Int. J. Morphol.,
44(1):206-210, 2026.

Relationship Between Neural Tube Closure Defects and the Presence of Sternal Muscle: A Case Report and an Embryological Hypothesis

Relación Entre Defectos del Cierre del Tubo Neural y la Presencia del Músculo Esternal: Un Reporte de Caso y una Hipótesis Embriológica

Emilio Farfán[1]; Oscar Inzunza[1]; Mark Echeverría[1]; Verónica Inostroza[1]; Jaritza Tramolao[1] & Natalia Sánchez[1]

FARFÁN, E.; INZUNZA, O.; ECHEVERRÍA, M.; INOSTROZA, V.; TRAMOLAO, J. & SÁNCHEZ, N. Relationship between neural tube closure defects and the presence of sternal muscle: A case report and an embryological hypothesis. *Int. J. Morphol., 44(1)*:206-210, 2026.

SUMMARY: The sternal muscle is an anatomic variation characterized by the presence of an accessory muscle in the ventral thoracic wall. Its occurrence is more prevalent in anencephalic fetuses or newborns, although the underlying reason remains unclear. Anencephaly results from a failure in the neural tube closure, leading to brain tissue protrusion and subsequent degeneration. Here, we report the presence of a sternal muscle in an anencephalic newborn. A white male anencephalic newborn was dissected to expose the ventral thoracic muscular wall. Morphometric analysis of the sternal muscle was performed. We identified a bilateral sternal muscle in an anencephalic newborn, with an unusual presentation: the right hemithorax contained a simple fascicle, whereas the left hemithorax presented a double fascicle, whit the superior fascicles bifurcating again in two fascicles. All the fascicles originated from the contralateral pectoralis major muscle. Innervation and vascularization could not be determined. The findings meet the criteria to be considered an uncommon bilateral sternal muscle. We discuss possible associations between the sternal muscle and anencephaly, considering genetic components such as PAX3, CXCR4, and PCP/non-canonical Wnt pathway. This hypothesis is based on the embryologic origin of thoracic ventral muscles and the process of neural tube closure. We also propose investigating the presence of sternal muscle in adults as a potential indicator of less severe neural tube closure defects.

KEY WORDS: Muscle development; Anencephaly; Embryology.

INTRODUCTION

The sternal muscle is an anatomic variant characterized by an accessory muscle in the ventral wall of the thorax. Its prevalence ranges from 3-8 % (Arraez-Aybar *et al*., 2003; Snosek *et al*., 2014), with higher occurrence in the Asian populations (11.5 %) compared with White (4-7 %) and Black populations (8.4 %) (Bergman *et al*., 1988). Regarding gender, it is more frequent in women (8.7 %) than in men (6.4 %) (Scott-Conner & Al-Jurf, 2002). The muscle was first described by Cabrolius in 1604 (Testut, 1884), and has since been identified in diverse through cadaver dissections, and imaging techniques such as MRI and scanners. For classification as a sternal muscle, four criteria must be met: 1) location within the fascia of the pectoralis major muscle; 2) origin from the sternum or infraclavicular region; 3) insertion into the lower ribs, costal cartilages, aponeurosis of the external oblique, or sheath of the rectus abdominis; and 4) innervation by pectoral or intercostal nerves (Jelev *et al*., 2001). Its morphology can vary depending of the differences on the number of fascicles and the sites of origin and insertions, allowing for further classification (Jelev *et al*., 2001; Snosek *et al*., 2014).

The ventrolateral thoracic muscle wall, including the sternocleidomastoid, serratus anterior, intercostal, and, pectoralis major and minor muscles, derives from the somitic mesoderm. In the consensus model, the first muscle progenitors (myoblast) delaminate from the dorsomedial lip of the dermomyotome. These myoblasts proliferate, migrate, and organize to form the four corners of the primary myotome, then a population of myoblast derived from the central part of the dermomyotome form the secondary myotome (Hollway & Currie, 2005; Asghar *et al*., 2022). However, the embryological origin of the sternal muscle remains uncertain.

[1]Departamento de Anatomía, Escuela de Medicina, Pontificia Universidad Católica de Chile, Santiago, Chile.

Received: 2025-08-18 Accepted: 2025-11-25

Interestingly, a higher prevalence of sternal muscle has been described in anencephalic fetuses. Abraham (1883) observed it in 6 of 11 anencephalic fetuses (55 %), and Shepherd (1885) reported it in all 6 cases studied (100 %), 3 unilateral and 3 bilateral, suggesting an origin of the sternal muscle in the clavicular fibers of the pectoralis major muscle.

