CASE REPORT

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Pediatric Cervical Chondromesenchymal Hamartoma: Case Report of a Rare Tumor

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ABSTRACT

Cervical chondromesenchymal hamartoma (CMH) is a rare, benign tumor that typically occurs in the soft tissues of the neck. We present a case of a 4-year-old male child who presented with a slowly expanding, painless left paramedian neck mass. Imaging studies revealed a well-circumscribed, lobulated solid lesion with a heterogeneous appearance, extending from the left thyroid lobe to the upper mediastinum. The mass was completely excised and histopathologically diagnosed as a CMH. This case highlights the importance of considering this rare entity in the differential diagnosis of a neck mass in a child. The imaging characteristics and histopathological findings are discussed, and the treatment and outcome are reported. This case report adds to the limited literature on cervical CMH in children and emphasizes the importance of complete surgical excision as the treatment of choice.

Keywords: Chondromesenchymal hamartoma, Computerized tomography, Magnetic resonance imaging, Neck

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INTRODUCTION

Hamartomas are abnormal growths of cells and tissues that originate in the same organ or tissue where they are found. Histologically, this entity is composed of a combination of immature connective tissue (mesenchymal) and cartilage cells.

Hamartomas of the head and neck are rather uncommon, despite the possibility of presentation in any part of the body. They have been discovered in a number of places, including the endotracheal and endobronchial areas, hypothalamus, dermis, nose, lingua, thyroid gland, and larynx.[1]

Here was a report of a histopathologically proven chondromesenchymal hamartoma (CMH) located in the cervical region adjacent to the left lobe of the thyroid gland.

CASE PRESENTATION

A 4-year-old male child presented to the pediatric surgery clinic in our hospital with a slowly expanding painless left paramedian neck mass. Medical history was unremarkable, and there was no history of drug usage. On physical examination, the patient had a painless mobile mass on the left side of the neck (Fig. 1).

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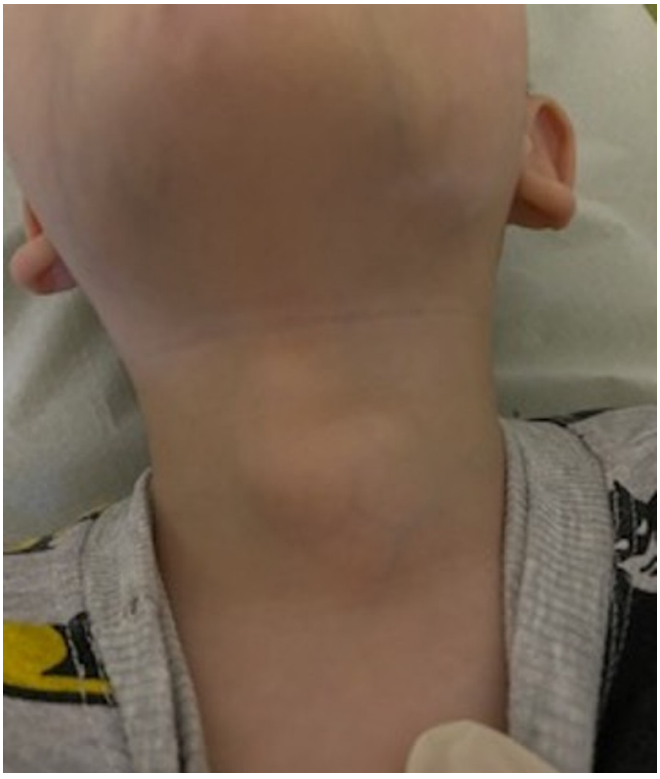

Figure 1. A 4-year-old patient with a left paramedian mass in the neck.

Ultrasound examination revealed a lobulated, contoured solid mass lesion adjacent to the left thyroid lobe, hypoechoic compared to the gland parenchyma, with no vascularization. Computed tomography (CT) and magnetic resonance examinations were performed for optimal evaluation of the lesion borders and neighborhood.

CT examination revealed a 31×48×46 mm, well-circumscribed, lobulated solid lesion with a heterogeneous appearance starting from the localization of the left thyroid lobe and extending toward the upper mediastinum in the paramedian neck (Fig. 2). The trachea was deviated to the right due to mass effect. There was no destruction in the adjacent fatty and bony structures. The right thyroid lobe was normal appearing, but the left lobe could not be differentiated separately.

