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Review

DIAGNOSIS AND HABILITATION OF CONGENITAL MUSCULAR TORTICOLLIS: A NARRATIVE REVIEW

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SUMMARY – Congenital muscular torticollis (CMT) is one of the most common congenital musculoskeletal deformities, and is defined as tilted head position with hypertonic sternocleidomastoid muscle and limited cervical spine mobility. Incidences up to 16% have been recorded, more often in male children. Clinical features are characterized by an inclined position of the head on the diseased side and a gaze directed towards the healthy side, deformities such as plagiocephaly, and various complications. In order to prevent this, it is recommended to screen all infants up to four months of age. Diagnosis is usually made through history and physical examination. CMT treatment is carried out conservatively, and the habilitation program is created individually, depending on the degree of CMT. The habilitation program includes neck stretching exercises, neck muscle strengthening exercises, motor activities that encourage a symmetrical movement pattern, adaptation to the environment, and education and support of parents or guardians to ensure a daily, intensive home program. Early treatment is one of the main prognostic factors on which the outcome and duration of treatment depend. As this is the only prognostic factor that we can act on, our goal is to speed up the diagnosis and therapy implementation until the cure.

Key words: *Congenital torticollis; Infant; Physical therapy modalities*

Introduction

Congenital muscular torticollis (CMT) is one of the most common congenital musculoskeletal deformities. It is characterized by an inclined position of the head resulting from contracture of the sternocleidomastoid muscle (SCM). In more serious cases, plagiocephaly, or deformation of the skull bones also occurs. The diagnosis is based on a well-taken history and detailed physical examination. Only in some cases does the need of further diagnostic processing with various imaging methods appear. The vast majority of infants and children with CMT achieve good to excellent results with early conservative treatment, i.e., physical therapy (habilitation)[1,2]. As the age of the child at the start of therapy is one of the main prognostic factors on which

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the success of treatment depends, early treatment is the main goal we can act on. This thought prompted us to write these guidelines to facilitate the process from identifying children with CMT to curing them. The aim of these guidelines is to present the scientific opinion on this topic in a simple and systematic way, as well as our own experience, and thereby help all health care professionals who will encounter children with CMT.

Methods

A thorough literature review was conducted on October 28, 2022 for inclusion in this narrative review. Medline and Google Scholar databases were searched with the following keywords, including wildcards as appropriate to maximize search results: congenital torticollis, physical therapy, and diagnostic procedure. The bibliographies of each pertinent article, as well as those in review articles that may have included appropriate information, were also reviewed for additional literature. All articles discussing CMT were reviewed. Duplicates were removed and abstracts were screened for relevance according to our clinical experience, which resulted in a total of 27 articles used for this narrative review; there were no disagreements between reviewers.

Definition and Epidemiology of Torticollis

The clinical entity torticollis was created by fusion of two Latin words, *tortum collum*, translated as crooked (twisted) neck. Therefore, torticollis is defined as an inclined position of the head with hypertonus of the SCM and limited mobility of the cervical spine[3]. It is usually not considered a diagnosis, but a manifestation of a number of underlying conditions.

Torticollis is mainly manifested in the neonatal period as CMT. The latter is one of the three most common deformities that children are born with, along with hip dislocations and foot deformities[3]. Previous studies state that the incidence of CMT is below 1%, but today new data appear with a higher incidence, 0.3%-3.92%, while some studies stating it as the most common congenital musculoskeletal deformity, with an incidence of up to 16%[4,5]. The frequency of occurrence is higher in male children and on the right side of the neck.

There are numerous risk factors that increase the incidence of torticollis. Newer methods of treating infertility have increased the number of multiple pregnancies, which is considered one of the main risk factors for the development of CMT[6]. An important risk factor is the number of pregnancies of the mother; torticollis occurs more often in primiparous women, as well as in pregnancies with a reduced volume of amniotic fluid. Likewise, the incidence of CMT is higher during a traumatic birth, with the use of forceps during birth, and with intrauterine exposure to opioids[7] .

Classification and Pathophysiology of Torticollis

Torticollis can be the result of numerous conditions and can appear at any age, therefore its classification has always caused divisions among authors. Depending on the dynamics, some authors divide it into non-dynamic (non-paroxysmal, such as CMT) and dynamic (paroxysmal, such as benign paroxysmal or cervical dystonia)[8]. Traditionally, torticollis is divided into congenital and acquired, and further depending on etiology.

