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Congenital muscular torticollis: Diagnosis, management, and prognosis in the pediatric population☆

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A B S T R A C T

Congenital muscular torticollis (CMT) typically is due to unilateral shortening of fibrosis of sternocleidomastoid muscle. Timely diagnosis and management are essential for improving outcomes in children. Within CMT, there are several classifications with variable prognosis: postural, muscular, and sternomastoid tumor group. The diagnosis of CMT involves comprehensive physical assessment. Treatment of CMT typically involves conservative treatment that includes physical therapy, but can involve pharmacologic treatment with botulinum toxin injections, and at worst case, surgical treatment. Caregiver education is the key to successful treatment.

Introduction

Congenital muscular torticollis (CMT), also known as the twisted or wry neck, is a musculoskeletal anomaly presenting unilateral shortening or fibrosis of the sternocleidomastoid (SCM) muscle, resulting in ipsilateral cervical lateral flexion and contralateral rotation.[1] It is often detectable at birth or in the first few weeks of life, with the median age at the time of clinical manifestation being two months, and can persist until 1 year[2]

Relevance of early diagnosis and early intervention

Timely diagnosis and management are essential for improving outcomes in children with CMT.[3] Physical therapy (PT) applied in the first 1–3 months of life is associated with a better chance of complete resolution of limitation in cervical range of motion and prevention of secondary outcomes such as plagiocephaly and facial asymmetry.[4] Subsequently, delayed diagnosis can lead to long-lasting musculoskeletal deformities, prolonged therapies, and a greater likelihood of surgical intervention.[5]

Embryology and pathophysiology

Developmental anatomy of the sternocleidomastoid (SCM) muscle

The sternocleidomastoid is a prominent muscle located on the anterior side of the neck. It develops from the paraxial mesoderm and occipital somites during early gestation, appearing on the 14th day of gestation in animal models.[6] The inferior attachments of the SCM are split into the sternal head (medial) and clavicular head (lateral).[7] The sternal head attaches to the anterior surface of the upper aspect of the manubrium of the sternum and the clavicular head to the posterior-superior surface of the medial one-third of the clavicle.[7] The superior attachment is to the lateral surface of the mastoid process and the lateral one-half of the superior nuchal line on the occipital bone.[7] The SCM is primarily innervated by the spinal accessory nerve (cranial nerve XI) with some supplementary sensory (proprioceptive) information from the nerve roots from the mid and upper cervical plexus (C2–3).[6,7] Acting unilaterally, the SCM acts as a strong ipsilateral lateral flexor and contralateral cervical rotator. Bilaterally, the SCM acts as a strong flexor of the mid to lower cervical spine and mild extensor of the upper cervical spine, which promotes a forward head posture.[7] [Fig. 1]

Pathogenesis of CMT: fibrosis and shortening of SCM muscle

Congenital muscular torticollis is defined by pathological changes within the sternocleidomastoid (SCM) muscle, most notably unilateral fibrosis and muscle shortening. These structural changes are considered the primary contributors to limited flexibility, resulting in ipsilateral cervical lateral flexion and contralateral rotation.

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Theories on etiology: intrauterine crowding, birth trauma, vascular injury

While there is no singular cause for torticollis, there are several theories on etiology. The intrauterine theory proposes that abnormal positioning of the fetus within the uterus can lead to CMT. Large fetal size, breech position, oligohydramnios, or multiple gestations are all examples of crowding in the intrauterine space which could decrease fetal movement and force a fixed position of the neck.[1,8,] Another common theory is that birth trauma can cause physical tearing to the SCM, leading to fibrosis and the development of CMT.[8] Excessive twisting of the neck can occur during a difficult delivery or when there is a mismatch between the size of the fetus and the birth canal.[1] Additionally, the vascular theory elaborates on the birth trauma theory proposing that the trauma to the SCM can cause muscle ischemia and the development of a hematoma.[1] Therefore, the decreased blood supply to the SCM can also cause fibrosis and development of CMT.