On magnetic resonance imaging (MRI), a lobule contoured mass lesion was observed, which was hypointense on T1-weighted images, heterogeneously hyperintense on T2-weighted images, showing only thin wall contrast uptake on contrast-enhanced series, and no diffusion restriction on diffusion-weighted images (Figs. 3-6). The lesion was adjacent to the main vascular structures and extended to the upper mediastinum. No pathological lymph nodes were observed in the cervical chains.

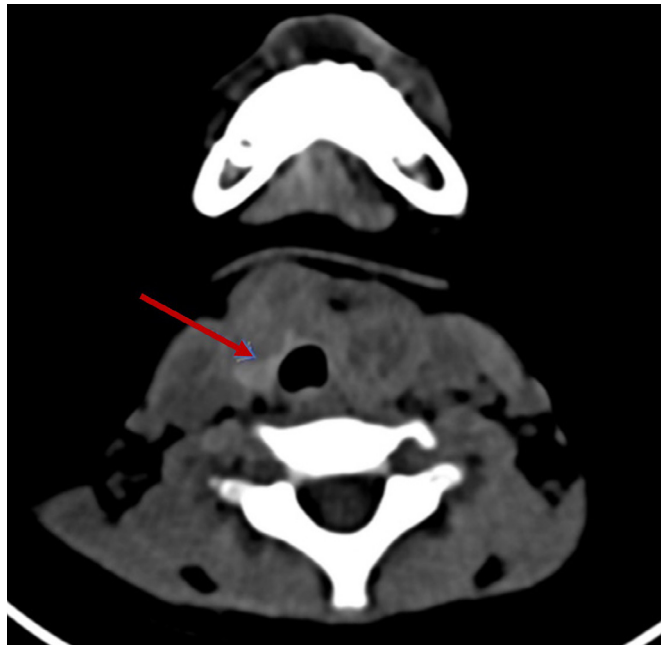

Figure 2. Non-contrast computed tomography scan shows a hypodense, heterogeneous solid mass at the level of the thyroid gland. The right lobe can be differentiated from the mass (arrow).

The patient was operated (Fig. 7).

The lesion, which was found to be anterior to the isthmus and left lobe of the thyroid gland, was completely excised and pathologically diagnosed as CMH (Figs. 8-10).

DISCUSSION

CMHs are rare, benign lesions characterized by disordered proliferation of mesenchymal and cartilaginous tissues. While most commonly reported in the sinonasal cavity as nasal CMHs (NCMHs), their occurrence in atypical locations, such as the cervical region or ectopic thyroid tissue, remains exceedingly rare.[2] Our case presents a 4-year-old male child with a slowly expanding, painless left paramedian neck mass, which was ultimately diagnosed as a cervical CMH. The anatomical presentation of CMHs varies significantly. One of the reported neonatal CMH cases developed within ectopic thyroid tissue.[3] In our patient, the mass arose adjacent to the left thyroid lobe, a feature shared with neonatal cervical CMH described by Yang et al.,[4] causing tracheal deviation. This feature highlights the potential for CMHs to mimic thyroid neoplasms or teratomas radiologically, necessitating histopathological confirmation. In contrast, nasal CMHs that have been reported predominantly involve the nasal cavity, presenting with obstructive symptoms such as nasal congestion or epistaxis.[2,5] Despite these differences, histopathology remains consistent across anatomical sites, with hyaline