Anencephaly is a neural tube closure defect resulting from failure of anterior neuropore closure during early gestation. Its prevalence is 1-5 per 1000 births, with higher rates in China, Mexico, Turkey, and the British Isles (Mai *et al*., 2019). Neural tube formation begins at 17-18 days of gestation trough neurulation. Neurulation occurs when the inductive signaling released from the notochord to the overlying ectoderm induces neural plate formation. Then, the edges of the neural plate begin to elevate, generating the neural folds. Those neural folds start to get close to finally fuse, forming the neural tube. The neural tube closure commences at cervical level (5th somite), and expands to the cranial and caudal end as a zipper, transiently generating the cranial and caudal neuropores. Failure to close at the cranial ends leads to anencephaly, a condition marked by disrupted cell migration, cell adhesion, and closure mechanisms (Sadler, 2016). Affected fetuses typically die in utero or within hours to days after birth, representing 100 % of mortality (Johnson *et al*., 2012). While the cerebellum and the brain stem may develop, the forebrain and calvarium are partially or completely absent (Sadler, 2016).

In this report, we describe a bilateral sternal muscle in an anencephalic, white, and male newborn, and discuss possible developmental link between anencephaly and sternal muscle considering their embryological development.

MATERIAL AND METHOD

A white, male, and anencephalic newborn body was donated to the Pathological Anatomy Service of Sótero del Río Hospital, Santiago, Chile and since 1978 it has been part of the teaching resources of the Anatomy Department at the Pontificia Universidad Católica de Chile. The body was preserved by perfusion with 10 % buffered formaldehyde and stored at 4 °C. The newborn showed no evidence of prior surgeries or interventions in the sternal region and was intact at the time of dissection.

Following rinsing in 0.9 % NaCl, dissection of the anterior thoracic wall was carried out. A longitudinal midline incision was made from the jugular notch of the sternum to the infrasternal angle, lifting the skin laterally while preserving the deep fascia. A muscle located between the superficial fascia and the pectoralis major muscle was identified, extending from the base of the neck toward the abdomen. After removal of the deep fascia, the sternal muscle was fully exposed. The cranial and caudal ends were examined to determine their insertions, as well as possible vascularization, innervation and anatomical relationships. Morphometric measurements of the muscle fascicles were performed. All procedures were conducted under Ethics Committee approval of the Pontificia Universidad Católica de Chile.

RESULTS

An anencephalic newborn with acrania and minimal brain tissue was dissected (Fig. 1A, B). During thoracic dissection, we identified an atypical muscle structure composed of two muscular fascicles located on the ventral surface of the thorax, beneath the skin and subcutaneous tissue, and superficial to the pectoralis major muscle.

In the right hemithorax, a single muscular fascicle originated from the sternocostal head of the left pectoralis major muscle. This fascicle crosses the midline from left to right, extending downward and laterally over the pectoralis major muscle, and terminating at the costal arch. The deep surface of the fascicle was separated from the pectoralis major muscle by a distinct fascia. Inferiorly, its tendinous fibers intertwined with the pectoral fascia and the aponeurosis of the abdominal external oblique muscle (Fig. 1C). The total length of the right fascicle was 37.65 mm, with a transversal diameter of 7.17 mm (Fig. 1D). Vascularization and innervation could not be determined.

In the left hemithorax, a fascicle originated from the sternocostal portion of the right pectoralis major muscle. This fascicle crossed deeply toward the right sternal muscle before branching into two superficial fascicles overlying the left pectoralis major muscle, one superior and one inferior (Fig. 1C). The superior fascicle ran horizontally, measuring 22.89 mm in length and 6.97 mm in transverse diameter. It divided into two smaller fascicles, whose fibers were continuous with the pectoral fascia. The inferior fascicle ran obliquely, measuring 18.41 mm in length and 6.96 mm in transverse diameter (Fig. 1D). It terminated in tendinous fibers that blended with the pectoral fascia and the aponeurosis of the abdominal external oblique muscle (Fig. 1C). At the right side, vascularization and innervation could not be determined.

In summary, the dissection revealed muscular fascicles consistent with an uncommon bilateral sternal muscle, and met at least three of the four established diagnostic criteria, with the exception of confirmed innervation.