Epidemiology

Approximately 0.3–2 % of children are diagnosed with CMT.[9–16] The incidence of CMT has actually risen in the past several years.[17] The rise is multifactorial including the Safe to Sleep campaign,[18–21] infant position equipment,[22] less frequent prone positioning for play, as well as cultural factors.[17] It is widely reported that CMT is more common on the right side compared to the left; bilateral cases do exist but are rare. There is a slight male preponderance as well as a predilection for firstborn children.[11,13,23,24] Breech presentation, assisted delivery, and intrauterine growth restriction are also thought to increase risk of CMT, specifically with the type involving fibrosis in the sternocleidomastoid.[8,25,] Craniofacial asymmetries (e.g., facial asymmetries, ear abnormalities, mandibular asymmetries, plagiocephaly) are present in nearly 90 % of children with CMT.[4,17] CMT may also be associated less commonly with other conditions such as brachial plexus injury, hip dysplasia, metatarsus adductus, and talipes equinovarus.[13,23,24]

Common types of congenital muscular torticollis (CMT)

Congenital muscular torticollis is clinically classified into three distinct types based on physical examination findings: Postural, Muscular, and Sternomastoid Tumor Group.[3] Diagnosis of the specific form of CMT is important for planning treatment and managing family expectations about the length of recovery.

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Postural Torticollis includes the absence of any palpable tightness or mass in the SCM.[4] This type is commonly regarded as the most benign, thus is more likely to be accompanied by environmental influences (e.g., intrauterine position).[26] Infants with Postural Torticollis generally have a mild presentation without muscular contraction, and they usually improve dramatically with conservative treatment (e.g., repositioning techniques and home exercise program)[27]

Muscular Torticollis is characterized by unilateral shortening of the sternocleidomastoid muscle, leading to measurable restrictions in passive cervical rotation and lateral flexion.[3] It involves more pronounced muscle tightness, often necessitating a more intensive physical therapy regimen. These cases typically require a structured treatment plan emphasizing targeted stretching to address soft tissue restrictions. Prognosis is generally favorable, particularly when intervention is initiated early in infancy[4]

Sternomastoid Tumor (SMT) Group is called fibromatosis colli or SCM Mass Type Congenital Muscular Torticollis (CMT). It is characterized by a palpable fibrotic mass that is palpable in the SCM muscle and results in limited cervical range of motion.[27] it is frequently noticed in first few weeks of life and is classified as the most severe and requires long-term therapy.[1] While the prognosis is generally favorable, outcomes are significantly improved when treatment begins early. Due to the presence of a fibrotic mass within the sternocleidomastoid muscle, these cases may exhibit a slower response to conservative interventions as compared to Postural and Muscular Torticollis[1] The tumor usually resolves between 4 and 8 months of age[2]

Clinical presentation and diagnosis

Typical physical findings

In addition to the classic posture of ipsilateral cervical lateral flexion and contralateral rotation, infants with CMT may demonstrate asymmetrical neck folds, facial asymmetry, positional plagiocephaly, hip dysplasia and decreased active cervical movement.[27] Persistent preference for looking in one direction during sleep or feeding may be an early parental concern prompting evaluation.

The diagnosis of CMT relies heavily on a comprehensive clinical evaluation. Detailed prenatal, birth and perinatal history is also important.[28] This would include questions regarding the presence of intrauterine constraint (fibroid), multiple birth, birth trauma (e.g., use of forceps or vacuum assistance), or prolonged labor.[8]

The diagnosis of CMT relies heavily on a comprehensive clinical evaluation.

During the physical examination of an infant with suspected congenital muscular torticollis (CMT), a comprehensive musculoskeletal and developmental assessment is essential. Clinicians should evaluate

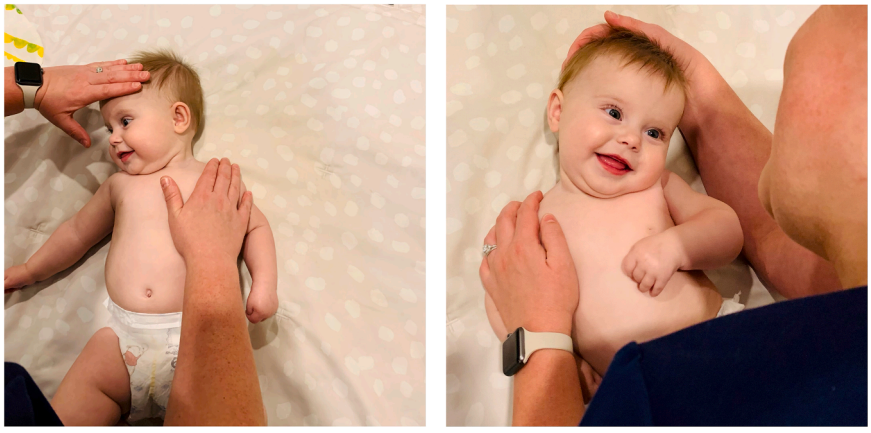

Fig. 1. Example of torticollis exercise, active stretching.