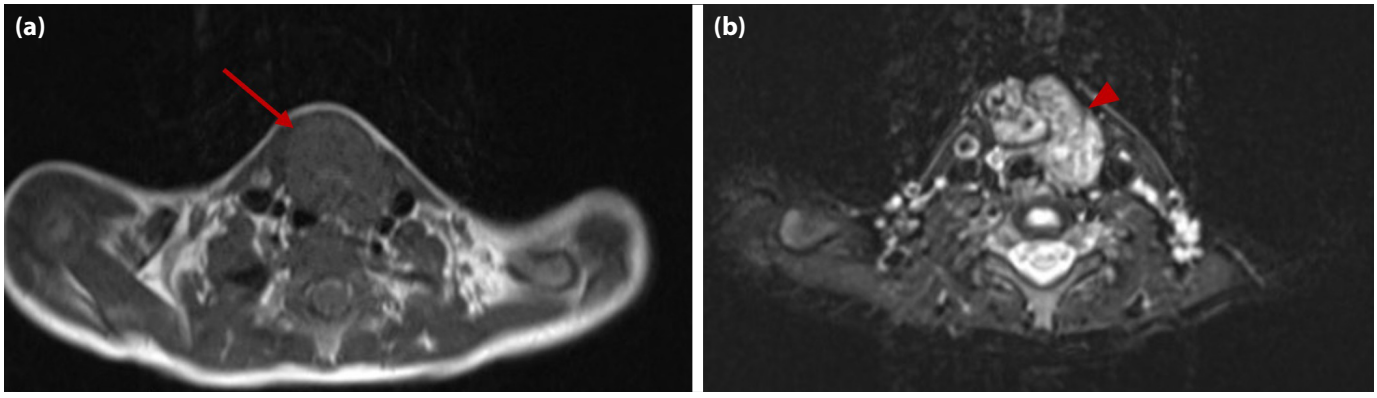

Figure 3. On magnetic resonance examination, the lesion was isointense with adjacent muscle planes in axial T1A **(a)** series and heterogeneous hyperintense signal in fat-suppressed axial T2A series **(b)**.

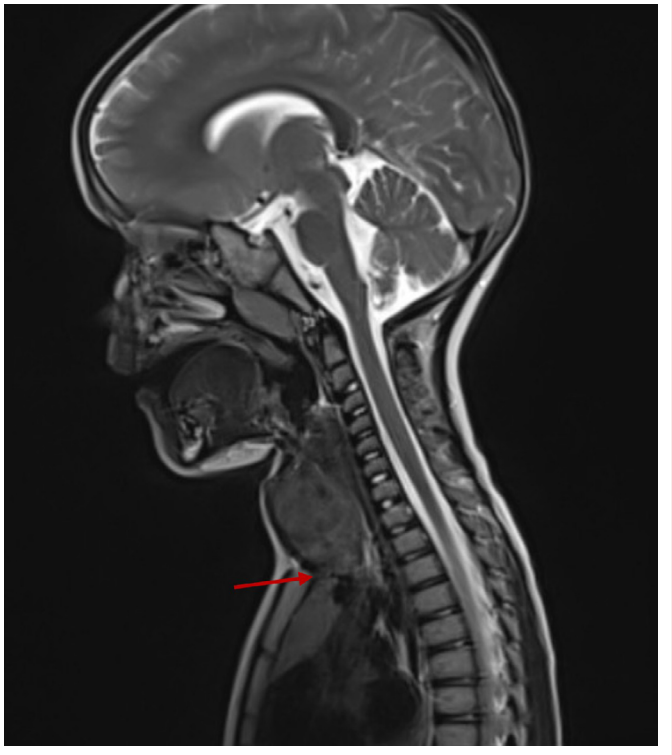

Figure 4. Sagittal T2-weighted images show extension of the lesion toward the upper mediastinum.

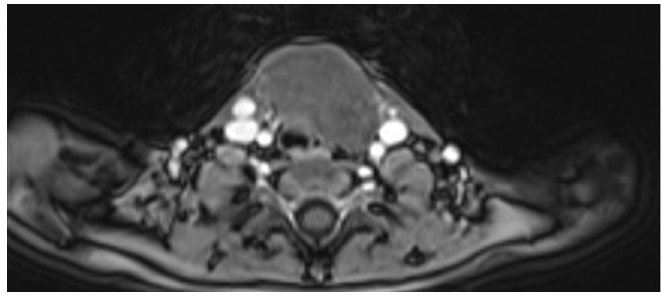

Figure 5. On fat-suppressed axial T1-weighted images, the lesion showed no contrast enhancement except for thin-walled contrast enhancement.

cartilage nodules, spindle cell stroma, and absence of cytologic atypia serving as diagnostic hallmarks.[3,6]

