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INSTITUTO NACIONAL DA ATROFIA MUSCULAR ESPINHAL

# FISIOPATOLOGIA RESPIRATÓRIA NA AME

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FISIOTERAPIA RESPIRATÓRIA NA AME



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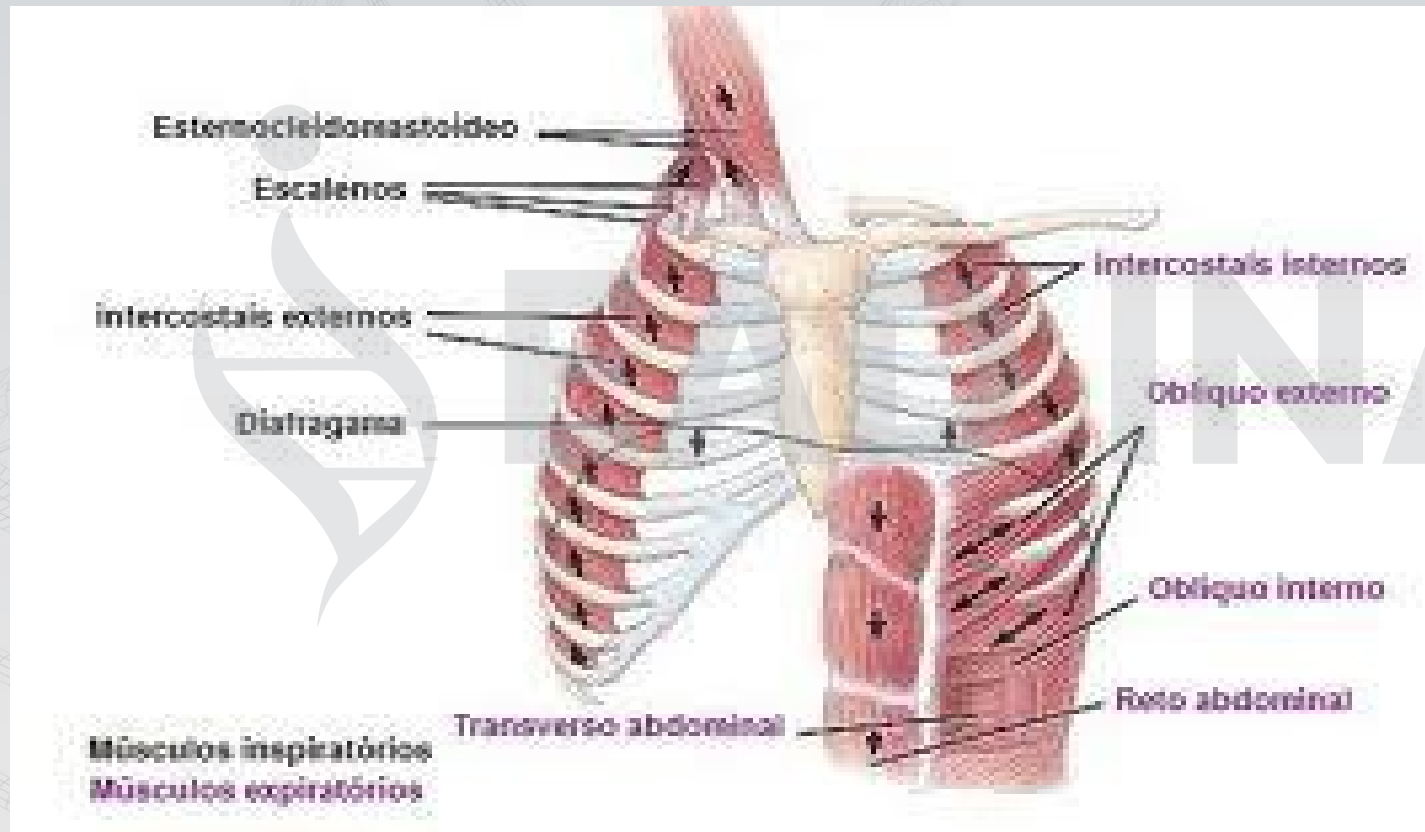


