Manifestazioni Nazionali UILDM 2016
Centro Ge.Tur. - Lignano Sabbiadoro (Udine)
12-14 maggio 2016
Giornata Scientifica – 13 maggio

Malattie Muscolari: Aspetti Nutrizionali

Complicanze gastrointestinali e disfagia nelle malattie muscolari

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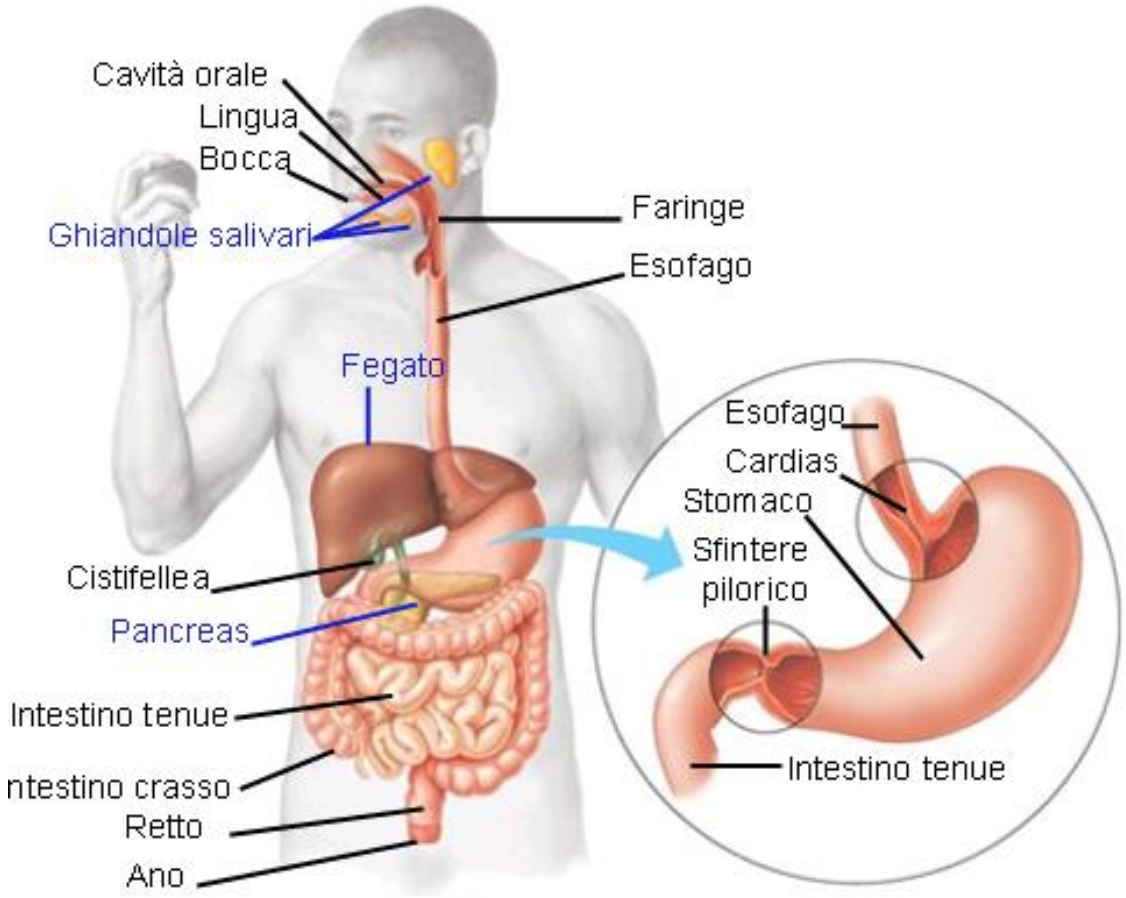
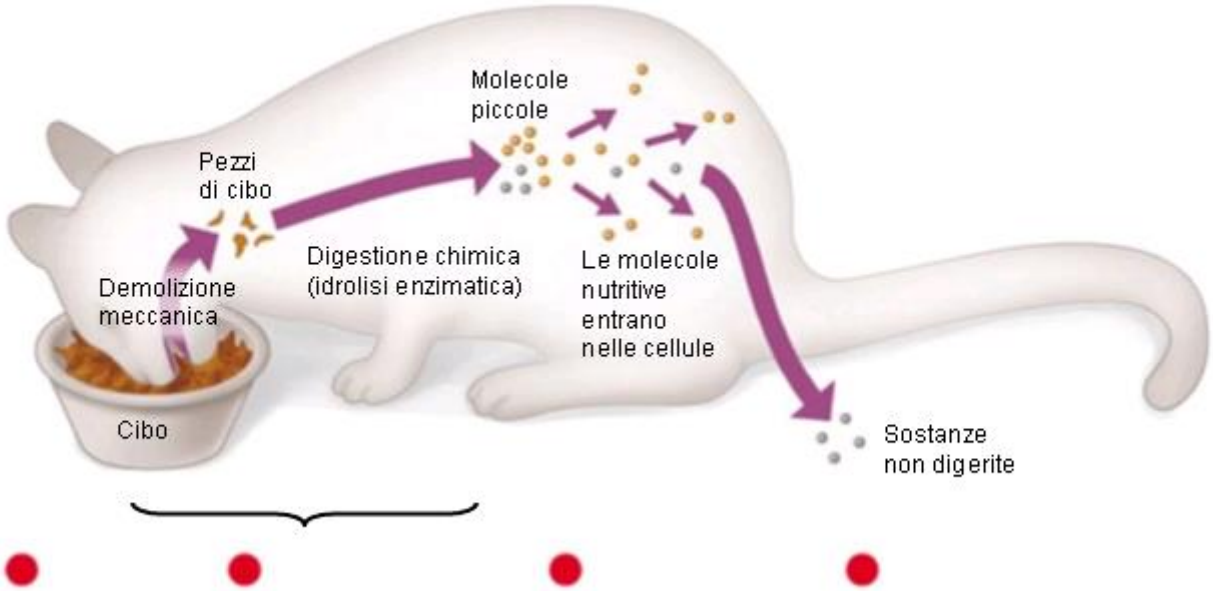
Università di Torino