Emerging genetic insights suggest a potential link between CMHs and somatic DICER1 mutations, particularly in nasal cases. In one study, hotspot mutations were identified in exon 25 of DICER1 in 55.6% of NCMH cases, aligning these lesions with the DICER1 tumor family.[5] While genetic testing was not performed in our cervical case or the neonatal ectopic thyroid CMH case,[3] this molecular association raises questions about shared pathogenic mechanisms across anatomical subtypes. Notably, DICER1-mutated tumors often arise in pediatric populations, consistent with the age distribution of CMHs.[5,6] CMHs exhibit a strong pediatric predominance, as evidenced by our case, the neonatal ectopic thyroid CMH,[3] and the neonatal cervical CMH.[4] However, adult NCMH cases were also reported (up to 55 years), demonstrating that these lesions are not exclusive to early childhood.[5] Regardless of age, complete surgical excision remains the cornerstone of management. In our patient, total resection resulted in symptom resolution without recurrence at 6-month follow-up – an outcome mirrored in both neonatal and adult cases.[3,5] This consistency reinforces the benign nature of CMHs but underscores the need for meticulous pre-operative imaging (MRI/CT) to delineate tumor margins, particularly in complex cervical or mediastinal locations.[2,7]

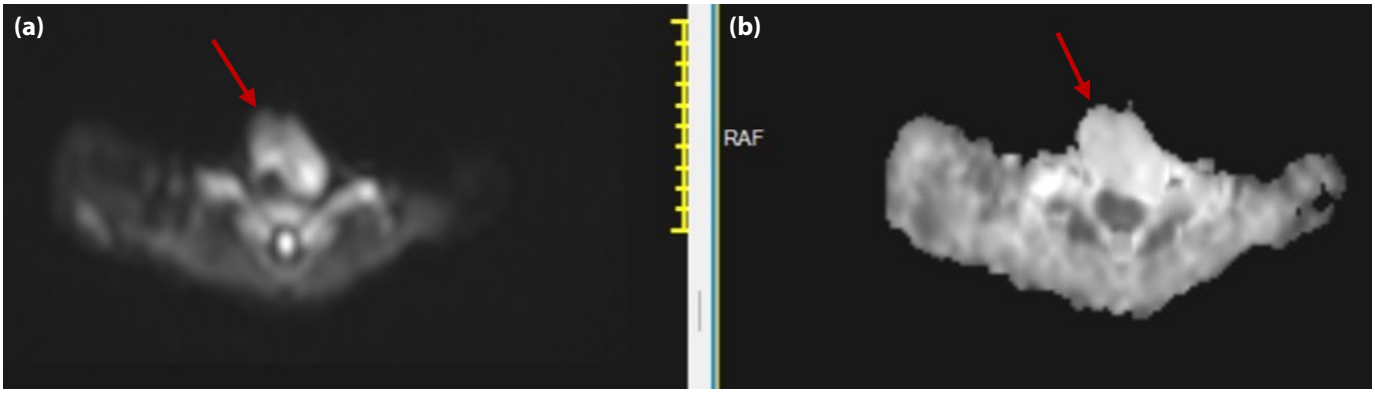

Figure 6. Diffusion **(a)** and ADC **(b)** images show no diffusion restriction of the lesion.

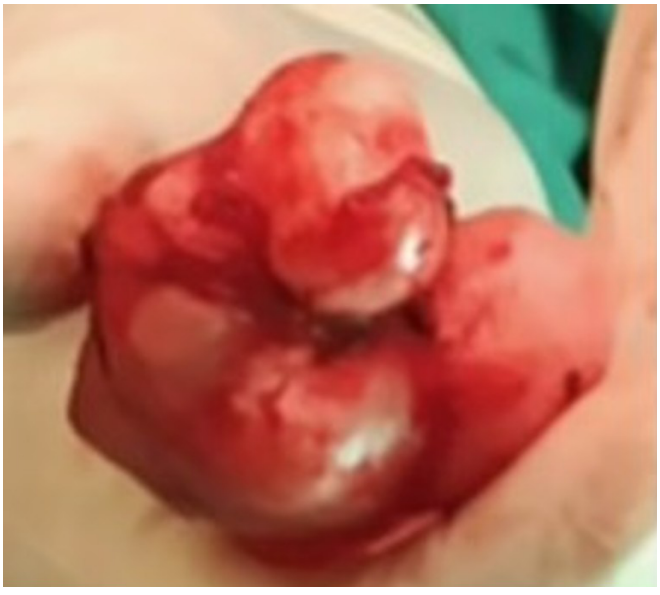

Figure 7. Macroscopic view of the total excised lesion.