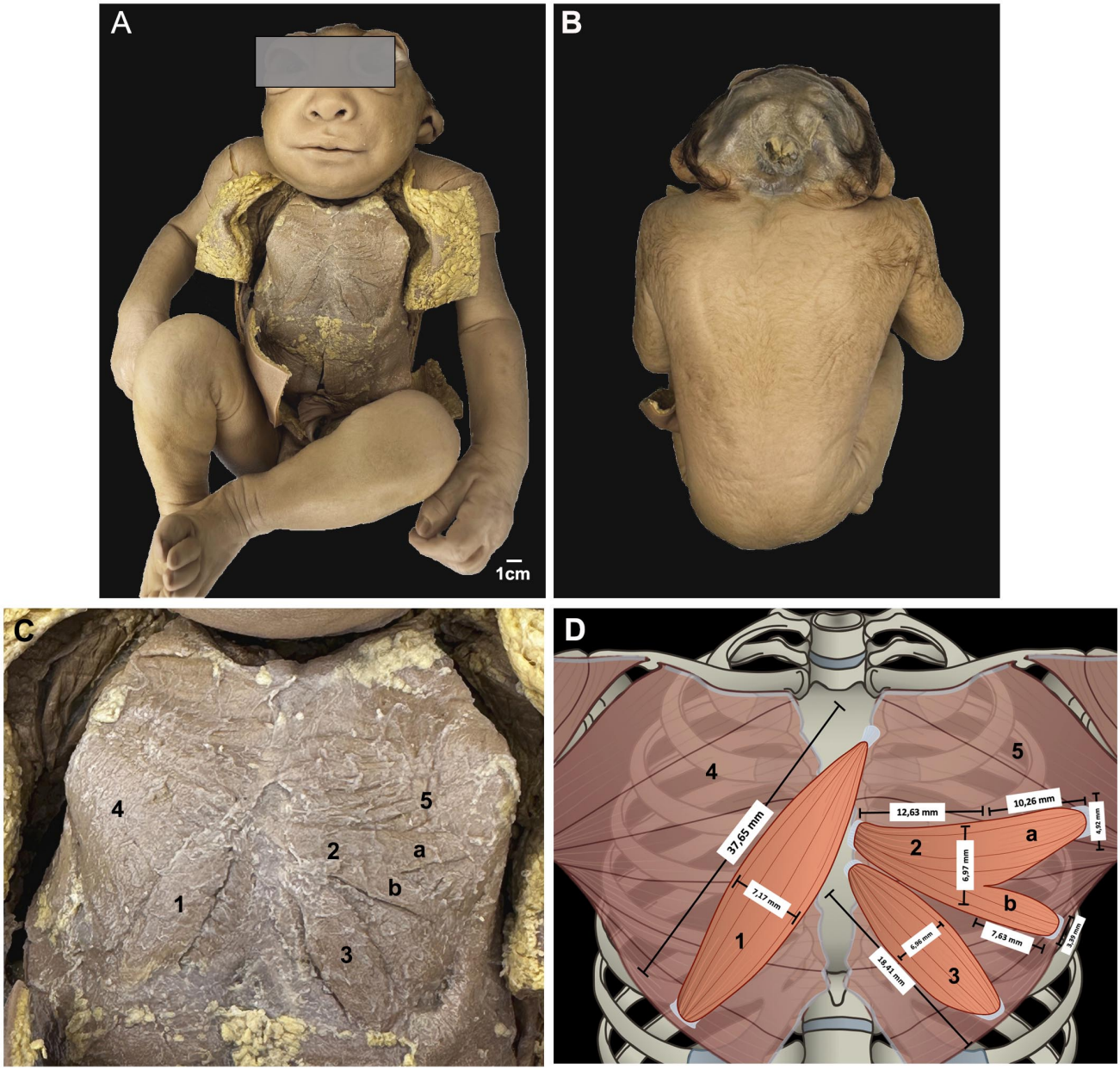

Fig. 1. Anencephalic newborn with thoracic dissection. A Ventral view showing the bilateral sternal muscles. B Dorsal view displaying acrania with minimal brain tissue. C Magnified view of the dissected area. D. Schematic representation of the sternal muscles in the anencephalic newborn, indicating the total lengths and transverse diameters of the muscular fascicles. 1. Right sternal muscle; 2. Left sternal muscle, upper fascicle; a. Upper subdivision of the left upper fascicle; b. Lower subdivision of the left upper fascicle; 3. Left sternal muscle, lower fascicle; 4. Right pectoralis major muscle; 5. Left pectoralis major muscle.

