SERVIZIO SANITARIO REGIONALE
EMILIA-ROMAGNA
Azienda Unità Sanitaria Locale di Bologna

Istituto delle Scienze Neurologiche
Istituto di Ricovero e Cura a Carattere Scientifico

DISTROFIE MUSCOLARI: ASPETTI NUTRIZIONALI

Luisa Zoni

UOC Dietologia e Nutrizione Clinica
AUSL Bologna

"l'uomo è quello che mangia"

Ludwig Feuerbach

3ª Edizione

P.zza XX Settembre Pordenone

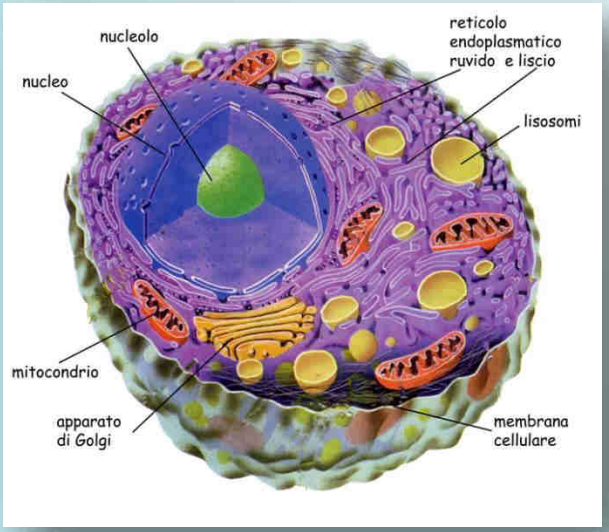
Risparmio Energetico, Bioedilizia,
Gestione dei Rifiuti, Mobilità Sostenibile,
Economia Consapevole, Prodotti a KM Zero

dal 3 al 27 maggio 2012

Incontri, dibattiti, teatro, film, laboratori

NELLA PRATICA

- Ogni molecola ha azioni complesse e diverse all'interno delle nostre cellule
- Le codifiche genetiche possono essere alterate
- Gli apporti possono essere inadeguati



- Cibi e farmaci possono interferire tra di loro
- Le scelte personali impattano sullo stato di salute

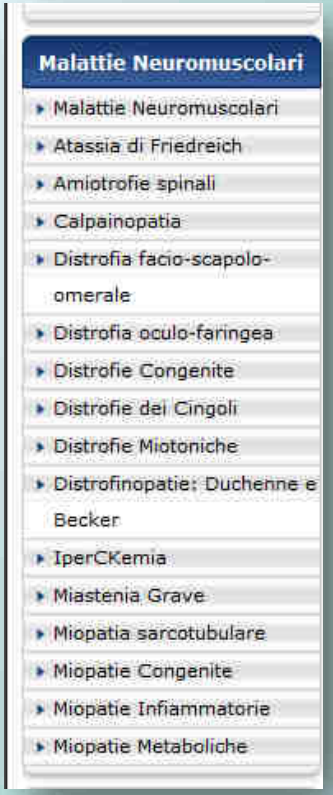

Uso limitato:
Carni rosse e Burro;
Cereali raffinati: pane, **pasta** e riso;
Patate; **Bevande gassate** e Dolci;
Sale
Alcool con moderazione e non per tutti
Latticini (1 o 2 porzioni al giorno) o alternative di Calcio + Vitamina D
Integratori specifici per alcune/ molte persone
Noci, **semi**, Legumi e Tofu
Pesce Pollame e Uova
Frutta e **Verdura**
Grassi e oli vegetali **salutari**
Cereali integrali
ACQUA naturale, **Attività fisica** giornaliera e Controllo del peso

MALATTIE NEURO-MUSCOLARI

- **NEUROPATIE PERIFERICHE** (**polineuropatie** su base tossica, metabolica, carenziale, infiammatoria o **multineuropatie** su base vascolare
- **MALATTIE DEL MOTONEURONE** (SLA, …)
- **MIOPATIE** (genetiche distrofiche, dismetaboliche, infiammatorie): singolarmente rare, globalmente numerose