Piccolo ripasso di anatomia: Il sistema digerente



Piccolo ripasso di fisiologia: il processo alimentare



Bocca, faringe: ingestione (masticazione + deglutizione)

Esofago e stomaco: transito e digestione

Intestino tenue e crasso: transito e assorbimento

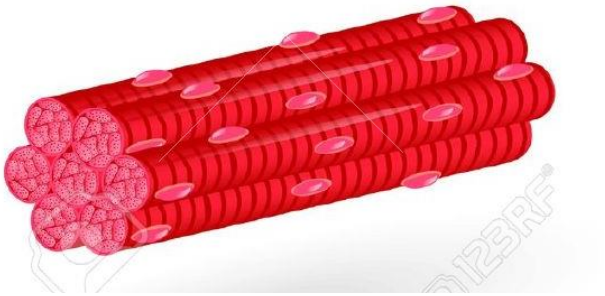
Intestino retto e sfinteri: eliminazione

Ci alimentiamo per….

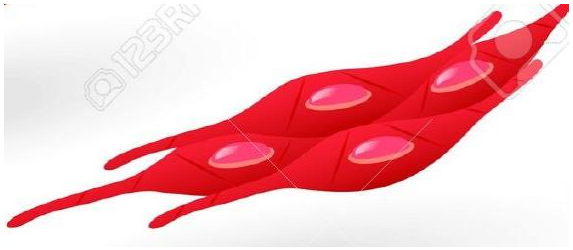
- Introdurre le sostanze nutritive
- Soddisfare il senso di fame
- Soddisfare il piacere del gusto e dei sapori
- Partecipare a un atto sociale e conviviale
- … quindi le complicazioni gastrointestinali hanno un forte impatto sullo stato di salute e sulla qualità di vita

Malattie muscolari e sistema GI: solo una questione di muscoli?

Muscolo scheletrico: innervato da motoneuroni, <u>movimenti volontari</u>

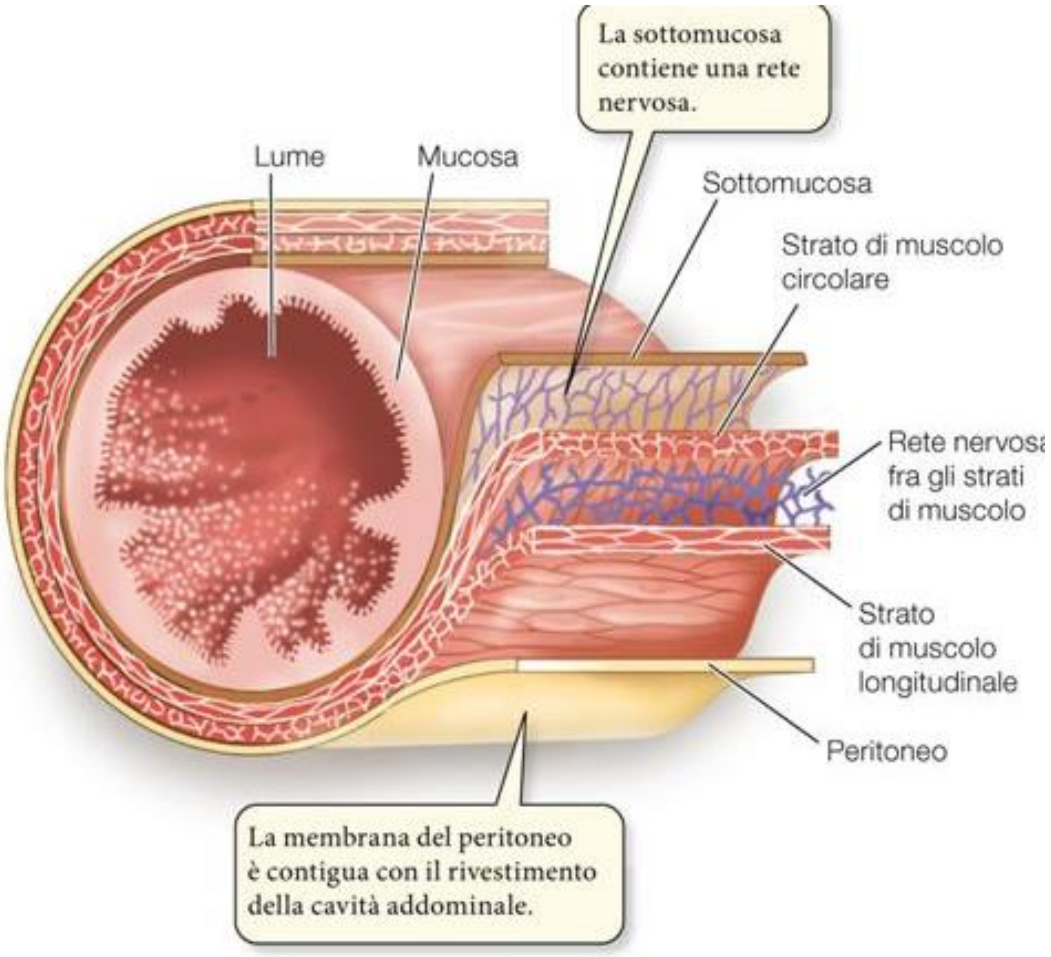


Muscolo cardiaco: contrazione spontanea, indipendente; influenzata da sistema vegetativo autonomo



