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CME article

Chest Wall Abnormalities and their Clinical Significance in Childhood

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EDUCATIONAL AIMS

1. The reader will become familiar with the anatomy and physiology of the thorax
2. The reader will learn how the chest wall abnormalities affect the intrathoracic organs
3. The reader will learn the indications for surgical repair of chest wall abnormalities
4. The reader will become familiar with the controversies surrounding the outcomes of the VEPTR technique

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SUMMARY

The thorax consists of the rib cage and the respiratory muscles. It houses and protects the various intrathoracic organs such as the lungs, heart, vessels, esophagus, nerves etc. It also serves as the so-called "respiratory pump" that generates the movement of air into the lungs while it prevents their total collapse during exhalation. In order to be performed these functions depend on the structural and functional integrity of the rib cage and of the respiratory muscles. Any condition (congenital or acquired) that may affect either one of these components is going to have serious implications on the function of the other. Furthermore, when these abnormalities occur early in life, they may affect the growth of the lungs themselves. The following article reviews the physiology of the respiratory pump, provides a comprehensive list of conditions that affect the thorax and describes their effect(s) on lung growth and function.

INTRODUCTION

The thorax comprises the upper body and it consists of multiple independent bony parts (spinal vertebrae, sternum, ribs) that form the rib cage, and several muscles that cover it from the outside and separate it from the abdominal cavity. The rib cage provides the "scaffolding" on which the muscles lay and connect, whereas the muscles provide stabilization and movement to the rib cage. Although, it is often viewed as just a "protective case" for the various intrathoracic organs (lungs, heart, vessels, esophagus, nerves etc), the thorax is in fact a dynamic apparatus (the so-called "respiratory pump") that performs the actual function of breathing by, generating the movement of air in and allowing or forcing the movement of air out of the lungs). Thus, any condition that results in its malfunction will have significant repercussions on the function of the respiratory system and frequently on other intrathoracic organs as well.

"Chest wall abnormalities" refer to any abnormality that affects the normal structure and/or limit the function of the thorax. Chest wall abnormalities are often referred to as chest or thoracic dysplasias or dystrophies. Although there is a certain overlap between these terms, in this article, dysplasia refers to abnormal anatomic structures that result from the abnormal growth or development of cells or tissues, and it is primarily used for bony abnormalities (e.g. Spondyloepiphyseal dysplasia). The term dystrophy is conventionally used for muscle abnormalities (e.g. muscular dystrophy). This article reviews in detail the common types of chest wall abnormalities and the effects they have on the respiratory system.

TYPES OF CHEST WALL ABNORMALITIES

Many of the chest wall abnormalities (especially the dysplasias) are congenital but they can also develop later in life as a result of a disease (e.g. ankylosing spondylitis) or injury that can be accidental (e.g. flail chest secondary to trauma), or iatrogenic (e.g. thoracotomy). Specific genes and modes of inheritance have been identified for many of the congenital dysplasias, whereas others are assumed to be caused by accidental exposures. The chest wall abnormalities are either primary or part of a syndrome

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Table 1
Conditions associated with abnormalities of the thorax

CONDITION	THORACIC SHAPE	STERNUM	RIBS	SPINE	VERTEBRAE
Aarskog syndrome		PE/PC		(X)	(X)
Achondrogenesis	Small thoracic cage				X
Achondroplasia	Small thoracic cage				X
Allagile Syndrome (arteriohepatic dysplasia)			X		X
Beals syndrome				X	X
Camptomelic Dysplasia	Small thoracic cage		X	(X)	X
Cerebro-Costo-Mandibular syndrome	Small thoracic cage		X		X
Chondroectodermal dysplasia	Small thoracic cage				
Chondroplasia punctate				X	(X)
CHARGE syndrome			(X)		
CHILD syndrome			(X)		
Cleidocranial dysostosis	Small thoracic cage		(X)		
Coffin-Lowry syndrome		PE/PC		X	X
Cohen syndrome				X	X
Diastrophic dysplasia	Small thoracic cage			X	
Down syndrome		(PE/PC)	(X)		(X)
Dyggve-Melchior-Clausen syndrome		PE/PC		X	X
Early Amnion Rupture sequence				X	
Ehlers-Danlos syndrome				(X)	
Escobar syndrome			X	(X)	X
Fetal Hydantoin Effects			X		
Fetal Alcohol syndrome			(X)		(X)
Fetal Aminopterin Effects			(X)		
Fetal Valproate Effect			(X)		X
Fibrochondrogenesis	Small thoracic cage				X
Frontometaphyseal dysplasia					X
Generalized Gangliosidosis syndrome, Type I			X	(X)	X
Gorlin syndrome			X	X	(X)
Haldu-Cheney syndrome				(X)	X
Homocystinuria syntrome		PE/PC		X	
Hunter syndrome			X		
Hurler syndrome			X		X
Hypophosphatasia	Small thoracic cage				
Incontinentia PPigmenti syndrome			X		(X)
Jarcho-Levin syndrome	Small thoracic cage				X
Jeune syndrome	Small thoracic cage				X
Klippel-Feil sequence			(X)	(X)	X
Kniest Dysplasia				X	X
Kozlowski spondyloepiphyseal dysplasia		PE/PC			X
Langer-Giedion syndrome			X		X
Lenz-Majeswski hyperostosis syndrome			X		
Lethal multiple pterygium syndrome	Small thoracic cage				
Marfan syndrome		PE/PC		X	(X)
Marinesco-Sjogren syndrome		PE/PC		(X)	
Maroteaux-mucopolysaccharidosis			X	X	X
Melnick-Needles syndrome	Small thoracic cage	PE/PC	X		X
Meningomyelocele					X
Metaphyseal chondrodysplasias	Small thoracic cage				X
Metatropic Dysplasia	Small thoracic cage			X	X
Morquio syndrome			X	X	X
Mucopolysaccharidosis VII		PE/PC	X		(X)
Multiple synostosis					X
Multiple Lentigines syndrome		PE/PC			
Multiple neuroma syndrome		PE/PC		(X)	
MURCS association			(X)		X
Neurofibromatosis syndrome			(X)	(X)	(X)
Noonan syndrome		PE/PC	(X)	(X)	X
Osteogenesis imperfect	Small thoracic cage	PE/PC			X
Oto-Palato-Digital syndrome	(Small thoracic cage)	PE/PC	X		X
Pallister Hall syndrome			(X)		X
Partial Trisomy 10q syndrome		PE/PC	X	X	(X)
Poland anomaly			X		(X)
Progeria syndrome	(Small thoracic cage)				X
Proteus syndrome			(X)	X	(X)
Pseudoachondroplasia Sponyloepiphyseal dysplasia	Small thoracic cage			X	X
Pyle Metaphyseal Dysplasia			X	(X)	X
Rhizomelic Chondroplasia Punctuta					X
Robinow syndrome			(X)		X
Rokitansky sequence					X
Rubenstein-Taybi syndrome					(X)
Ruvalcaba syndrome		PE/PC		X	X
Sanfilippo syndrome			X		X
Seckel syndrome			X	(X)	
Short rib syndrome	Small thoracic cage				X
Shprintzen syndrome				(X)	
Shwachman syndrome					
Sponyloepiphyseal dysplasia congenita		PE/PC		X	