We have chosen the traditional classification because it emphasizes the importance and frequency of occurrence of congenital torticollis. Despite the fact that the term congenital torticollis includes various prenatal and perinatal causes of a crooked (twisted) neck, in the literature this term is often equated with the term CMT because it is by far the most common form of congenital torticollis.

Congenital muscular torticollis is typically categorized into three types, as follows: postural, muscular without a fibrous mass, and muscular with a fibrous mass. Patients with CMT with a tumor have a palpable mass, those without a tumor have increased muscle tone of the SCM without a palpable mass, while in those with postural CMT we find neither a tumor mass nor increased muscle tone[9].

Different pathophysiological mechanisms leading to the mentioned conditions have also been described, mainly the process of ischemia and the consequent fibrotic changes. Most often, ischemia occurs when blood vessels are compressed due to intrauterine malposition of the fetal head. Also, depending on the time of occurrence, there is a logical sequence, i.e., if the ischemia occurred earlier, CMT will appear with the fibrous mass due to sufficient time for the development of fibrotic changes and the formation of a hard palpable mass. If the ischemia lasts for a shorter time, there is a higher probability of postural

CMT without the fibrous mass and without muscle hypertonus because there was no major muscle damage and thus the contracture is of a lower degree. Birth trauma is also described, mainly in cases of premature or prolonged birth. Stretching the neck during childbirth leads to tearing of muscle fibers and bleeding in the muscle itself. Blood collects within the fascia and leads to a local increase in pressure, causing ischemic muscle injury and, consequently, to the replacement of muscle tissue with fibrous tissue. Also, a developmental disorder in the form of an imbalance in the development of myofibrils and connective tissue of muscles leads to fibrosis and muscle shortening and the consequent occurrence of torticollis. If CMT occurs with congenital hip dysplasia, a hereditary mechanism is suspected[4,10].

Other causes of congenital torticollis include vertebral anomalies such as congenital scoliosis, unilateral atlantooccipital fusion, Klippel-Feil syndrome, various neurological and ophthalmological disorders, paroxysmal torticollis that changes sides, and unilateral lack of the SCM muscle[11,12].

Clinical Features of CMT

In order to establish the diagnosis correctly and as early as possible, it is essential to define clinical indicators of CMT. Clinically, we distinguish early and late signs and complications.

Early signs

The earliest signs of CMT are noticeable immediately after birth. A newborn baby performs lateral flexion of the head ipsilaterally, i.e., towards the diseased side, and rotates the head contralaterally, looking towards the healthy side. Gradually there is development of limitation of mobility in the neck. Initially, rotation towards the diseased side is reduced, and then there is a limitation of lateral flexion towards the healthy side[1]. Two to three weeks after delivery, a painless palpable mass, shortening of the SCM muscle and its hypertonus appear. A palpable mass can grow for two months until it reaches the approximate size of an almond when it begins to recede, and it can disappear completely by the eighth month of life[13,14]. Thickening of the SCM muscle often occurs in the middle and distal third, but it is not always present. Slowly, drooping of the head, i.e., plagiocephaly begins to be noticed[3].

Late signs

Due to unilateral muscle shortening, a child with CMT prefers to sleep in a prone position with the face facing the healthy side. This position causes asymmetrical forces on the skull, which affects the development and remodeling of facial bones. The result of these processes is, in addition to plagiocephaly, hemihypoplasia of the facial bones, which manifests itself in facial asymmetry, usually around the third month of life. Asymmetry of the face is manifested by developmental stagnancy of the diseased side, the eyebrow on the diseased side is placed lower than on the healthy side, and the ear is flattened.

Complications

Complications of CMT occur very quickly, so early recognition and prompt treatment are crucial. It is difficult for the child to breastfeed due to the inability to take the correct position. Asymmetry of the head and face persists as a constant finding. Through the child's motor development and development of balance in positions such as sitting and crawling, compensatory mechanisms from different systems are reflected in distortion of the cervical spine in the frontal plane, that is, in the development of scoliosis of the cervical and thoracic spine. In some children, visual and hearing disorders are also observed[3,13]. Some authors also describe the importance of the influence of the SCM muscle on the development of large muscle groups, especially trunk muscles. Due to the weakness of large muscle groups, these children are less often exposed to stimulation from the environment, which consequently leads to a weaker development of cognitive functions and fine motor skills, i.e., overall neuromotor development[1,4].