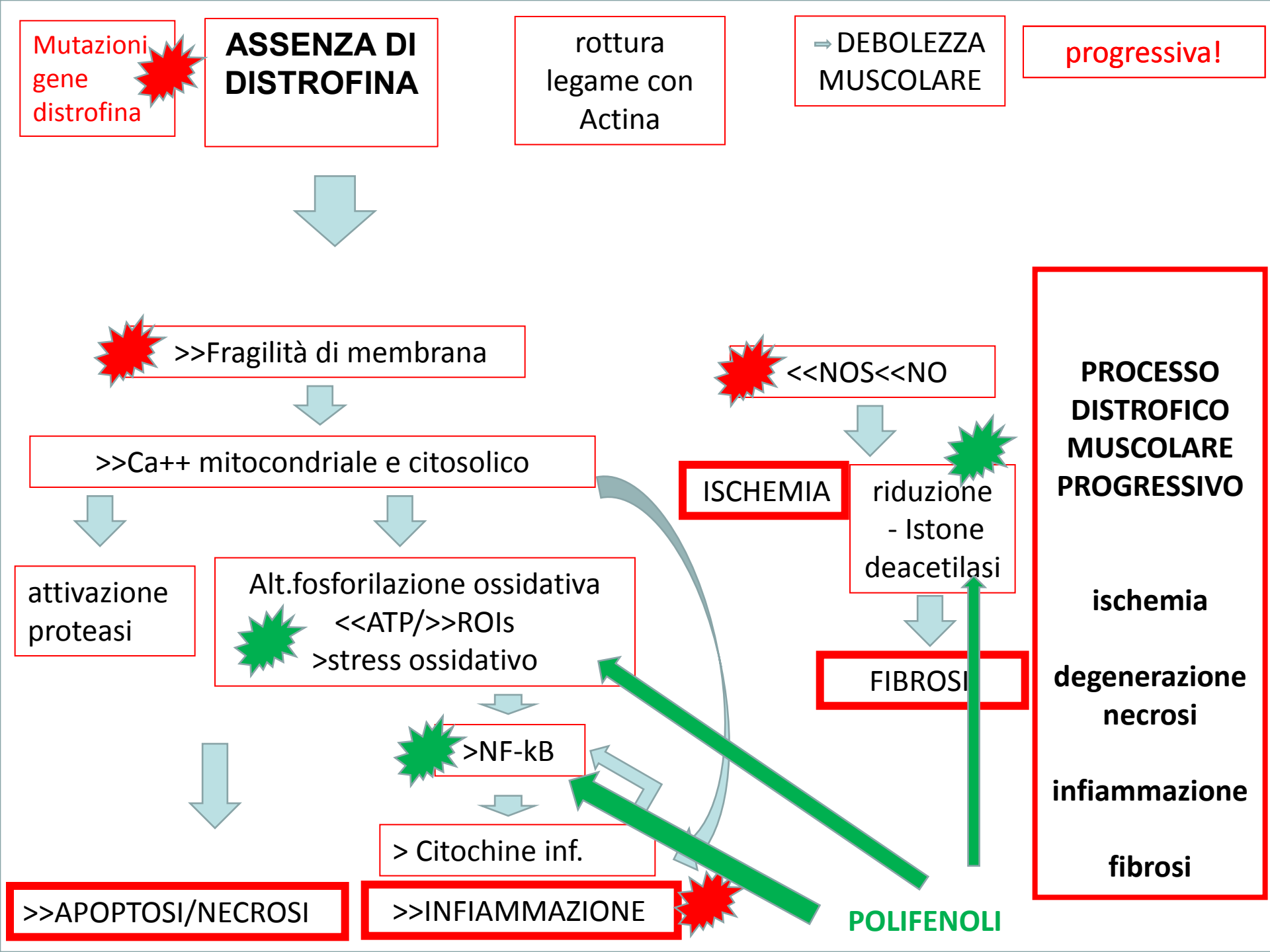
UN FILONE PARTICOLARE

- Le **MALATTIE NEUROMUSCOLARI DISTROFICHE**
  - Distrofia di Duchenne (1/3500 nati vivi)
  - Distrofia di Beker
  - Distrofia Miotonica di Steinert (1/10000 n v)
  - Distrofia Facio-Scapolo-Omerale (1/20000 n v)
  - Atrofia Spinale (SMA) tipo 1-2-3
  - SLA, sporadica o eredo-familiarre (2-3/100000 abitanti)



COME INTERVIENE LA NUTRIZIONE

- **La nutrizione gioca un ruolo rilevante per alcune patologie neurologiche e neuro-muscolari, a stadi diversi:**
  - **Esclusione di nutrienti o supplementazioni mitrate**
  - **Distribuzione Proteica Differenziata (DPD)**
  - **Prevenzione/cura della malnutrizione**
  - **Aumento nutrienti anti-infiammatori, anti-ossidanti, funzionali**
  - **Prevenzione/gestione obesità e dismetabolismi**
  - **Modifiche di consistenza per la disfagia**
  - **Nutrizione Artificiale**


