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Congenital torticollis in an infant

Wrodzony kręcz szyi u niemowlęcia

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Abstract

Introduction: Fibromatosis colli is an uncommon congenital infantile tumour occurring in 0.4–1.3% of live births. They are non-malignant, however early detection and treatment are crucial. This is because, if left untreated, fibromatosis colli tumours can cause permanent disfigurements to the skull and spine of the infant. The main aetiology that causes congenital fibromatosis colli is believed to be muscle injury, either intrauterine or during childbirth. A common symptom of congenital fibromatosis colli is head tilting toward the affected side. **Case report:** This case report discusses the management of an 82-day-old infant with fibromatosis colli. **Discussion:** Clinical examination including palpation and examination of the range of movement of the sternocleidomastoid muscle is the key to diagnosing fibromatosis colli. Early physiotherapy represents the first-line treatment in affected infants.

Keywords: fibromatosis colli, torticollis, congenital infantile tumour, sternocleidomastoid muscle

Streszczenie

Wprowadzenie: *Fibromatosis colli* (fibromatoza szyi) jest rzadkim wrodzonym guzem wieku niemowlęcego, występującym z częstością 0,4–1,3% żywych urodzeń. Choć guzy tego typu są niezłośliwe, ich wczesne wykrywanie i leczenie ma istotne znaczenie, ponieważ brak interwencji niesie ze sobą ryzyko trwałego zniekształcenia czaszki i kręgosłupa niemowlęcia. Uważa się, że głównym czynnikiem etiologicznym wywołującym *fibromatosis colli* jest uszkodzenie mięśnia: wewnątrzmacicznie lub podczas porodu. Częsty objaw wrodzonej fibromatozy szyi stanowi wymuszone przechylanie głowy w stronę dotkniętą zmianami. **Opis przypadku:** W pracy omówiono postępowanie wdrożone u 82-dniowego niemowlęcia z rozpoznaniem fibromatozy szyi. **Omówienie i wnioski:** Kluczowe znaczenie dla prawidłowego rozpoznania *fibromatosis colli* ma badanie przedmiotowe pacjenta obejmujące ocenę palpacyjną oraz określenie zakresu ruchomości mięśnia mostkowo-obojczykowo-sutkowego. U niemowląt z fibromatozą szyi leczeniem pierwszego rzutu jest wcześnie podjęta fizjoterapia.

Słowa kluczowe: *fibromatosis colli*, kręcz szyi, wrodzony guz wieku niemowlęcego, mięsień mostkowo-obojczykowo-sutkowy

INTRODUCTION

Fibromatosis colli (FMC) is an uncommon congenital infantile tumour that typically involves the sternocleidomastoid muscle (SCM), either unilaterally or bilaterally. It is a rare tumour with an incidence of 0.4–1.3% of live births. Traditionally, FMC is characterised by the presence of head tilting towards the affected muscles, while the chin points away from the involved muscles[1]. FMC tumours are benign, not malignant, and they do not metastasise, but they can cause disfigurements[2].

There are many hypotheses pertaining to FMC aetiology. The most notable one includes injury or mass of the SCM. Muscle injury can happen because of abnormal intrauterine foetal positioning, especially during the 3rd trimester, or venous occlusion due to lateral flexion and neck rotation[3]. Difficult labour is another hypothesised cause, as is reduced foetal movement, oligohydramnios, large infant birth weight, breech position, and multiple births[4].

Following injury, tearing and bleeding of the muscles of the neck, scars form within the injured muscle. Fibrosis limits the contractility of the muscles, which reduces the range of neck motion. Deformations of the back, hips, and feet might be among the sequelae of FMC if no early treatment is initiated[5]. This case report discussed an 82-day-old toddler diagnosed with FMC and referred to our centre for further management.

CASE SUMMARY

A 2-month-old female infant presented with left neck swelling, first noticed during the first-month check-up in a private paediatrician's clinic. According to the mother, the swelling was not increasing in size, and was not painful, caused no discharge, and was not associated with any body temperature rise. The infant did not appear uncomfortable at rest but wound cry if the mother tried to turn her head to the left side. The infant was tolerating expressed breast milk, but the mother claimed that the infant found it difficult to latch onto the breast. Occasionally, mild regurgitation was noted post-feeding. There was no other noticeable swelling besides the left neck. The child's limb and eye movement appeared normal. The infant was delivered through spontaneous vaginal delivery (SVD), and no history of trauma birth history was reported.

