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Craniofacial Muscles

A New Framework for Understanding the Effector Side of Craniofacial Muscle Control

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Part I
Overview

Chapter 1
The Craniofacial Muscles: Arguments for Uniqueness

Francisco H. Andrade and Linda K. McLoon

1.1 Introduction

The craniofacial muscles are small skeletal muscles associated with head and neck structures and involved in a wide array of non-locomotor activities such as mastication, swallowing, breathing, vocalization, facial expression, and even vision and other special senses. These muscles are the new kids on the block, starting with their relatively recent appearance with the evolution of the head and neck in vertebrates and to our growing understanding of their distinctive development programs, functions, and pathologies. For convenience, we can group the craniofacial muscles according to their developmental origin: extraocular muscles, branchiomeric muscles (facial, masticatory, pharyngeal, and laryngeal muscles), and tongue muscles (Noden and Francis-West 2006). There is growing recognition of clinical relevance of the craniofacial muscles in terms of diseases that are specific to them (strabismus, laryngeal dystonias, facial paralysis, and many others), but also their characteristic divergent response to certain systemic neuromuscular disorders (sparing by some muscular dystrophies, targeting by myasthenia gravis, to name a few). These are the basic arguments for the uniqueness of the craniofacial muscles that serve as the central theme for the following chapters.

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1.2 Origin of Head and Neck Musculature

Craniofacial evolution is one of the key steps in the origin and diversification of vertebrates (Trainor et al. 2003). The Craniata, a clade within the phylum Chordata (animals with notochords at one point in their lives), arose during the Cambrian explosion (~525 million years ago). In contrast to other chordates, the craniates have heads with complex (vesicular) brains encased in a rigid cranium and specialized sensory organs, including laterally placed eyes. Properly speaking, a skull is composed of the cranium and the jaw. The latter is an even more recent evolutionary feature: its presence separates the "jawed vertebrates" (Gnathostomata, the great majority of vertebrates, including mammals) from the older members of the Craniata clade, the jawless fishes (lampreys and hagfishes). The appearance of the head and neck is one of the factors that allowed vertebrates to shift from filter feeding to active predation, greater motility, and a faster metabolic rate. At least partly due to these characteristics, vertebrates have a presence in a broad range of terrestrial and aquatic environments. Among many other distinguishing features, vertebrates have a complex muscular digestive system, and the cardiovascular system has a heart with two or more chambers, all additions required to cope with greater metabolic demands imposed by their larger size and active lifestyle.

The craniofacial muscles allow for a broad motor repertoire in the head and neck. It will become apparent in the next chapters that these muscles build on the striated muscle stereotype to serve the unique motor needs of craniofacial structures. In other words, the craniofacial muscles do not have quite the same developmental origin as other striated muscles, skeletal or cardiac. To start, craniofacial muscles are non-somitic; they do not follow the progression from mesoderm to segmented somites, which characterizes the development of trunk and limb muscles. Instead, the cranial mesoderm that originates craniofacial muscles differentiates along limits set by molecular rather than anatomical boundaries (Sambasivan et al. 2011). Craniofacial muscles can trace their most primitive origins to trunk musculature, as evidenced by shared steps in their respective myogenic programs. However, the developmental programs for craniofacial muscles show a level of complexity that befits both their relatively recent appearance and the allocation of craniofacial mesoderm for multiple fates. For example, some craniofacial muscles follow a developmental program very similar to that of the heart, hinting at a common evolutionary origin (Kelly 2010). Neural crest plasticity and independent gene regulation (from other muscle groups) are needed for greater adaptability to environmental pressures (Trainor et al. 2003). This inherent plasticity in the craniofacial developmental program gives rise to the diverse cranial phenotypes we see around us today: wide range of beaks in birds, the repurposing of mandibular bones for hearing (all the way to four middle ear bones in mammals), the sensory and motor function of the elephant's trunk, the muscles of facial expression in humans, and the enhanced sensory role of the nose of the star-nosed mole.

