Respiratory insight to congenital muscular dystrophies and congenital myopathies and its relation to clinical trial.

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Abstract
Congenital muscular dystrophies and congenital myopathies represent a heterogeneous group of disorders of the muscle characterized by an early onset of hypotonia and muscle weakness and consequently, a high respiratory morbidity and mortality. The diagnosis and characterization of the weakness of the respiratory muscles is crucial for clinical management of patients and the evaluation of innovative therapies. Routine respiratory evaluation is based on noninvasive volitional tests, such as the measurement of lung volumes, spirometry, and maximal static pressures, which may be difficult or impossible to obtain in young children. Tests using natural maneuvers such as a sniff, a cough or a whistle, are easier to perform and may be more informative in young children. The combination of multiple tests of respiratory muscle function is essential and both increases diagnosis accuracy and the strength of the data in case of clinical trials assessing new therapies for these diseases.

KEYWORDS: Child; Congenital muscular dystrophies; Congenital myopathies; Lung function; Respiratory muscles; Sniff nasal inspiratory pressure

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