Muscolo liscio: meno organizzato, può avere <u>contrazione spontanea</u> (peristalsi) o mediata da <u>neuroni del sistema nervoso autonomo</u>

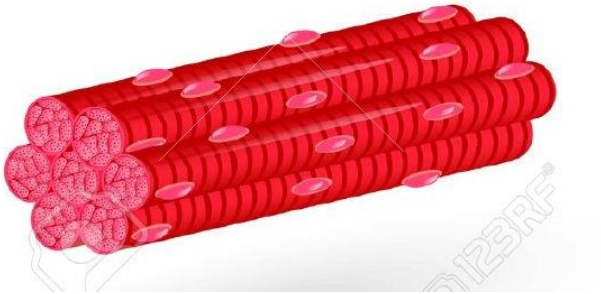
La struttura a ‘tubo cavo’ del sistema digerente

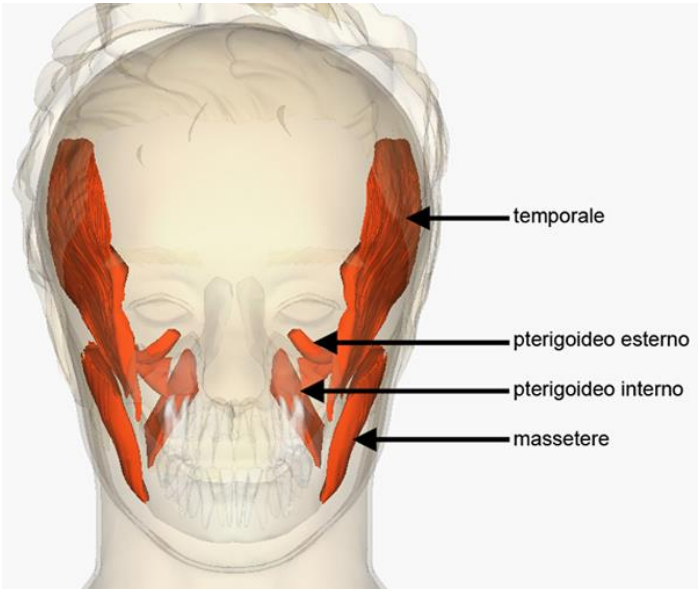


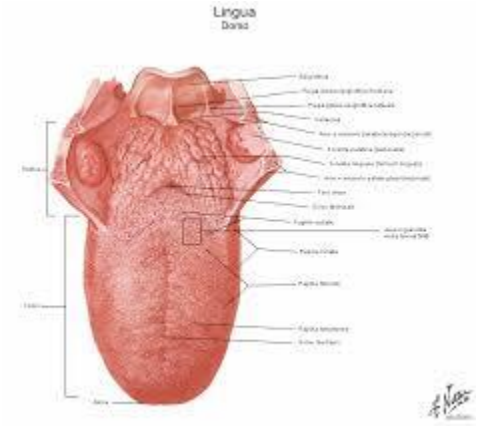
Malattie muscolari e alterazioni gastrointestinali: quale relazione?

- I muscoli scheletrici hanno un ruolo parziale ma importante per il funzionamento GI
- In alcune MM i muscoli scheletrici correlati al tratto GI vengono coinvolti in modo primario rispetto ad altri, in altre compaiono solo in fasi più avanzate
- In altre MM il muscolo liscio può essere compromesso per alterazioni genetiche di proteine o enzimi in comune con il muscolo scheletrico
- In alcune forme di MM anche l'innervazione viene compromessa
- La ridotta mobilizzazione, alterazioni metaboliche, alterazioni osteo-articolari aggiungono ulteriori complicazioni