both passive and active cervical range of motion in lateral flexion and rotation to identify limitations in mobility.[5] Observation of head shape is equally important, as occipital plagiocephaly may indicate the infant's inability or reluctance to rotate the head toward the contralateral side.[5,27] For infants older than four months, bilateral head righting reactions in ventral suspension should be assessed to gauge symmetrical neck control.[5] Palpation of the sternocleidomastoid (SCM) muscle is critical for detecting tightness or a fibrotic mass, which may suggest the muscular or tumor subtype of CMT.[4,5] Additionally, evaluation of craniofacial asymmetry can provide insight into the chronicity and severity of postural imbalance.[5,27] Since ocular abnormalities, such as strabismus or impaired visual tracking, may mimic or contribute to torticollis, clinicians should monitor for abnormal eye movements and refer for ophthalmologic evaluation as needed.[27] Careful observation of the infant's postural preferences during spontaneous movement can help differentiate CMT from other neuromotor or positional conditions.[5] In addition to cervical and craniofacial assessments, examination should include screening for associated musculoskeletal conditions such as developmental dysplasia of the hip (DDH) and early signs of scoliosis. Hip instability or asymmetrical hip abduction may signal DDH, which is present in approximately 20 % of cases.[2] Likewise, in longstanding or untreated cases, spinal curvature should be monitored, as altered muscle tone and persistent head tilt may contribute to compensatory scoliosis.[29]

Role of imaging

Although CMT is primarily diagnosed through clinical assessment and evaluation, imaging is warranted in specific cases. To assess SCM musculature, ultrasound is the preferred modality for confirming fibrosis or excluding other soft tissue anomalies.[8] Radiographs of the cervical spine may be indicated if there is limited motion in cervical rotation and vertebral anomalies or atypical findings are suspected.[24] MRI is reserved for unusual cases when neurological, skeletal, or soft tissue abnormalities beyond muscular causes are being considered.[10] However, Imaging is not necessary for routine, typical, and uncomplicated presentations.[27]

Differential diagnosis

A thorough and systematic clinical assessment is essential for distinguishing congenital muscular torticollis from other potential causes of abnormal head posture, such as ocular, neurologic, or gastrointestinal[24]

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Ocular torticollis results from visual disturbances such as strabismus or nystagmus, prompting the child to adopt a compensatory head tilt to optimize visual alignment. Unlike congenital muscular torticollis, this form typically lacks true muscular tightness, although secondary muscle shortening may develop over time if the abnormal posture persists.[27] An ophthalmologic evaluation is essential to confirm the diagnosis and guide appropriate management.[27]

Benign paroxysmal torticollis (BPT) is a rare, self-limiting disorder of infancy characterized by recurrent episodes of head tilt that often alternate sides.[30] Unlike congenital muscular torticollis, infants with BPT typically demonstrate side-to-side variability in head posture. These episodes may last for hours, days, or even weeks, followed by intervals of normal head alignment. BPT has been linked to an increased risk of developing migraine disorders later in childhood.[30] Recognition of this distinct clinical pattern is essential, and referral to neurology is recommended for evaluation and long-term follow-up, especially if migraine symptoms emerge[31]

Sandifer syndrome is a movement disorder associated with gastroesophageal reflux disease (GERD), characterized by intermittent dystonic posturing of the head, neck, and trunk—often with sudden episodes of neck extension or back arching.[32] These movements can be mistaken for torticollis or seizure activity. Diagnosis relies on careful correlation between symptom onset and feeding history, along with confirmation of underlying GERD. Effective management of reflux symptoms typically results in resolution of the abnormal posturing.[32]

Management and treatment approaches

Conservative management

Physical therapy is the primary conservative intervention for CMT. It is highly effective when initiated in early infancy. Comprehensive physical therapy for CMT includes interventions to address passive and active range of motion of the neck and trunk, development of symmetrical movements, environmental adaptations, and caregiver education.[5]

If there are a range of motion restrictions, it is imperative to initiate a stretching program to target tight SCM. This includes both passive stretching and active stretching. Passive range of motion is when the therapist or caregiver gently moves the baby's neck through the full range of motion, gradually improving range of motion over time.[33] Stretches can be completed in a variety of positions depending on the child's tolerance including supine, side-lying, prone, and supported sitting. Passive cervical rotation should be completed with stabilization of the shoulder and gentle overpressure towards the contralateral side (image 1). Passive lateral cervical flexion should be completed with stabilization of the shoulder while gently bringing the ear towards the contralateral shoulder. (Image 2). Active range of motion exercises encourage the child to move their own neck, through age-appropriate developmental play.[33] For an infant, this can be facilitated with visual or auditory tracking towards a parent or toy (Fig. 2).