Diagnosis

The clinical guideline of the American Society of Physical Therapy strongly recommends examination and screening of all infants in the first 3 to 4 months of life in order to detect children with CMT on time[16]. First of all, diagnosis is based on the history obtained from the parents or guardians, and physical examination. During history taking, it is important to pay attention to the details related to the preferred positions of the child during sleep, and the birth of the child. Children born by complicated delivery, those with larger birth weight, children born in breech position,

and children born from multiple pregnancies, especially those positioned low in the womb, have an increased risk. Furthermore, important data are decreased fetal movements, lack of amniotic fluid and retention of the fetus in the same position, i.e., the back turned to one side. Mothers often state that the fetus did not change the position during pregnancy and that fetal back was always turned to one side. During physical examination, we should look for early and late signs, and in the case of late diagnosis, complications. Already during the first postnatal examination of newborns (up to the first three days of life), screening for CMT can be performed by checking passive rotation (the chin turns towards the shoulders) and lateral flexion (the ear approaches the shoulder) while the child is in supine position. It is important to note that the range of motion differs between the children aged up to three years and those aged over three years. In older children, the range of passive rotation is 90^0 (chin to shoulder), whereas in children up to three years of age it is 100-110^0, i.e., 10-20^0 behind the shoulder. The range of passive lateral flexion in older children is around 45^0, whereas in children up to three years of age it varies between 65^0 and 75^0[4]. If deviation/decreased range of motion in the neck is noted, the child is immediately referred for physical therapy to ensure a comprehensive and supportive habilitation program. In some cases, additional diagnostic processing is indicated, e.g., laboratory blood and urine testing (more in terms of excluding other diseases), ultrasound of the neck region soft tissue, and x-ray images of cervical spine (anteroposterior and lateral projection)[3,6]. Sometimes additional diagnostic processing is required, e.g., x-ray tomograms, functional x-ray images and imaging in oblique projection, especially imaging (transoral) of the odontoid process. Since children with muscular torticollis also have congenital hip dysplasia in 20% of cases (every fifth child), it is necessary to take an x-ray of the hip[3].

A comprehensive examination by a pediatric physiatrist includes assessment of the child's entire musculoskeletal system, classification of the CMT degree (Table 1), and assessment of parenting skills related to feeding, positioning, assessment of thorough understanding of the potential factors contributing to asymmetric positions. In addition, non-muscular causes of asymmetry and conditions associated with CMT are checked.

Table 1. Grading of congenital muscular torticollis (CMT). The values entered in the table were obtained through many years of experience in working with children with CMT

Degree	Clinical features
0 – no CMT	In the supine position, the child maintains symmetrical head position, full active and passive range of motion
1 – mild form	In the supine position, the child has asymmetric head position, full active and passive range of motion
2 – moderate form	In the supine position, the child has asymmetric head position, active range of motion is reduced by 1/3 of the physiological amplitude, full passive range of motion
3 – severe form	In the supine position, the child has asymmetric head position, active range of motion is reduced by up to 1/2 of the physiological amplitude, passive movements are terminally reduced
4 – very severe form	In the supine position, the child has asymmetric head position, active range of motion is reduced by more than 1/2 of the physiological amplitude, passive range of motion is reduced by 1/3 or more of the physiological amplitude

Treatment

After thorough medical history, physical examination and assessment of the child, a decision is made on the most effective treatment. Treatment of CMT is most often carried out conservatively. The habilitation program is created individually according to the patient's clinical status. The habilitation program includes neck stretching exercises, neck muscle strengthening exercises, motor activities that encourage a symmetrical movement pattern, environmental adaptation, and parent or guardian education and support to ensure a daily, intensive home program[17].

There is strong evidence that earlier initiation of physical therapy is more effective than later initiation of physical therapy. Beginning physical therapy before the first month of the child's age results in a normal range of motion in the neck within 1.5 months in 98% of infants with CMT. In infants with CMT, starting therapy after one month of age extends the duration of treatment to 6 months, and starting therapy after 6 months of age in infants most often results in a duration of treatment of 9 to 10 months with progressively less success in achieving normal range of motion in the neck[2,18].