La masticazione

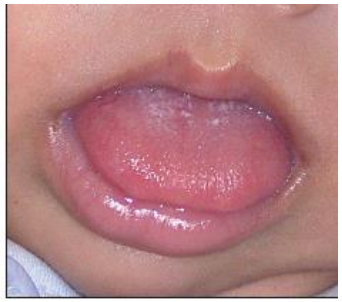






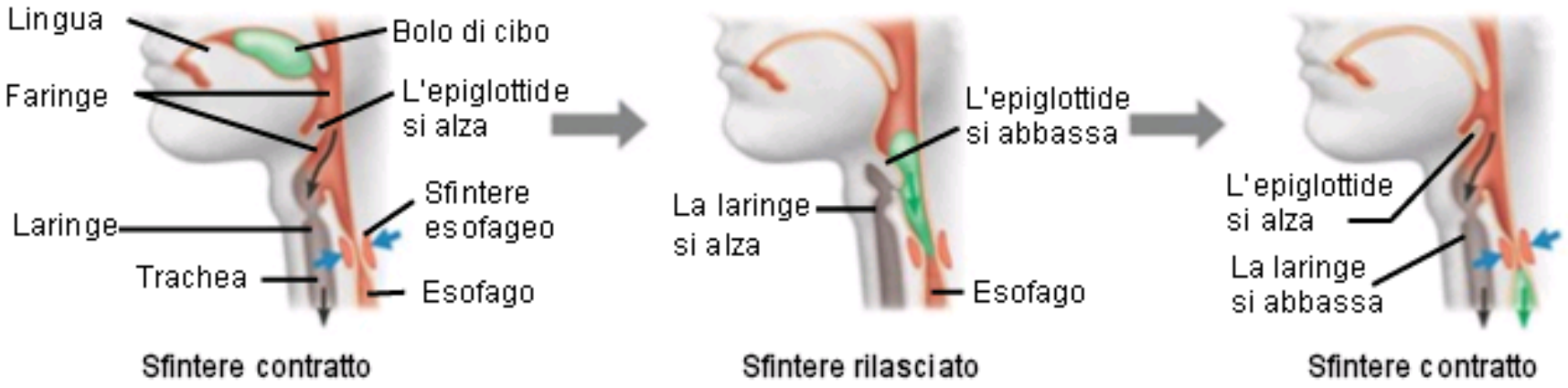
Malattie muscolari con compromissione della masticazione

- Distrofia miotonica (> congenita)
- Miopatie congenite (malocclusioni)
- Malattia di Pompe infantile (macroglossia)
- SMA
- DMD più grandi
- Distrofia facio-scapolo-omerale



Trattamento: modificare la consistenza del cibo

La deglutizione



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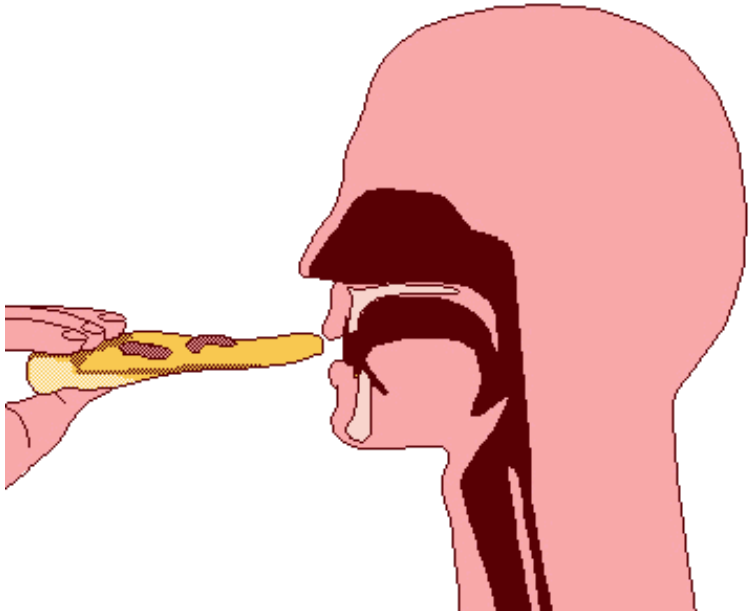


Malattie muscolari con compromissione della deglutizione (disfagia)

- Distrofia oculo-faringea
- Miopatie mitocondriali
- Distrofia miotonica
- Miopatie infiammatorie
- DMD più grandi e rare DMP (laminopatie)
- SMA
- Miastenie congenite
- Malattia di Pompe infantile (nuovo fenotipo)
- Miopatie congenite

Malattie muscolari con compromissione della deglutizione (disfagia)

- Disturbo causato anche da numerose affezioni neurologiche centrali e periferiche (miastenia gravis, SLA, m. Parkinson, neuropatie periferiche, disautonomie, SM, malattie neurodegenerative avanzate, es. MSA)
- complessa diagnosi differenziale; alto rischio di polmoniti ab-ingestis

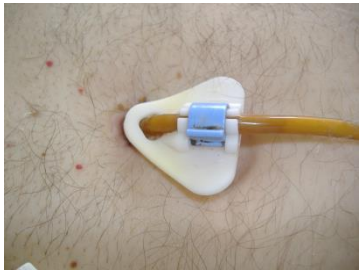
- Progressivo calo ponderale, disidratazione
- Frequente causa di decesso!

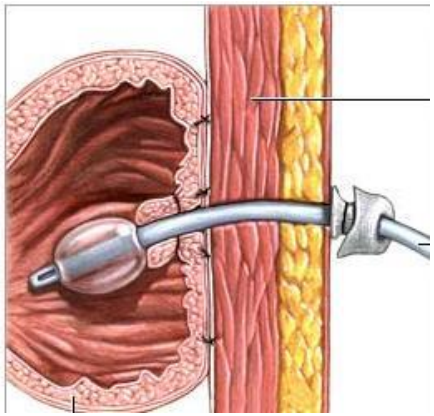
Malattie muscolari con compromissione della deglutizione (disfagia)

- Trattamento: consistenza dei cibi modificata (cremosa, evitare doppie consistenze); posizione del capo; esercizi →
- sondino temporaneo →
- PEG/RIG →
- nutrizione parenterale (EV)
- Per OPMD: miotomia cricofaringea