Mutazioni gene distrofina
ASSENZA DI DISTROFINA
rottura legame con Actina
⇒DEBOLEZZA MUSCOLARE
progressiva!
>>Fragilità di membrana
>>Ca++ mitocondriale e citosolico
attivazione proteasi
Alt.fosforilazione ossidativa
<<ATP/>>ROIs
>stress ossidativo
>NF-kB
> Citochine inf.
>>APOPTOSI/NECROSI
>>INFIAMMAZIONE
<<NOS<<NO
ISCHEMIA
riduzione - Istone deacetilasi
FIBROSI
POLIFENOLI
PROCESSO DISTROFICO MUSCOLARE PROGRESSIVO
ischemia
degenerazione necrosi
infiammazione
fibrosi

PUNTI CHIAVE

- Età infantile (= potenziale evolutivo di crescita staturale e sviluppo puberale)
- Valutazione dello stato nutrizionale (antropometrica, bioumorale, alimentare, clinica) x prevenzione della malnutrizione
- Problematiche specifiche della patologia interferenti sulla nutrizione
- Effetti dell'iponutrizione in quella determinata patologia

THE OFFICIAL JOURNAL OF THE BRITISH DIETETIC ASSOCIATION
Journal of Human Nutrition and Dietetics

Journal of Human Nutrition and Dietetics

2009

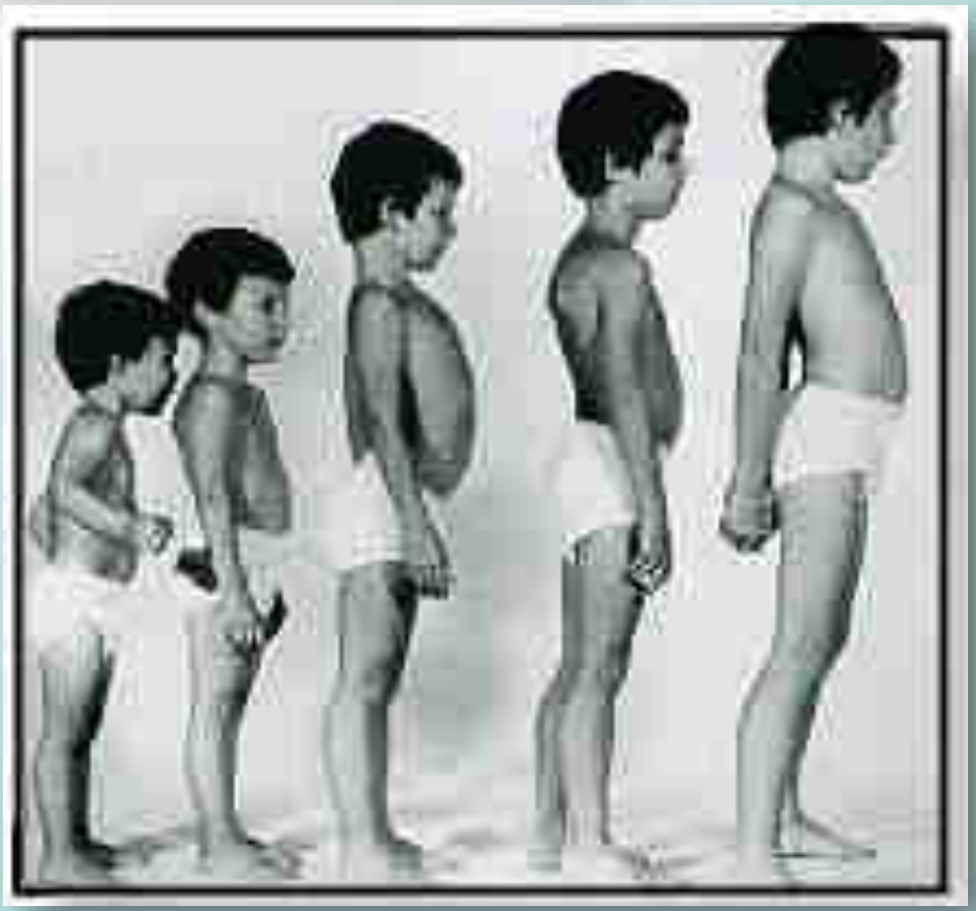
REVIEW

A review of nutrition in Duchenne muscular dystrophy

Z. E. Davidson & H. Truby

The University of Queensland, Children's Nutrition Research Centre, School of Medicine, Queensland, Australia

"Boys with DMD can move between the spectrum of over to under Nutrition within their shortened lifespan"



QUALE PESO

In a large cohort of patients with DMD, 44% of patients were obese by the age of 12 y, and 44% of patients had malnutrition by the age of 18 y.