DISCUSSION AND CONCLUSION

This report described the presence of a bilateral sternal muscle in the thorax of an anencephalic newborn. The subject, a white and male newborn, represents one of the most least likely combinations for sternal muscle occurrence. The presentation of the sternal muscle is unusual: the right sternal muscle closely resembles a simple type, while the left side shows a double fascicle with a single-diverging subtype (Snosek *et al*., 2014). However, the case does not fit neatly to the current classification, as both fascicles originated from the pectoralis major muscles, making this a unique and previously undescribed configuration.

The embryologic origin of the sternal muscle remains subject to discussion. Some authors regard it as an error in the early development of the ventrolateral thoracic muscle wall (Hollway & Currie, 2005). Historically, it was considered a rudimentary form of the pectoralis cutaneous seen on lower animals (Turner, 1867). Subsequent works propose origins from adjacent muscles such as panniculus carnosus, sternocleidomastoid, pectoralis major, or rectus abdominis (Kida *et al*., 2000; Jelev *et al.*, 2001). An alternative hypothesis suggest derivation from the somatic mesodermal plate, as a part of the ventral longitudinal muscular column arising from thoracic hypomeres, whose equivalent in the abdomen originates the rectus abdominis muscle (Kumar *et al*., 2003). Another model, the "in-out mechanism", proposes that pectoral muscle progenitors migrate transiently to the limb bud before returning to the lateral plate mesoderm of the trunk to form the pectoralis muscles, potentially including the sternal muscle (Valasek *et al*., 2011; Asghar *et al*., 2022). Variations in the pectoralis major muscles are often concurrent with sternal muscle development, suggesting a developmental link between both muscles (Abraham, 1883). Vascularization and innervation patterns can also provide insights into the muscle's embryological origin. In this dissection, the vascularization or innervation of these muscles could not be determined, probably due to its small size; therefore, it was not possible to suggest an embryological origin.

The correlation between sternal muscle presence and anencephaly has previously been documented (Abraham, 1883; Shepherd, 1885), though the underlying cause remains unclear. Given the shared developmental timelines of neural tube closure and thoracic muscle wall formation, it is plausible that they share common molecular pathways susceptible to disruption. The etiopathology of anencephaly includes environmental and genetic causes and it is often associated with other defects, including cleft palate, cardiac and pulmonary defects, club foot, overlapping fingers, and other musculoskeletal anomalies (Martins Santana *et al*., 2018).

While exploring candidate genes, we identified Pax3 (paired box 3), Cxcr4, and the non-canonical or PCP Wnt pathway as plausible shared regulators for neural tube and muscle development.

Pax3, a helix-loop-helix transcription factor, is essential for hypaxial dermomyotome formation and muscle progenitor migration. Pax3 knockout mice show severe muscle hypoplasia of limb, ventral thoracic and abdominal muscles (Scaal, 2021). Mutations in Pax3 are also linked to neural tube defects, including anencephaly, which can be prevented by maternal folic acid supplementation (Sudiwala *et al*., 2019). Cxcr4, a chemokine factor, participates in the "in-out mechanism" of muscle formation (Rehimi *et al*., 2010), and is expressed in the neural tube neuroepithelium (Denny *et al*., 2013). The PCP/non-canonical Wnt pathway regulates neuroepithelial cell divisions and neural tube closure (Gao *et al*., 2012), and also participates in the maintenance of the dermomyotome, the formation of abdominal muscles and ventral body wall closure (Krück & Scaal, 2012). These overlapping roles suggest that alteration in these pathways could explain the higher incidence of sternal muscle in anencephalic fetuses.

Notably, the presence of the sternal muscle and anencephaly have been reported more frequently in Asian and Chinese populations (Bergman *et al*., 1988; Mai *et al*., 2019), suggesting a genetic background effect. Consequently, it has been described which mice with the Pax3 mutation have greater frequency of anencephaly in certain strains, suggesting that genetic background is relevant for the expression of this phenotype (Fleming & Copp, 2000).

Environmental teratogens also play a major role in anencephaly. In this regard, the timing of disruption of molecular control is critical: early perturbations in shared molecular pathways could affect both neural tube and myogenesis, whereas later disturbances might only alter muscle differentiation. This may explain why sternal muscle is sometimes found in adults without neural tube defects.