Nutrizione enterale

- PEG: gastrostomia endoscopica percutanea





- RIG: gastrostomia percutanea radiologica
- Aumento delle necessità assistenziali, riduzione del rischio ab ingestis

La progressione esofagea e la motilità gastrica

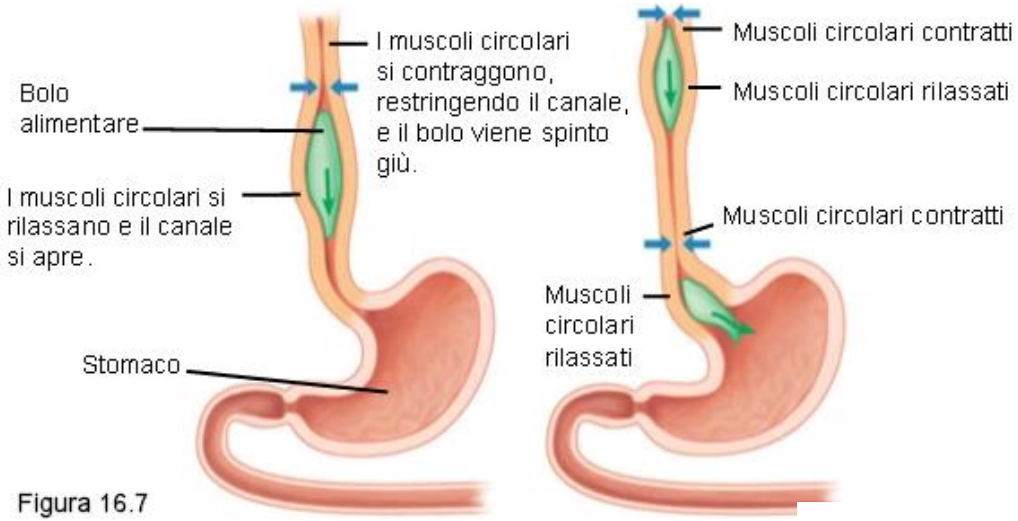


Figura 16.7

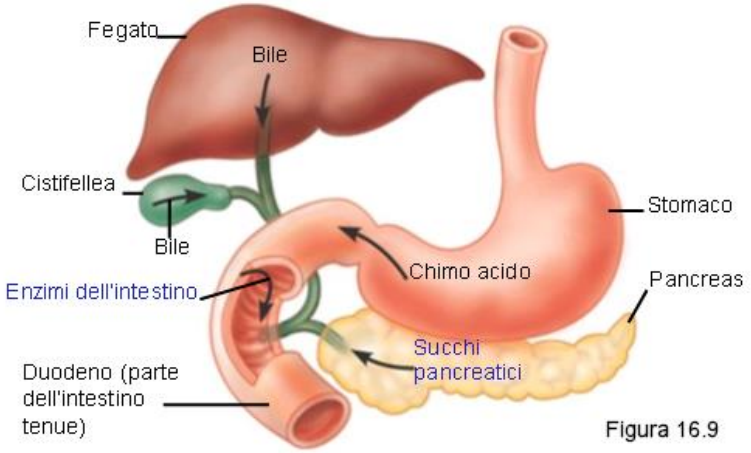
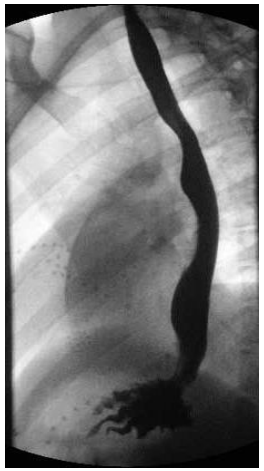


Figura 16.9

Malattie muscolari con disturbi del transito esofageo

- Comuni nella popolazione generale, sono particolamente accentuati nelle distrofie miotoniche e nelle collagenopatie (acalasia)
- Causano reflusso notturno, dolore toracico, dolore epigastrico, disfagia, alitosi



Malattie muscolari con disturbi del transito esofageo

Trattamento:

- antiacidi per tamponare il pH dei succhi gastrici
- procinetici per regolare la motilità dell'esofago

Malattie muscolari con ipocinesia e atonia gastrica

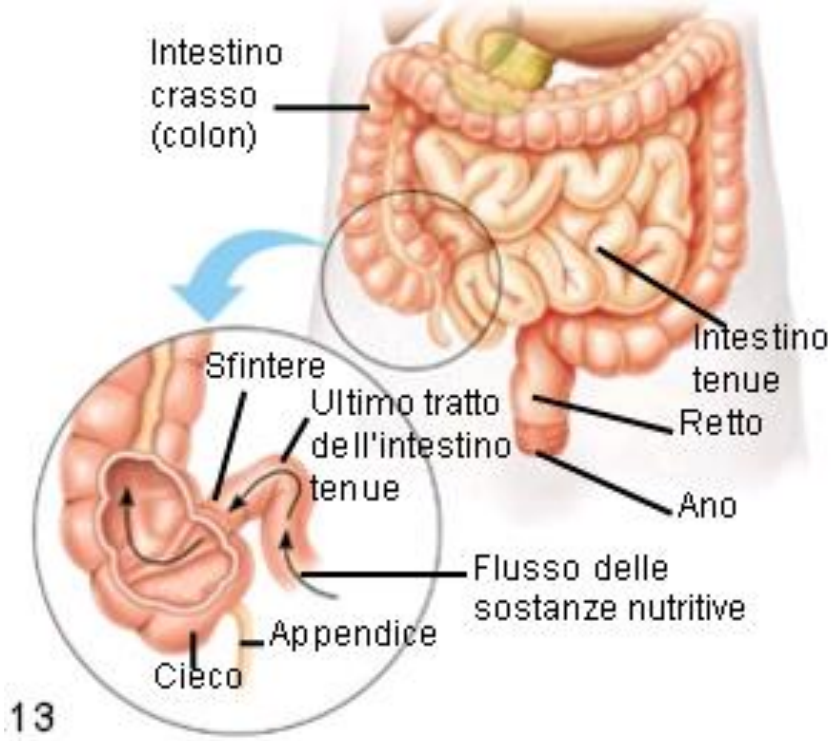
- DMD più grandi e alcune DMP, soprattutto in NIV
- Distrofia miotonica
- (SMA)
- Sintomi vegetativi, nausea, inappetenza, scialorrea, ipotensione;
-  attenzione al rischio di insufficienza respiratoria acuta per ridotta escursione diaframmatica