Simple environmental modifications can provide opportunities to increase the active range of motion of the neck and promote positive changes to head shape. Parents should consider what position their child is in throughout the day and reposition them to allow the affected muscle to lengthen. Caregivers should consider rotating the side of the crib they place the child's head towards or pick them up from. Families should aim to limit the amount of time their baby is in a container that can restrict movement such as swings, bouncy chairs, and car seats. Babies should be repositioned throughout the day to allow for skills to develop in supine, side-lying, and prone. The American Academy of Pediatrics recommends a total of 30 min to 1 hour of supervised "tummy time" by 2–3 months of age.[34] This can start in multiple short bouts throughout the day and build up as the child grows. Modified prone on the parent's chest or an inclined surface such as a boppy pillow can help young infants be more successful in tummy time as they gain head control.

Caregiver education on a clear home exercise program is critical for successful treatment of torticollis. Caregiver education should include demonstration of the exercises with an opportunity for the parent to practice with the guidance of a physical therapist.[33] Home exercise programs can include pictures, written instructions, and/or videos depending on the family's preferred learning style. Families should be encouraged to build the exercises into their daily schedule such as during diaper changes and bedtime routines to improve consistency and adherence. Providers should set goals with the caregivers and re-assess progress over time to adjust the exercise program as needed.

Caregiver education on a clear home exercise program is critical for successful treatment of torticollis.

Chemodenervation with botulinum toxin

When therapy and the home exercise program are insufficient, botulinum toxin injections may be considered. The aims of botulinum toxin injections are to improve the ability to stretch on the side of muscle shortening while also allowing strengthening of the opposite side.[23, 27, 4] The primary targets for the injections are the ipsilateral

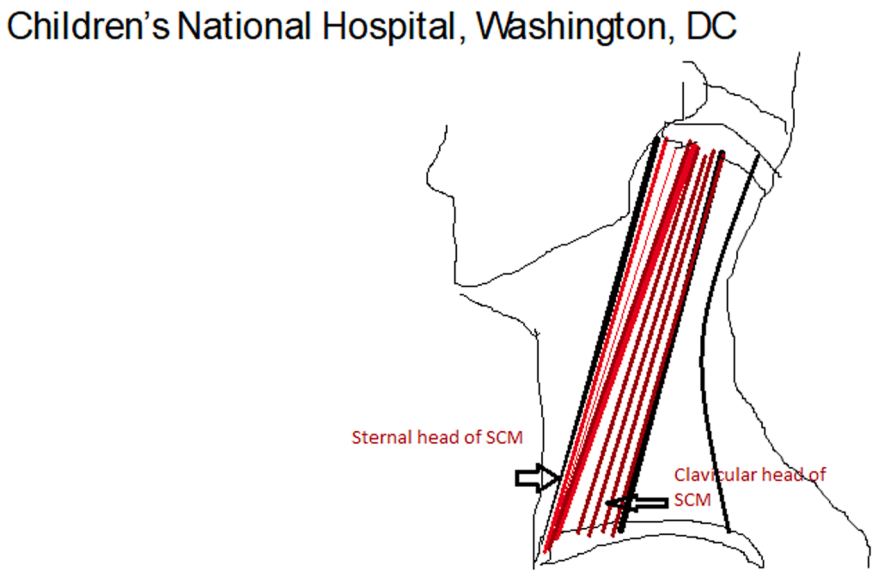

Fig. 2. Anatomy depiction of SCM muscle. Reference source: Fehrenbach, M. J., 2012.

sternocleidomastoid, upper trapezius, and scalene muscles.[23] It is important to note that none of the commercially available botulinum toxin preparations (onabotulinumtoxinA, abobotulinumtoxinA, incobotulinumtoxinA, and rimabotulinumtoxinB) have official FDA approval in the United States for the treatment of torticollis in pediatric patients.