J. Nutritional Assessment in Duchenne Muscular Dystrophy.Dev Med Child Neurol 1993;35:1074–82

- **Obesity affects more than half of 13-year-old boys ….**
- **… On the other hand, adolescents and adults with DMD frequently experience feeding difficulties and …**
- **… Underweight affects more than half of 18-year-old**

British Journal of Nutrition (2011), 105, 1486–1491

↑ peso inizia verso 8 anni, picco a 13 anni e poi ↓ in alcuni pazienti

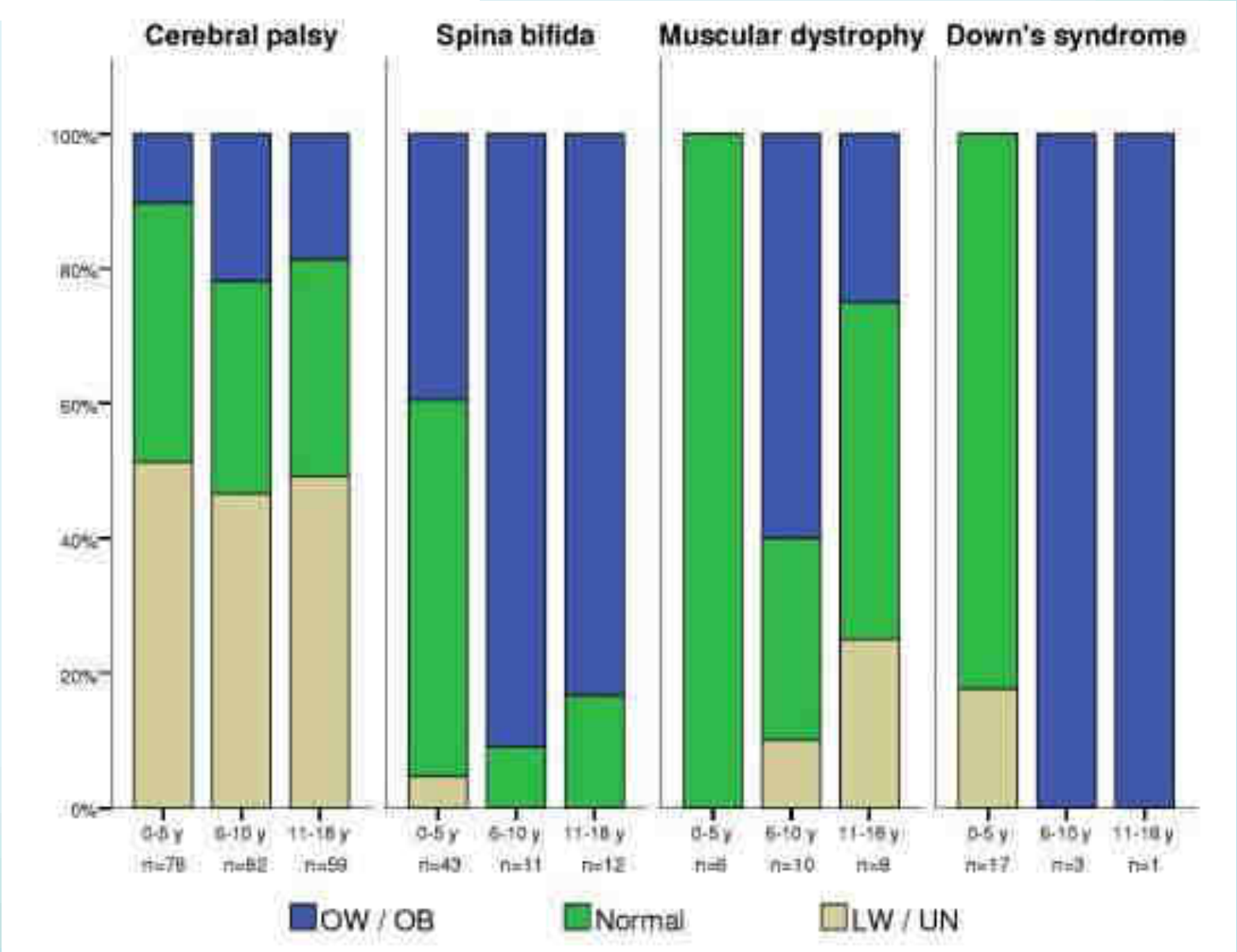
Vega-Sanchez et al. BMC Research Notes 2012, 5:218
http://www.biomedcentral.com/1756-0500/5/218

BMC Research Notes

RESEARCH ARTICLE Open Access

Weight-based nutritional diagnosis of Mexican children and adolescents with neuromotor disabilities

Per età



Eur J Clin Nutr. 2014 Aug;68(8):892-7. doi: 10.1038/ejcn.2014.93. Epub 2014 May 14.