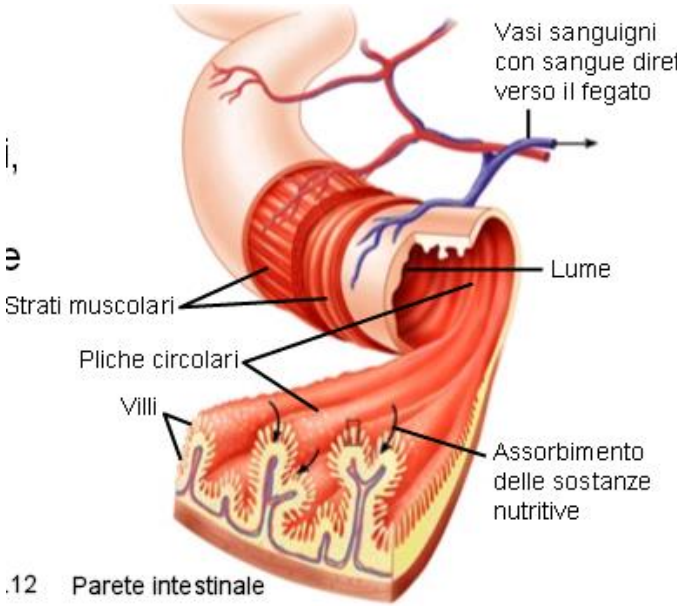
Malattie muscolari con ipocinesia e atonia gastrica

Trattamento:

- pasti piccoli e frequenti, no bevande gasate
- manovre per favorire la fuoriuscita dell'aria, come il cambiamento di posizione e massaggi; se PEG, sfiatare la valvola
- antiacidi
- inibitori di pompa ionica
- procinetici (ad esempio domperidone, eritrocina).
- In acuto: sondino!!!

L’assorbimento e l’espulsione





Malattie muscolari con alterazione del transito intestinale

- Disturbo molto comune nelle patologie con ridotta mobilizzazione e nelle neuropatie in genere
- Megacolon → DMD, distrofie miotoniche, mitocondriopatie
- Sindrome da pseudo-ostruzione cronica intestinale:
  - Miopatie mitocondriali (MNGIE)
  - DMD e alcune DMP in età avanzata
  - Distrofia miotonica
- Sintomi: dilatazione addominale; coliche ricorrenti e intense; alvo alternante (stipsi e diarrea alternate); tenesmo.

Malattie muscolari con alterazione del transito intestinale

Trattamento estemporaneo:

- Idratazione locale
- Rimozione dei fecalomi
- Introduzione di abbondanti liquidi
- intervento chirurgico (se occluso)
- no lassativi osmotici.

Trattamento preventivo:

dieta povera di scorie e ricca di liquidi;
cicli di antibiotici per regolare la flora intestinale

Cicli di fermenti lattici (per sostituire i batteri ad alta fermentazione)

Neuromuscular disorder	Oral-phase involvement	Pharyngeal-phase involvement	Esophageal-phase involvement	Other GI manifestations
Oculopharyngeal muscular dystrophy	Mild to moderate	Severe	Absent	Absent
Myotonic dystrophy	Moderate	Moderate	Moderate	Gallstones, intestinal pseudo-obstruction
Duchenne dystrophy	Mild	Moderate	Mild to moderate	Gastric dysmotility and dilatation, intestinal pseudo-obstruction
Inflammatory myopathies	Mild to moderate	Moderate to severe	Mild to moderate	Heartburn, nausea/vomiting, diarrhea or constipation, and fecal incontinence
Mitochondrial myopathies	Mild	Moderate	Mild	Intestinal pseudo-obstruction, rare
Nemaline rod myopathy	Moderate	Moderate	Absent	
Myasthenia gravis	Moderate	Moderate	Absent	
Lambert-Eaton myasthenic syndrome	Mild	Mild to moderate	Absent	
Inflammatory neuropathies	Moderate	Moderate to severe	Absent or mild	
Bulbospinal muscular atrophy	Mild	Moderate	Absent	
Amyotrophic lateral sclerosis	Moderate	Severe	Absent	

Jaradeh S, GI motility online 2006

LA ‘CLASSIFICA’

1. Distrofia miotonica
2. DMD ‘grandi’
3. Miopatie mitocondriali/miopatie infiammatorie

Available online at www.sciencedirect.com

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Neuromuscular Disorders ■■ (2016) ■■–■■

www.elsevier.com/locate/nmd

Dystrophic changes in masticatory muscles related chewing problems and malocclusions in Duchenne muscular dystrophy

L. van den Engel-Hoek [a,*], I.J.M. de Groot [a], L.T. Sie [b], H.W. van Bruggen [c], S.A.F. de Groot [a], C.E. Erasmus [d], N. van Alfen [d]

THE JOURNAL OF PEDIATRICS • www.jpeds.con (J Pediatr 2016;171:183-8).