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# MECÂNICA PULMONAR



# FISIOPATOLOGIA

Tipo 1	Tipo 2	Tipo 3
Mais grave	Intermediária	Benigna
Antes dos 6m	6 – 18m	Após 18m
Nunca sentam óbito nos primeiros 24m	Criança senta Podem chegar à vida adulta	Maioria deambula Podem ou não desenvolver sintomas respiratórios
	Complicações respiratórias são comuns	

**Table 1**  
Classification of Spinal muscular atrophy (Adapted from Mercuri 2012 [84] and Finkel 2015 [65]).

	Age of onset	Maximal milestone achieved	Defining clinical features <sup>1</sup>	SMN2 copy number (CN) <sup>2</sup>
Type 0	Birth, prenatal signs	Nil	Severe hypotonia, respiratory failure at birth, congenital contractures	
Type 1A	First 2 weeks of life	Nil	Severe hypotonia, may require respiratory/feeding support during neonatal period, no head control	
Type 1B	After neonatal period but <3 months of life	Never rolls or sits	Severe hypotonia, no head control, paradoxical breathing pattern	1 or 2 CN in 80% of patients 3 CN in 20% of patients
Type 1C	3-6 months	Never rolls or sits	May gain head control, severe weakness (proximal > distal, lower limbs > upper limbs)	
Type 2	7-18 months	Sitting, but never stands	See Finkel 2015	2 CN in 11% of patients, 3 CN in 82% and 4 CN in 7% of patients
Type 3	>18 months	Stands and walks	See Finkel 2015	1 CN in 0% of patients, 2 CN in 4% and 3 or 4 CN in 96% of patients
Type 4	10-30 years	Stands and walks	See Finkel 2015	4 or more CN

Key: CN - Copy number.

<sup>1</sup> Adapted from Finkel 2015 [65]; only symptoms relevant for respiratory failure are listed. For comprehensive description of symptoms see Finkel 2015 [65].

<sup>2</sup> Feldkotter 2002 [4].