Finally, it would be valuable to investigate whether sternal muscle presence in adults could be linked to subclinical or less severe neural closure tube defects, which may go undetected in routine dissection or imaging. Exploring the potential shared embryological origins of the neural tube and thoracic muscles could yield insights into developmental biology, refine diagnostic criteria and therapeutic considerations.

ACKNOWLEDGEMENTS. As researchers and teachers, we thank the people who donate their bodies to science to contribute to morphological research and training new and better professionals in an enormous act of generosity and greatness.

Ethical approval and consent for publication.The body was donated by the Pathological Anatomy Service of Sótero del Río Hospital, Santiago, Chile, and since 1978 it has been part of the teaching resources of the Anatomy Department of the Pontificia Universidad Católica de Chile (PUC), which complies fully with the World Medical Association's Declaration of Helsinki and national legal and ethical requirements. Consequently, the study was approved by the MED-UC Scientific Ethics Committee of the Pontificia Universidad Católica de Chile (No: 190115002).

FARFÁN, E.; INZUNZA, O.; ECHEVERRÍA, M.; INOSTROZA, V.; JARITZA TRAMOLAO, J. & SÁNCHEZ, N. Relación entre defectos del cierre del tubo neural y la presencia de músculo esternal: Reporte de un caso y una hipótesis embriológica. *Int. J. Morphol., 44(1)*:206-210, 2026.

RESUMEN: El músculo esternal es una variación anatómica caracterizada por la presencia de un músculo accesorio en la pared torácica ventral. Su aparición es más frecuente en fetos o recién nacidos anencefálicos, aunque la causa subyacente no está clara. La anencefalia se debe a una falla en el cierre del tubo neural, lo que provoca protrusión del tejido cerebral y su posterior degeneración. En este trabajo, informamos de la presencia de un músculo esternal en un recién nacido anencefálico. Se diseccionó a un recién nacido anencefálico, varón de raza blanca, para exponer la pared muscular torácica ventral. Se realizó un análisis morfométrico del músculo esternal. Identificamos un músculo esternal bilateral en un recién nacido anencefálico, con una presentación inusual: el hemitórax derecho contenía un fascículo simple, mientras que el hemitórax izquierdo presentaba un fascículo doble, con los fascículos superiores bifurcándose en dos. Todos los fascículos se originaron en el músculo pectoral mayor contralateral. No fue posible determinar la inervación ni la vascularización. Los hallazgos cumplen los criterios para ser considerados un músculo esternal bilateral poco común. Discutimos las posibles asociaciones entre el músculo esternal y la anencefalia, considerando componentes genéticos como PAX3, CXCR4 y la vía PCP/Wnt no canónica. Esta hipótesis se basa en el origen embriológico de los músculos ventrales torácicos y el proceso de cierre del tubo neural. También proponemos investigar la presencia de músculo esternal en adultos como un posible indicador de defectos de cierre del tubo neural menos graves.

PALABRAS CLAVE: Desarrollo muscular; Anencefalia; Embriología.

REFERENCES

Abraham, P. S. Notes on the occurrence of the musculus sternalis in human anencephalic foetuses. *Trans. Acad. Med. Ireland, 1(1)*:301-4, 1883.

Arraez-Aybar, L. A.; Sobrado-Perez, J. & Merida-Velasco, J. R. Left musculus sternalis. *Clin. Anat., 16(4)*:350-4, 2003.

Asghar, A.; Naaz, S.; Narayan, R. K. & Patra, A. The prevalence and distribution of sternalis muscle: A meta-analysis of published literature of the last two hundred years. *Anat. Sci. Int., 97(1)*:110-23, 2022.

Bergman, R.; Thompson, S.; Afifi, A. & Saadeh, F. C*ompendium of Human Anatomic Variations.* Baltimore-Munich, Urban & Schwarzenberg, 1988.

Denny, K. J.; Jeanes, A.; Fathe, K.; Finnell, R. H.; Taylor, S. M. & Woodruff, T. M. Neural tube defects, folate, and immune modulation. *Birth Defects Res. A Clin. Mol. Teratol., 97(9)*:602-9, 2013.

Fleming, A. & Copp, A. J. A genetic risk factor for mouse neural tube defects: Defining the embryonic basis. *Hum. Mol. Genet., 9(4)*:575-81, 2000.

Gao, Y.; Chen, X.; Shangguan, S.; Bao, Y.; Lu, X.; Zou, J.; Guo, J.; Dai, Y. & Zhang, T. Association study of PARD3 gene polymorphisms with neural tube defects in a Chinese Han population. *Reprod. Sci., 19(7)*:764-71, 2012.