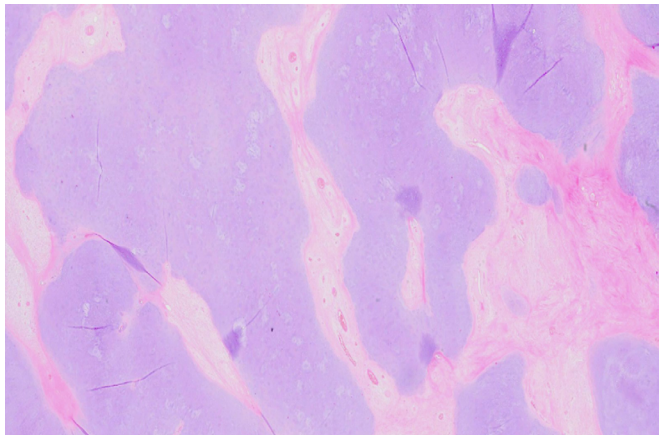

Figure 8. Pathological characteristics of the chondromesenchymal hamartoma (HE staining) in case. Hyaline cartilage with endochondral ossification surrounds.

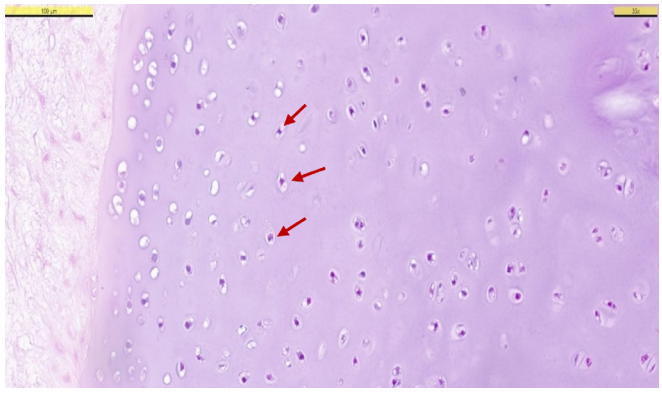

Figure 9. Chondrocytes in lacunae, arranged diffusely. Cellular atypia, mitotic figures, and cellularity were not seen.

The rarity of CMHs increases their susceptibility to misdiagnosis. In our case, imaging initially suggested a thyroid neoplasm, while according to the literature, 44% of NCMHs were misclassified preoperatively as nasal polyps or cartilaginous lesions.[5] Similarly, previous studies emphasized the non-specific radiological features of their neonatal ectopic thyroid CMH.[3] Key differentials for cervical CMHs include teratomas, rhabdomyosarcoma, and lymphatic malformations, all requiring distinct management approaches.[1,7]

Histopathology remains critical for definitive diagnosis, as it reveals the characteristic admixture of cartilage, spindle cells, and the absence of malignancy.[3,6]

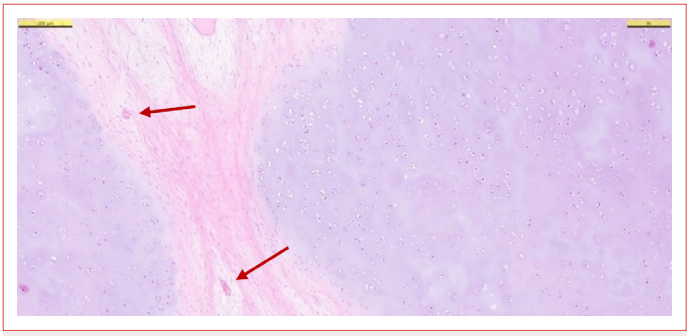

Figure 10. A number of mesenchymal spindle-shaped cells interwoven with multilobulated hyaline cartilage.

Congenital and developmental anomalies may contribute to CMH pathogenesis. The neonatal ectopic thyroid CMH exemplifies this, as aberrant thyroid migration likely provided a nidus for hamartomatous growth.[3] While our patient had no congenital abnormalities, this association highlights the importance of prenatal imaging and postnatal vigilance in neonates with neck masses. The role of hormonal or embryonic factors, such as the progesterone receptor expression noted in a scalp CMH, warrants further exploration.[8]

CONCLUSION

Our case highlights the importance of considering CMH in the differential diagnosis of a neck mass in a child. The imaging characteristics and histopathological findings in our case are consistent with this diagnosis, and the tumor was successfully treated with complete surgical excision.

DECLARATIONS

Ethics Committee Approval: This is a single case report, and therefore ethics committee approval was not required in accordance with institutional policies.

Informed Consent: Informed consent was obtained from the parents of the child.

Conflict of Interest: The authors declare that there is no conflict of interest.

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