Upon examination, the baby was generally well, active on handling, pink, not dysmorphic, not in respiratory distress, with warm peripheries and a good pulse volume. The lungs were clear, and no abnormality was noted on heart auscultation. The abdomen was soft, and no hepatosplenomegaly was palpable.

Head examination revealed haemangioma at the skull vertex measuring 3 × 3 cm, flat, blanchable and not pulsatile. A naevus was noted on the left side of haemangioma, measuring 5 × 4 cm, with an irregular surface, and no sign of infection. The head was fixed and tilted to the left side, and

Fig. 1. Head fixed and tilted to the left side

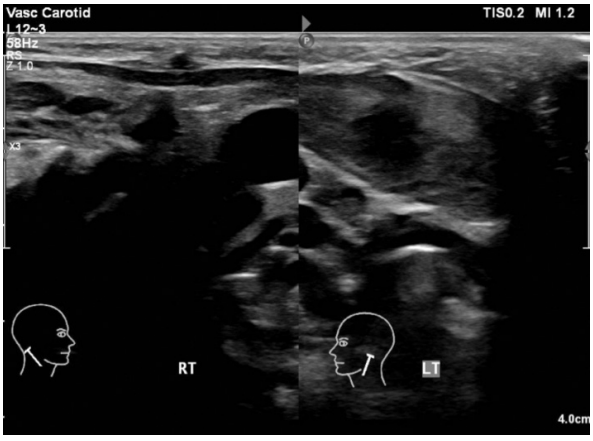

Fig. 2. Ultrasound of the neck shows a fusiform dilatation within the SCM, measuring 1.1 × 3.9 cm at left neck as compared to the right neck, suggestive of FMC

passive neck movement was about 90 degrees to the right side and 30 degrees to the left side (Fig. 1). A palpable hard mass was noted at the left SCM, non-tender, with no discharge, and with normal skin colour. No facial asymmetry and plagiocephaly were observed.

Ultrasound of the neck showed a fusiform dilatation within the SCM, measuring 1.1 × 3.9 cm at the left neck as compared to the right neck (Fig. 2). Cervical lymph nodes with preserved fatty hilum were noted bilaterally. The ultrasound result was suggestive of FMC.

With the consensus from a conference call between the radiologist and otorhinolaryngology head and neck team, the patient was diagnosed with FMC of the left SCM.

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Fig. 3. Improved posture of the head post physiotherapy

The patient was then referred to physiotherapy for neck exercise. A follow-up examination one month post neck exercise shows an improved head posture at rest (Fig. 3). Passive neck movement on the left side increased from 30 degrees to 75 degrees, while the right side remains at 90 degrees. The left neck mass remains palpable. The patient was scheduled for monthly follow-ups in the department.

DISCUSSION

A typical clinical presentation of an infant with FMC of the SCM is head tilting toward the side of the affected muscle, while the chin is pointing towards the opposite side[3]. However, this clinical presentation can be easily missed if the infant is born with a short neck or if FMC occurs in the SCM bilaterally. In bilateral FMC, infants usually present with masses at both SCM muscles and bilaterally restricted neck movement. Thus, physical examination of the clinical presentation is important. Checking for the range of motion of the muscles of the neck and palpating for masses or swelling in the neck region are mandatory[6]. Normal full passive range of motion in neck rotation for a child below the age of 3 years is 100° to 110°. Any movement less than that should be further investigated to rule out FMC. In addition, any facial asymmetry involving the eyes, ears, and mandible should be a cause for alert, as these manifestations can be the sequelae of FMC[5]. Plagiocephaly is another clinical presentation in infants with FMC[7]. Plagiocephaly refers to unilateral flattening of the cranium, often associated with some facial asymmetry. Usually, the frontal areas of the affected side and the parietal occipital area of the contralateral side are flattened.

Limited movement of the affected side of the neck causes the infant to rely on the unaffected side for postural control, which leads to atypical postural and gross motor development. As a result, the infant is unable to breastfeed equally well from both breasts.

According to Boyko et al., clinical examination is sufficient to diagnose congenital torticollis in infants. Imaging with radiographs, computed tomography, and magnetic resonance imaging is not necessary[8]. Palpation of the muscles involved is the primary means of investigation. A fibrous mass is typically appreciated in the SCM region. The mass is usually more prominent in the first 3 to 4 weeks after birth[9]. Clinicians must also be aware that FMC can be found bilaterally on the neck to prevent misdiagnosis. Imaging should only be performed if the infant fails to respond to early physiotherapy treatment after one year or if the clinician suspects that non-muscular pathologies and other neurological findings are present[8]. Invasive diagnostic procedures (such as FNAC) should be avoided, as they can cause further injury to the muscle and worsen the existing muscle fibrosis.

