screening not only assisted doctors in making an early diagnosis, but it also provided an opportunity for regular treatment and avoided surgery in patients with CMT, thereby improving the prognosis. More importantly, with the promotion of screening, the awareness of congenital musculoskeletal abnormalities can be raised in pediatricians and parents.

This study had several limitations. First, due to insufficient resources, it was not possible to perform ultrasonography of the SCMs in every neonate. Secondly, not all parents were compliant with the study protocol; therefore, some infants with CMT may have been lost to follow-up. Besides, due to the nature of this





**Figure 8.** Three years after birth: sonograms of the right and left sternocleidomastoid muscles (a); photographs of the right and left SCMs (b).

retrospective study and the shortage of diagnostic standards for CMT, some unavoidable biases, such as ultrasound, selection, or reporting bias may be present in this study.

In conclusion, the findings in this clinical study and our previous studies suggested that prenatal and postnatal factors play important roles in CMT. Namely, the congenital muscular torticollis may be a developmental disease, and not a congenital disease.

#### Author contributions

**Data curation:** Shuaidan Zeng, Xin Qiu, Huanxiong Chen.

**Formal analysis:** Gen Tang.

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**Methodology:** Yu Tang.

**Project administration:** Sheng-ping Tang.

**Supervision:** Sheng-ping Tang.

**Writing – original draft:** Zhu Xiong, Shuaidan Zeng.

**Writing – review & editing:** Sheng-ping Tang.

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