Hollway, G. & Currie, P. Vertebrate myotome development. *Birth Defects Res. C Embryo Today, 75(3)*:172-9, 2005.

Jelev, L.; Georgiev, G. & Surchev, L. The sternalis muscle in the Bulgarian population: Classification of sternales. *J. Anat., 199(Pt. 3)*:359-63, 2001.

Johnson, C. Y.; Honein, M. A.; Dana Flanders, W.; Howards, P. P.; Oakley Jr., G. P. & Rasmussen, S. A. Pregnancy termination following prenatal diagnosis of anencephaly or spina bifida: A systematic review of the literature. *Birth Defects Res. A Clin. Mol. Teratol., 94(11)*:857-63, 2012.

Kida, M. Y.; Izumi, A. & Tanaka, S. Sternalis muscle: Topic for debate. *Clin. Anat., 13(2)*:138-40, 2000.

Krück, S. & Scaal, M. Divergent regulation of Wnt-mediated development of the dorsomedial and ventrolateral dermomyotomal lips. *Histochem. Cell Biol., 138(3)*:503-14, 2012.

Kumar, H.; Rath, G.; Sharma, M.; Kohli, M. & Rani, B. Bilateral sternalis with unusual left-sided presentation: A clinical perspective. *Yonsei Med. J., 44(4)*:719-22, 2003.

Mai, C. T.; Isenburg, J. L.; Canfield, M. A.; Meyer, R. E.; Correa, A.; Alverson, C. J.; Lupo, P. J.; Riehle-Colarusso, T.; Cho, S. J.; Aggarwal, D.; *et al*. National population-based estimates for major birth defects, 2010–2014. *Birth Defects Res., 111(18)*:1420-35, 2019.

Martins Santana, E. F.; Araujo Junior, E.; Tonni, G.; Costa, F. D. S. & Meagher, S. Acrania-exencephaly-anencephaly sequence phenotypic characterization using two- and three-dimensional ultrasound between 11 and 13 weeks and 6 days of gestation. *J. Ultrason., 18(74)*:240-6, 2018.

Rehimi, R.; Khalida, N.; Yusuf, F.; Morosan-Puopolo, G. & Brand-Saberi, B. A novel role of CXCR4 and SDF-1 during migration of cloacal muscle precursors. *Dev. Dyn., 239(6)*:1622-31, 2010.

Sadler, T. W. Langman's Medical Embryology. 13th ed. Philadelphia, Wolters Kluwer Health/Lippincott Williams & Wilkins, 2016.

Scaal, M. Development of the amniote ventrolateral body wall. *Dev. Dyn., 250(1)*:39-59, 2021.

Scott-Conner, C. E. H. & Al-Jurf, A. S. The sternalis muscle. *Clin. Anat., 15(1)*:67-9, 2002.

Shepherd, F. J. The musculus sternalis and its occurrence in (human) anencephalous monsters. *J. Anat. Physiol., 19(Pt. 3)*:310.2-319, 1885.

Snosek, M.; Tubbs, R. S. & Loukas, M. Sternalis muscle: What every anatomist and clinician should know. *Clin. Anat., 27(6)*:866-84, 2014.

Sudiwala, S.; Palmer, A.; Massa, V.; Burns, A. J.; Dunlevy, L. P. E.; de Castro, S. C. P.; Savery, D.; Leung, K. Y.; Copp, A. J. & Greene, N. D. E. Cellular mechanisms underlying Pax3-related neural tube defects and their prevention by folic acid. *Dis. Model. Mech., 12(11)*:dmm042234, 2019.

Testut, L. *Anomalies Musculaires chez l'Homme*. Paris, G. Masson, 1884.

Turner, W. On the musculus sternalis. *J. Anat. Physiol., 1(2)*:246-53, 1867.

Valasek, P.; Theis, S.; DeLaurier, A.; Hinits, Y.; Luke, G. N.; Otto, A. M.; Minchin, J.; He, L.; Christ, B.; Brooks, G.; *et al*. Cellular and molecular investigations into the development of the pectoral girdle. *Dev. Biol., 357(1)*:108-16, 2011.

Corresponding author:
Dr. Natalia Sanchez
Departamento de Anatomía
Escuela de Medicina
Pontificia Universidad Católica de Chile
Av. Libertador Bernardo O'Higgins 340
Santiago
CHILE

E-mail:nlsanche@uc.cl