A similar presentation of head tilt posture can also be observed in infants with other syndromes. Consequently, clinicians should rule out other possible syndromes before coming out with a diagnosis. One of the differentials is Sandifer syndrome[9]. It is a combination of hiatal hernia and abnormal posturing of the head and neck. The abnormal posturing has been attributed to attempts to decrease the pain of oesophagitis resulting from gastroesophageal reflux and hiatal hernia. Torticollis resolves after the underlying disease is treated.

Another differential diagnosis to rule out is Klippel–Feil syndrome. An infant with this syndrome presents with congenital vertebral anomalies that cause abnormal head positioning[9]. Thus, a detailed examination of fused cervical vertebrae is important to rule out this syndrome.

Ocular abnormalities such as superior oblique, lateral rectus muscles, palsy or nystagmus can cause ocular torticollis[9]. In such cases, abnormal head posture is adopted to optimise and maintain binocularity. Early referral to an ophthalmologist to identify the affected eye and perform correction of the extraocular muscles involved resolves ocular torticollis.

Torticollis occurring without any associated symptoms is called paroxysmal torticollis[9]. It may be accompanied by pallor, vomiting, irritability, or ataxia. Paroxysmal torticollis resolves spontaneously without any treatment.

The main treatment modality for FMC is physiotherapy[10]. Early physiotherapy of torticollis is important to ensure satisfactory treatment outcomes and potentially eliminate the need for surgical intervention in the future. Early neck stretching of the affected muscle is crucial to prevent

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further muscle fibrosis, as intermittent stretching will stimulate myoblasts to undergo hypertrophy and hyperplasia. This improves muscle formation and reduces fibrosis of the affected site[9].

The recommended neck exercise involves stretching and holding the position for 30 to 60 seconds with three repetitions 6 to 8 times a day[9]. For better outcomes, stretching should be done for about 5 to 10 minutes if the infant is in compliance with the treatment. Often, the caretaker struggles with determining the amount of force to be applied while doing neck stretching exercises with the infant. Caution is advised, as excessive force can injure the infant. An experienced physiotherapist's guidance is mandatory in such circumstances. Alternatively, the caretaker can engage the infant during the physiotherapy session itself[10]. Encouraging the infant to look in the direction of the affected site is a very good way to treat the infant without making them feel uncomfortable. This can be done by holding the baby while trying to gain their attention from the affected side using toys or sounds. Constant flexing of the neck of the infant towards the unaffected side is a good passive neck-stretching exercise[11].

Close follow-up of the progress of the infant is recommended by most of the guidelines[12]. Adjunct therapies such as microcurrent, myokinetic stretching, kinesiology taping and orthoses may be added, if needed. Only infants with complete cervical passive range of movement within 5 degrees of the unaffected side, symmetrical active movement patterns, age-appropriate motor development, and no visible head tilt are recommended to be discharged from formal physical therapy[12].

Infant developmental delay is the most common side effect of untreated FMC. Inability to turn their head to look and interact with their full environment leads to a delay in infants' development. Since the infant is not able to turn their neck to look around at the other side of their body, musculoskeletal issues including abnormalities in their spine alignment and hip dysplasia can occur. This is because children tend to compensate by moving their spine, hip and pelvis in an unnatural way.

The asymmetrical tonic neck reflex (ATNR) will not be integrated in an FMC infant, if left untreated. An integrated ATNR means that infants can turn their head to either side without involuntary limb movement. The ATNR helps the infant to roll, but if it is not integrated, crawling and walking may be delayed.

Infants with torticollis tend to always be looking in one direction. This leads to an increase in pressure on the back and side of their head. Long-term pressure over these regions results in plagiocephaly. Plagiocephaly is the flatness on the back and side of the infant's head and an altered shape of their head, which can lead to asymmetries in facial features such as the position of the ears and eyes. Besides changes in appearance, plagiocephaly can also cause developmental delays in gross and motor skills, language development, and cognitive skills.

CONCLUSIONS

FMC is a rare congenital tumour of the head and neck region. Even though it is benign in nature, early diagnosis and treatment are crucial to prevent permanent developmental deformities to the skull, spine, and other associated body parts in infants. Non-invasive diagnostic modalities should be used as primary diagnostic tools to prevent further worsening of fibrosis. In addition, clinicians should be able to rule out other syndromes with similar presentations, as treating the underlying disease can resolve torticollis.

Conflict of interest

The authors do not declare any financial or personal links to other persons or organisations that could adversely affect the content of this publication or claim rights thereto.

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