Dosing is variable. A prospective study by Limpaphayom et al. in 2019 examined the use of botulinum toxin injections in 39 children aged 13.5–27.5 months.[35] Weight-based dosing of 2–3 units/kg was used and divided between 2–3 sites along the sternocleidomastoid.[35] A recent retrospective study by Sinn et al. of 134 children aged 6–24 months over a 10-year period reported dosing of 15–30 units per muscle (ipsilateral sternocleidomastoid, upper trapezius, and/or scalene muscles).[23] In this study, muscle selection was based on where the patient had limitations in motion, meaning that the sternocleidomastoid was injected if rotation was limited, and the upper trapezius and scalenes were injected if lateral flexion was limited.[23] Of the 134 children, 61 % had a successful outcome as determined by the authors' criteria for achieving 45 degrees of active lateral flexion and 80 degrees of active cervical rotation.[23] Two additional retrospective studies described the benefits of botulinum toxin – Oleszek et al. found that 74 % of 27 children demonstrated improvement[36] and Joyce et al. reported on improvement in 93 % of 15 children.[37] Of note, there were several differences between the above studies including but not limited to inclusion/exclusion criteria, dosing, and outcome measures. In 2020, a meta-analysis of botulinum toxin uses in torticollis by Qui et al. including 10 articles and 411 patients was published.[38] They reported that their study revealed botulinum toxin was 84 % effective.

Risks of botulinum toxin injections are rare but can be serious and even life-threatening. Possible side effects of the injections include pain, bleeding, bruising, and erythema. More serious potential risks include dysphagia, dyspnea and infection. In their meta-analysis, Qui et al. found the adverse reaction rate to be 1 % with dysphagia and erythema at the injection site the being most commonly reported.[38] Importantly, performing botulinum toxin injections does not preclude the need for therapy and a home exercise program. In fact, botulinum toxin injections are almost always coupled with therapy and continuation of a home exercise program.

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Surgical intervention

When therapy, home exercise program, bracing, and/or injections are insufficient, then surgical intervention may be considered. Surgery is targeted at the fibrotic tissues.[23] It is recommended that surgical intervention be done when the child is between 1–3 years old.[39, 29] Waiting until after 3 years old may increase the risk of scoliosis.[39, 29] Traditionally surgery involves either unipolar or bipolar release of the sternocleidomastoid. Since there have been advancements in minimally invasive surgical techniques, a few different endoscopic surgical techniques have been described including transaxillary subcutaneous endoscopic release.[39–46] There are data to suggest that endoscopic surgery is done in children from 1–180 months with even some data in children over 16 years old.[39, 47] Indications for surgical intervention include passive or active limitations in rotation or/and lateral flexion ranging from 10–15 degrees in children >1 year old, sternocleidomastoid mass in children >7 months old, and children who have shown insufficient response to conservative treatment.[48, 17] Surgery generally well tolerated without significant side effects. However, in addition to the risks of surgical procedures themselves, there is also the risk of the recurrence of torticollis. Interestingly, recurrence can occur with or without reconnection of the sternocleidomastoid muscle, such as residual fibrosis, asymmetrical muscle function, or inadequate postoperative rehabilitation—may contribute to the recurrence of torticollis.[48] Unipolar release has a reoccurrence rate of 5.5 % to 7 %; and Bipolar release is approximately 2 % to 2.9 %[48]

Conclusion

Congenital muscular torticollis (CMT) is a relatively common musculoskeletal condition of infancy, most often resulting from unilateral shortening of the sternocleidomastoid muscle, leading to characteristic tilt and contralateral rotation. CMT has an excellent prognosis when conservative treatment (physical therapy) is initiated at a young age. For an infant with CMT that begins physical therapy intervention before 1 month of age, the prognosis for a good outcome is 99 %..[5] Evidence has shown that good outcomes, defined as no head tilt and full passive cervical rotation, decline as the age of onset of treatment increases. .[5] Factors associated with a good outcome include participation in physical therapy and a home exercise program, younger age at onset of intervention, CMT severity including less difference between sides of cervical rotation PROM or SCM muscle thickness. .[5] Delayed treatment can cause persistent asymmetry and fibrosis of the muscle and require

more invasive treatment. Surgery may be considered after one year of age with failure to respond to physical therapy and physical changes to the facial and bony structures on the tight side. .[1] Therefore, early recognition of the signs of torticollis and referral to a local torticollis clinic or physical therapist is essential. Early intervention has been shown to improve clinical outcomes and shorten episodes of care which reduces the cost of care and burdens on families.[5]

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