Pathophysiology of Bolus Inefficiency in Oculopharyngeal Muscular Dystrophy
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Purpose: Although dysphagia is common in individuals with oculopharyngeal muscular dystrophy (OPMD) [1], few studies have studied swallowing efficiency profiles and contributing pathophysiology in this population. We therefore aimed to: 1) delineate global swallowing impairment profiles and 2) determine pathophysiology contributing to swallowing inefficiency in individuals with OPMD.

Methods: 22 OPMD patients were recruited from a university neurology clinic and underwent a standardized videofluoroscopic swallow study (thin, nectar, honey-thick liquids, pudding, cracker). Blinded frame-by-frame analyses were completed and included: Dynamic Imaging Grade of Swallowing Toxicity (DIGEST), Normalized Residue Ratio scale (NRRS), and mechanisms of laryngeal vestibule closure (LVC). Descriptive statistics were performed to characterize swallowing profiles (Aim 1). Between-group ANOVAs were utilized to compare physiologic swallowing parameters across efficiency groups (Aim 2).

Results: Of the 163 individual bolus clips analyzed, 87.9% were characterized as inefficient (NRRSv≥0.07; NRRSp≥0.20). Further, DIGEST analyses indicated efficiency was impacted in 96.2% of the cohort. Inefficient OPMD swallowers demonstrated incomplete LVC attributable to incomplete epiglottic inversion and arytenoid to base of epiglottis contact, resulting in significant vallecular [NRRSv: F(1,108)=12.979, p<0.001] and piriform sinus residue [NRRSp: F(1,113)=40.437, p<0.001].

Conclusions: Swallowing inefficiency was noted in nearly every analyzed swallow and was attributable to poor epiglottic inversion and incomplete laryngeal vestibule closure. Elucidating specific pathophysiology contributing to dysphagia provides rationale for dysphagia treatment including implementation of rehabilitative and compensatory strategies. Strategies targeting epiglottic inversion (eg., via laryngeal elevation and pharyngeal contraction), such as the Mendelsohn maneuver and effortful swallow may be efficacious rehabilitation approaches.