Constipation in Duchenne Muscular Dystrophy: Prevalence, Diagnosis, and Treatment

Dror Kraus, MD, PhD[1], Brenda L. Wong, MD[1], Paul S. Horn, PhD[1], and Ajay Kaul, MD[2]

Table I. Patient characteristics of a cohort of 120 patients with DMD

Patient characteristics	Age <18 y	Age >18 y	Total	*P* value (<18 y vs >18 y)
Number of participants (% of total)	95 (79.2)	25 (20.8)	120	n/a
Age in y (average ± SD)	10.8 ± 2.8	21.3 ± 3.3	12.9 ± 5.2	<.01
Age range in y	5-17	18-30	n/a	n/a
Ambulatory (%)	80 (84.2)	5 (20)	85 (70.8)	<.01
North Star Ambulatory Assessment (average ± SD)	21.6 ± 8.1	n/a	21.3 ± 8.1	-
Functional Mobility Scale	2.33 ± 1.6	5.44 ± 1.87	2.98 ± 2.08	<.01
Steroid treatment (%)	91 (95.7)	21 (84)	112 (91.8)	NS
Calcium supplementation (%)	51 (56)	15 (60)	66 (56.9)	NS

Symptomatic Treatment for Constipation

A survey of symptomatic treatments for constipation revealed several unexpected findings. Of 56 patients that fulfilled criteria for constipation, only 24 patients (43.6%) received treatment for constipation (Figure). Twenty-three of the 24 patients received polyethylene glycol 3350; one patient received a compound consisting of sodium picosulfate, magnesium oxide, and anhydrous citric acid. In the treated group, 13 patients (54.1%) were still constipated according to Rome-III criteria, even after correction for patients, whose positive result on the questionnaire was due to a previous diagnosis of constipation.

[Intervention Review]

Interventions for dysphagia in long-term, progressive muscle disease

Katherine Jones[1a], Robert DS Pitceathly[2,3], Michael R Rose[1], Susan McGowan[4], Marguerite Hill[5], Umesh A Badrising[6], Tom Hughes[7]

[1]Department of Neurology, King's College Hospital NHS Foundation Trust, London, UK. [2]Department of Basic and Clinical Neuroscience, Institute of Psychiatry, Psychology and Neuroscience, King's College London, London, UK. [3]MRC Centre for Neuromuscular Diseases, UCL Institute of Neurology and National Hospital for Neurology and Neurosurgery, London, UK. [4]National Hospital for Neurology and Neurosurgery, London, UK. [5]Department of Neurology, Morriston Hospital, Swansea, UK. [6]Department of Neurology, Leiden University Medical Centre, Leiden, Netherlands. [7]Neurology, University Hospital of Wales, Cardiff, UK

Cochrane Library

Cochrane Database of Systematic Reviews

Interventions for dysphagia in long-term, progressive muscle disease (Review)

Jones K, Pitceathly RDS, Rose MR, McGowan S, Hill M, Badrising UA, Hughes T

J Neurol (2015) 262:2225–2231
DOI 10.1007/s00415-015-7836-y

ORIGINAL COMMUNICATION

Development of a new scale for dysphagia in patients with progressive neuromuscular diseases: the Neuromuscular Disease Swallowing Status Scale (NdSSS)

Ayako Wada[1] · Michiyuki Kawakami[2] · Meigen Liu[2] · Eri Otaka[2] · Atsuko Nishimura[2] · Fumio Liu[2] · Tomoyoshi Otsuka[1]

Table 1 Neuromuscular disease swallowing status scale (NdSSS)

Level 1	Tube feeding with saliva suctioning in the oral cavity necessary. A patient can neither discharge nor swallow saliva
Level 2	Tube feeding without suctioning. Although a patient cannot take anything by mouth, can discharge and/or swallow saliva
Level 3	Tube feeding with occasional oral intake. A patient sometimes take orally for the fun, not for nourishment
Level 4	Totally orally fed and tube-free with supplemental nutrients, such as enteral solution. A patient usually take supplemental nutrients by mouth although don't take general food
Level 5	Totally orally fed with easy-to-swallow food and supplemental nutrients, such as enteral solution. A patient sometimes/often take supplemental nutrients by mouth
Level 6	Totally orally fed with only easy-to-swallow food. A patient eat foods processed in a mixer and drink thicken water
Level 7	Totally orally fed with no difficulties. A patient eat without something difficult to eat
Level 8	Totally orally fed with no restrictions. A patient eat all kinds of food

"Tube feeding" modes include feeding by a nasogastric tube, a gastrostoma tube, and so on

DISABILITY AND REHABILITATION, 2016
http://dx.doi.org/10.3109/09638288.2015.1111434

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RESEARCH PAPER

Dysphagia in Duchenne muscular dystrophy: practical recommendations to guide management