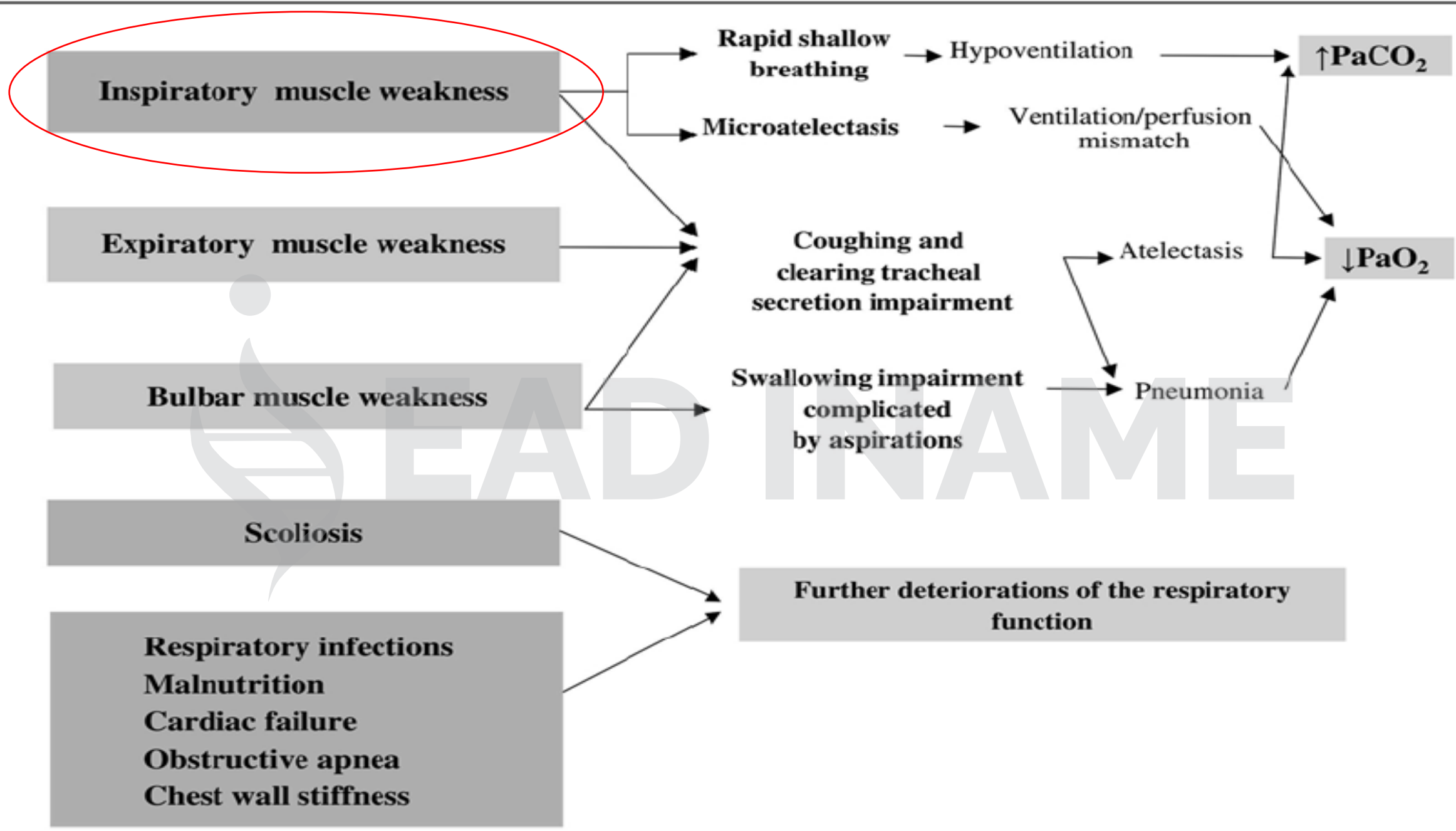


Figure 1.—Respiratory issues in patients with neuromuscular diseases.