1.3 Craniofacial Muscle Function

The structures of the head and neck protect the brain, provide support for delicate sensory organs, and are needed for feeding and exploratory behaviors. The craniofacial muscles, which include the extraocular, facial, masticatory, pharyngeal, laryngeal, and tongue muscles, are required for all motor activities of the head and neck. The extraocular muscles are responsible for the coordinated voluntary and reflexive movement of the vision organs, the eyes. Their anatomical arrangement and innervation are highly conserved, suggesting that the ocular motor system is fairly ancient, maybe predating some trunk and limb muscles. With rare exceptions, even the most primitive vertebrates have at least six extraocular muscles for each eye (Noden and Francis-West 2006). As is the case in other underused motor systems, these muscles are poorly developed in a non-vision-dependent species, the naked mole-rat (McMullen et al. 2010). An even more extreme example of functional extraocular muscle adaptation occurs in the billfish, whose enlarged dorsal (superior) extraocular muscles serve as heat-generating organs to maintain the brain warmer than the environment (Block and Franzini-Armstrong 1988).

The branchiomeric muscles control jaw movement, facial expression, and laryngeal and pharyngeal function. Jaw (masticatory) muscles are important for feeding and sound production; together with some neck muscles, they arise from the first and second branchial arches (Kuratani 2004). These are versatile muscles that are adapted to diverse jaw articulation plans and feeding behaviors. Mammals are unique in having muscles of facial expression and external ear movement. The pharyngeal and laryngeal muscles in mammals are mostly responsible for coordinated swallowing and breathing (i.e., normally, we do not swallow and breathe at the same time). The laryngeal muscles are also involved in airway protective reflexes and sound production (Lang et al. 2002).

A tongue containing voluntary muscles is present in most amphibians, and all reptiles, birds, and mammals, pointing to an association with the terrestrial lifestyle, and adapted for feeding and a sensory role (Iwasaki 2002).

1.4 Craniofacial Muscles and Neuromuscular Disease

The non-locomotor activities of feeding, sound production, breathing, facial expression, and vision are performed by the craniofacial muscles, and are altered by diseases affecting these muscles. For example, strabismus impairs vision secondary to the loss of coordinated eye movements, laryngeal muscle dysfunction alters phonation and airway protective reflexes, facial muscle paralysis affects facial expression and mastication. In addition, structural factors such as craniofacial shape and the position of its muscles predispose to conditions such as obstructive sleep apnea, which affect humans almost exclusively, apparently because of the morphological adaptations required for speech (Davidson 2003).

The aforementioned examples are commonly the specific craniofacial consequences of systemic neuromuscular diseases. Of greater interest and perhaps more significant is the divergent response of at least some craniofacial muscles to major neuromuscular disorders. The most extensively studied example are the extraocular muscles, which are spared by Duchenne muscular dystrophy and other dystrophies affecting the dystrophin–glycoprotein complex, yet targeted by myasthenia gravis and certain mitochondrial myopathies (Kaminski and Ruff 1997; Porter and Baker 1996; Rowland et al. 1997). The extraocular muscles, and more appropriately their motor neurons, are also spared by amyotrophic lateral sclerosis (ALS); at most, patients on long-term ventilator support may eventually have ocular motor involvement (Hayashi et al. 1987). The laryngeal muscles are also insensitive to dystrophin deficiency, the primary defect in Duchenne muscular dystrophy (Fry et al. 2010; Thomas et al. 2008). Oculopharyngeal muscular dystrophy, a trinucleotide repeat disease, affects many craniofacial muscles. Ptosis and dysphagia are the most common signs, and include gaze limitations, tongue weakness and atrophy, dysphonia, and facial weakness. There are also dystonias that target specific subgroups of craniofacial muscles, blepharospasm and laryngeal dystonia being the frequent examples.

It is unquestionably important to elucidate the pathogenesis of each neuromuscular disease in order to cure it or at least minimize its consequences. The contrasting response of craniofacial muscles to major generalized neuromuscular disorders gives us a window into disease-modifying strategies.

1.5 A Preamble

The purpose of this chapter is to set the stage for this book, presenting a brief argument for the uniqueness of the craniofacial muscles. The biology of these small muscles is relatively unexplored. We are just beginning to understand their developmental programs and the features that make them extreme examples of the skeletal muscle stereotype. The following chapters present an extensive survey of our knowledge of the craniofacial muscles by addressing their development, structure, function, and pathology.

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