Michel Toussaint[a], Zoe Davidson[b,c], Veronique Bouvoie[a], Nathalie Evenepoel[a], Jurn Haan[a] and Philippe Soudon[a]

[a]Acute Neurorespiratory Rehabilitation Unit, Neuromuscular Excellency Centre and Centre for Home Mechanical Ventilation, Vrije Universiteit Brussel-Inkendaal Rehabilitation Hospital, Vlezenbeek, Brussels, Belgium; [b]Department Nutrition and Dietetics, Monash University, Melbourne, Australia; [c]Murdoch Childrens Research Institute, Melbourne, Australia

➤ IMPLICATIONS FOR REHABILITATION

- Little guidance is available for the management of dysphagia in Duchenne dystrophy.
- Food can penetrate the vestibule, accumulate as residue or cause aspiration.
- We propose recommendations and an algorithm to guide management of dysphagia.
- Penetration/residue accumulation: prohibit solid food and promote intake of fluids.
- Aspiration: if cough augmentation techniques are ineffective, consider tracheostomy.

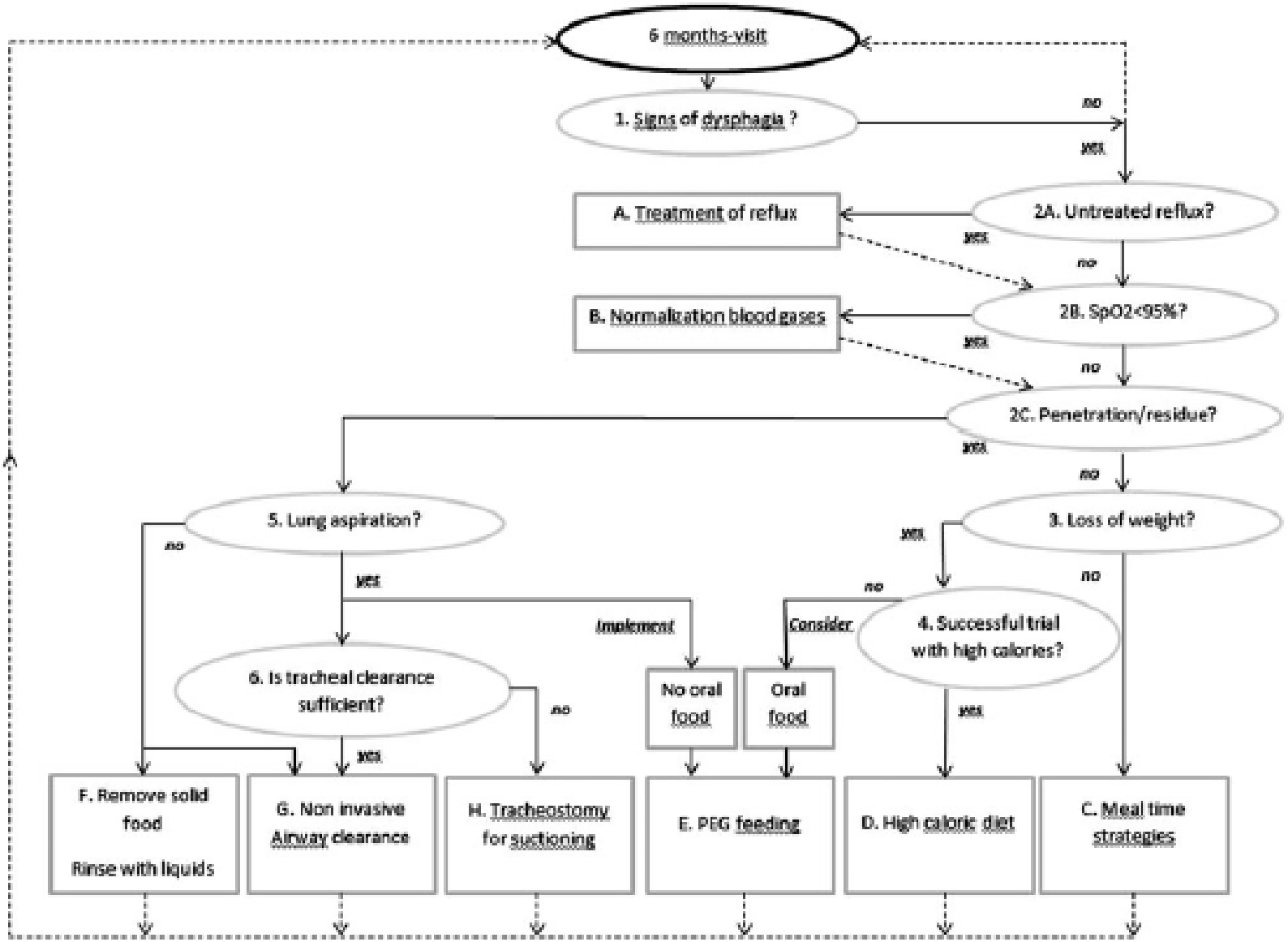

Figure 2. Clinical algorithm to guide management of dysphagia in the DMD population. See explanations in the text.

Liliana Vercelli, Valentina Ponzalino, Sara Bortolani

Loredana Chiadò-Piat, Silvia Boschi

(SCDU Neurologia 1, Dipartimento di Neuroscienze)

Federica Ricci, Enrica Rolle, Francesca Rossi

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