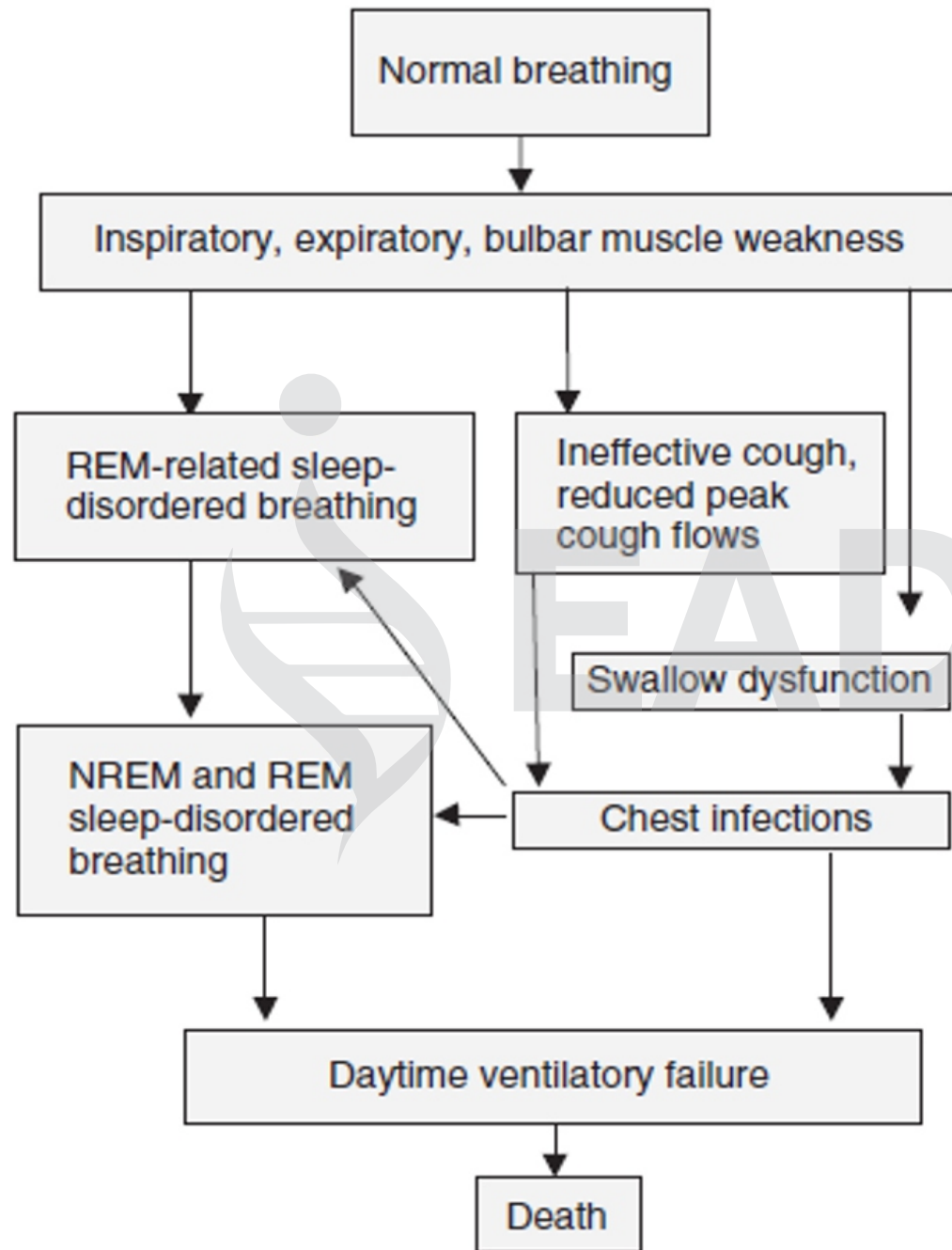
# COMPLICAÇÕES AME TIPO II

**FIGURE 2** Interventions Used to Address Primary Respiratory Complications of SMA Type 2

Primary Respiratory Complications	Definition	Intervention
Ineffective cough with decreased airway clearance	Ineffective cough secondary to reduced vital capacity and tidal volume with inability to forcibly exhale secondary to expiratory muscle weakness	Mechanical Insufflator-Exsufflator Abdominal Thrusts Manual or mechanical chest physiotherapy
Nocturnal Hypoventilation	Shallow breathing secondary to weak inspiratory muscles most often during REM sleep that often manifest as nocturnal desaturations due to gas exchange abnormalities	Bi-Level Positive Airway Pressure
Diminished lung and chest wall development	Inspiratory and expiratory muscle weakness results in alterations of chest wall and lung compliance	Resuscitator bag and mask Mechanical Insufflator-Exsufflator Bi-level positive airway pressure
Pulmonary Infection	Respiratory weakness and poor airway clearance of secretions results in mucus plugging, atelectasis, inflammation, and pneumonia	Mechanical Insufflator-Exsufflator Oral airway suction Manual or mechanical chest physiotherapy Bi-level positive airway pressure Antibiotics and Immunizations as indicated

Note. Based on data from Wang et al. (2007) and Schroth (2009).

Natural History



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# PONTOS PRINCIPAIS DA FISIOPATALOGIA DA AME

FRAQUEZA MM INSPIRATÓRIA	FRAQUEZA MM EXPIRATÓRIA	FRAQUEZA MM BULBAR	DEFORMIDADES TORÁCICAS	OUTROS FATORES
INICIO ATÉ INTRA UTERO	TOSSE PREJUDICADA	DISFAGIA	TORÁX EM SINO	DESNUTRIÇÃO
PREDOMINIO DA FRAQUEZA DA MUSCULATURA INTERCOSTAL EM RELAÇÃO AO DIAFRAGMA	ACUMULO DE SALIVA OU SECREÇÕES	DISARTRIA OU ANARTRIA	TORAX EM EXCAVATUM	SIALORREIA EXCESSIVA
RESPIRAÇÃO PARADOXAL	INCAPACIDADE DE MANTER AS VIAS AEREAS PÉRVIAS	BRONCOASPIRAÇÃO	TORAX EM CARINATUM	DISAUTONOMIA
INSUFLAÇÃO INADEQUADA DOS PULMÕES	ROLHAS E TAMPÕES MUCOSOS	NECESSIDADE DE VIA ALTERNATIVA DE ALIMENTAÇÃO	ESCOLIOSE E CIFOSE	INFECÇÕES RECORRENTES
FALÊNCIA VENTILATÓRIA HIPOXEMIA SECUNDARIA A HIPERCAPNIA	PNEUMONIA	PNEUMONIA	DESVANTAGEM MECÂNICA PARA O FLUXO DE AR	NARCOSE E ÓBITO



EAID INAME