Education

Some studies point to the need to educate parents about CMT before the birth of a child because in this way, they will participate in the treatment in a better way, which will prevent the development of asymmetry and clinical features of CMT. An educated parent can spot postural asymmetry and the first signs of CMT in time, which significantly contributes to the success and duration of treatment[19].

Future parents and parents of newborns should know how to follow the child's development, know the importance of children's play in the prone position in which the child should be when awake (three or more times a day). Likewise, it is necessary for parents to stimulate symmetrical motor development patterns of movement, and in case they notice postural asymmetry, they should immediately seek help of a professional[2].

Placing the child in the prone position while awake significantly prevents the development of postural asymmetry, and prone position of the child activates neck muscles and enables proper development of the skull bones[20,21].

When postural asymmetry or CMT has already developed, good parent education will speed up the trip to the pediatrician, and thus physical therapy will be started in a timely manner[19] .

Kinesiotherapy

The physical therapy program for the treatment of CMT is comprehensive and focuses on five essential components: 1) passive neck and trunk range of motion exercises; 2) active neck and trunk range of motion exercises; 3) development of active symmetrical movement patterns; 4) environmental adaptation; and 5) educating parents or guardians on how to conduct therapy at home so that it becomes part of their daily routine[17].

The Hospital for Special Surgery guidelines for non-operative treatment of CMT based on the latest 2018 Academy of Pediatric Physical Therapy guidelines and clinician experience are categorized into 3 stages, as follows:

- the first phase (weeks 1-8) is aimed at increasing the passive and active range of motion in the neck, increasing the time the child keeps the head in the central line and encouraging a symmetrical pattern of gross and fine motor skills. It is necessary to adapt the environment and educate parents/caregivers about carrying out exercises at home, proper positioning and monitoring of the child
- the second phase (weeks 9-16) is aimed at achieving full passive and active range of motion in the neck, control of the head in the central, i.e., medial line 95% of the time and 50%-75% of the time in other positions, and strengthening neck muscles to overcome gravity in different positions
- at the end of the third phase (weeks 17-24), the child is expected to achieve full active and passive range of motion in the neck, lateral flexion and rotation, age-appropriate antigravity neck strength, and the ability to maintain the head in the midline 95% of the time in all age-appropriate developmental positions.

Domicile implementation of therapy includes proper handling of the child in different positions, thereby encouraging turning the child's head in the desired direction, for example, during breastfeeding or feeding, changing clothes, and staying in the prone position[5]. This comprehensive physical therapy program improves outcomes as the therapist acts as partner to the family to determine the best ways to integrate exercises and positioning activities into the

infant's and family's daily routines. Re-evaluation is required 3-12 months after the end of therapy.

Passive stretching of the SCM muscles before the age of 12 months is the most effective physical therapy method[22]. The range of motion in the neck, in addition to passive stretching of the SCM muscle, is also increased by massage of hypertonic neck muscles and subcutaneous tissue, joint mobilization, myofascial relaxation, and therapeutic taping.

Massage is one of the widely accepted treatment techniques. By manipulating the muscles and soft tissue of the patient's neck (including gentle, continuous and vigorous application of force to the affected area), relaxation is achieved. Studies have shown that massage can effectively relieve muscle spasms in CMT patients and increase the range of motion in the neck. Massage is widely used during the treatment of CMT, it is painless, pleasant, and well tolerated by patients[23].

Thermotherapy

Heat treatment or thermotherapy is effective in the form of superficial heat modalities, and it is applied before stretching the muscles (heat and stretch) in order to achieve the best possible extensibility of soft structures, and predominantly light therapy and paraffin are used[24].

Orthoses

Spinal orthoses (soft) have their place in the conservative treatment of CMT, and are most often used to fix the effect achieved by active and passive exercises. The orthosis directs movement, prevents wrong movement and position, and positions and maintains the effect obtained by kinesiotherapy or surgery.

Surgical treatment

Previous research has shown that persistence of CMT despite conservative treatment after the child is one year old requires surgical treatment. The operation is most often performed between 12 and 18 months of age of the child after conservative treatment has failed. Operative treatment includes relaxation of the sternoclavicular part of the muscle by tenotomy. In rare cases, when there is an undoubted failure of conservative treatment and a rapid progression of disease complications is noticeable (facial asymmetry, etc.), operative treatment is applied to children at the age of six months[3].

After surgical treatment, facial asymmetry will regress over a longer period of time, and patients with more severe facial asymmetry have a better potential to later achieve facial symmetry, which is probably a consequence of earlier intervention[25].

Duration of treatment

Physical therapy is stopped when infants meet these five criteria: 1) cervical passive range of motion is within 5° in relation to the unaffected side; 2) the child presents symmetrical motor patterns of movement; 3) motor development is appropriate for age; 4) head tilt is not observed during static or dynamic positions; and 5) parents or guardians are trained to monitor further (future) motor development of the child[17].

Reviewing the current literature on CMT, we found three prognostic factors significantly associated with longer duration of treatment, worse outcomes, and increased risk of requiring surgical treatment. Those three prognostic factors are: 1) presence of a fibrous mass in the SCM; 2) a more serious deficit in the passive range of motion of the cervical spine compared to the healthy side; and 3) higher age of the child at the beginning of treatment[1].

Untreated torticollis

Reports of untreated CMT are rare, but the literature mentions 'unresolved' or recurrent CMT in older children or adults, who later undergo botulinum neurotoxin injections or surgery[26,27]. The frequency of spontaneous healing is not known, there is no method that could be used to assess in which direction CMT will move, i.e., whether it will develop milder, more severe or stubborn forms.

Conclusion

Congenital muscular torticollis is one of the three most common deformities that children are born with. Considering the increasing incidence of CMT in recent times, there is a need of greater education of parents and experts, so that they can recognize it in time and start treatment in a timely manner. The ultimate goal is to optimize outcomes for infants with CMT by shortening the duration of treatment, reducing family burden and medical costs. In order to recognize children with CMT in time, it is recommended to screen all infants up to 4 months of age, and for primary pediatricians to collect important history data

on risk factors for CMT. Early conservative treatment with kinesiotherapy in three phases achieves significant success and reduces the need of further surgical treatment. Therefore, early intervention is of utmost importance for more effective treatment, prevention of consequences, and minimization of health care costs, and we hope that these guidelines will be a guiding thread towards that goal.

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Sažetak

DIJAGNOSTIKA I HABILITACIJA KONGENITALNOG MIŠIĆNOG TORTIKOLISA: NARATIVNI PREGLED

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Kongenitalni mišićni tortikolis (KMT) jedan je od najčešćih kongenitalnih muskuloskeletnih deformiteta, a definira se kao nagnut položaj glave uz hipertonus mišića sternokleidomastoideusa te ograničenu pokretljivost vratne kralježnice. Zabilježene su incidencije sve do 16%, a javlja se češće u muške djece. Kliničku sliku obilježava nagnut položaj glave na bolesnu stranu i pogled usmjeren prema zdravoj strani, zatim deformiteti poput plagiocefalije, a naposljetku i razne komplikacije. Kako bi se to spriječilo, preporuča se provoditi probir sve dojenčadi do 4. mjeseca života. Dijagnoza se obično postavlja pomoću anamneze i fizikalnog pregleda. Liječenje KMT provodi se konzervativno, a habilitacijski program se izrađuje individualno, ovisno o stupnju KMT. Habilitacijski program uključuje vježbe istezanja vrata, vježbe jačanja vratnih mišića, motoričke aktivnosti kojima se potiče simetričan obrazac kretanja, prilagodbu okoline te izobrazbu i potporu roditelja ili skrbnika kako bi se osigurao svakodnevni, intenzivni kućni program. U terapiji se primjenjuje i masaža, termoterapija, ortoze, a ponekad i kirurško liječenje. Rano započeto liječenje jedno je od glavnih prognostičkih čimbenika o kojima ovisi ishod i duljina trajanja liječenja. Kako je to i jedini prognostički čimbenik na koji se može djelovati, ovim smjernicama želimo ubrzati i precizirati postavljanje dijagnoze i provođenje terapije sve do izlječenja.

Ključne riječi: *Dojenčad; Kongenitalni tortikolis; Modaliteti fizikalne terapije*