Observations of body mass index in Duchenne muscular dystrophy: a longitudinal study.

Davidson ZE[1], Ryan MM[2], Kornberg AJ[2], Sinclair K[3], Cairns A[3], Walker KZ[4], Truby H[4].

Abstract

BACKGROUND/OBJECTIVES:

Nutritional issues that are associated with Duchenne muscular dystrophy (DMD) remain poorly understood. The aim of this analysis was to describe and explore longitudinal observations of body mass index (BMI) in a cohort of children with DMD.

SUBJECTS/METHODS:

Anthropometric and clinical characteristics were collected retrospectively and longitudinally for boys with DMD seen in two large neuromuscular clinics. BMI Z-scores were determined using the Centers for Disease Control and Prevention reference values for children (2000).

RESULTS:

Medical records (**n=193**) were examined from which 75% were included for analysis. The mean age of the cohort at the time of data collection was 11.9 years, with 72% of patients currently or previously using steroids. The highest prevalence of obesity based on the BMI Z-score was 50% at the age of 10 years. Longitudinally, BMI Z-scores from the age of 2 to 12 years plot approximately one s.d. above the mean, after which there is a marked and progressive decline. BMI gainers were identified for whom BMI Z-score increased by 1.65 units compared with the 0.09 units in non-gainers. **BMI gainers were younger when they had their first BMI measurement (5.9 vs 7.2 years), and this measure was significantly lower compared with the non-gainers (BMI Z-score: 0.04 vs 1.17).** **In this cohort, BMI was associated with age, ambulatory status and lung function**.

CONCLUSIONS:

This study demonstrates that boys with DMD using steroid therapy experience shifts in BMI. A declining BMI appears to be associated with increasing age. Interpretation of growth patterns is limited here by a lack of normative growth references in DMD

frontiers in
AGING NEUROSCIENCE

ORIGINAL RESEARCH ARTICLE
published: 07 October 2014
doi: 10.3389/fnagi.2014.00274

Sarcopenia and sarcopenic obesity in patients with muscular dystrophy

Luciano Merlini[1]*, *Alessandro Vagheggini*[2] and *Daniela Cocchi*[2]

[1] Laboratory of Musculoskeletal Cell Biology, Istituto Ortopedico Rizzoli, Bologna, Italy
[2] Department of Statistical Sciences, University of Bologna, Bologna, Italy

Altogether, our study suggests the relevance of a proper evaluation of body composition in MD and we propose to use, both in research and practice, the measurement techniques that has already been demonstrated effective in aging sarcopenia.

La perdita di massa muscolare dovuta alla patologia, che comporta la sostituzione del muscolo con tessuto fibro-adiposo, viene aggravata dalla malnutrizione spesso presente in questi pazienti

Bach JR. (1999). *Guide to the Evalution and Management of Neuromuscolar Diseases*. Philadelphia, Hanley and Belfus, pp 23-34

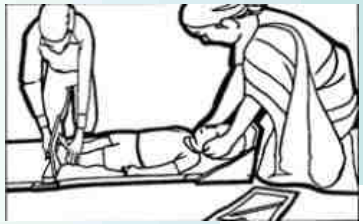
Bach JR, Tippett DC, McCray MM. (1992). *Bulbar disfunction and associated cardiopulmonary considerations in polio and neuromuscolar disease*. J Neuro Rehabil;6:121-8.

Il 70% dei pazienti affetti da DMD e sottopeso necessitavano ulteriormente di un supporto ventilatorio artificiale

VALUTAZIONE ANTROPOMETRICA

COSA FARE

- **Ad ogni controllo periodico (2 v/anno in fase di crescita, 1 v/anno poi) rilevare:**
  - **Peso**
  - **Altezza / Lunghezza**
  - **BMI**
  - **Controllo dei Percentili**
  - **Consensus internazionale nella definizione di rischio di**
    - **Sottopeso se BMI <3°P**
    - **Sovrappeso se BMI >85°P**
    - **Obesità se BMI >95°P**



A new chart for weight control in Duchenne muscular dystrophy

R D GRIFFITHS AND R H T EDWARDS

Muscle Research Centre, Department of Medicine, University of Liverpool, Liverpool

Referenza di curve per la distrofia muscolare di Giffin ed Edwards del 1988.

Ipotizza una perdita muscolare del 4% all'anno.

Non considera la statura dei soggetti.

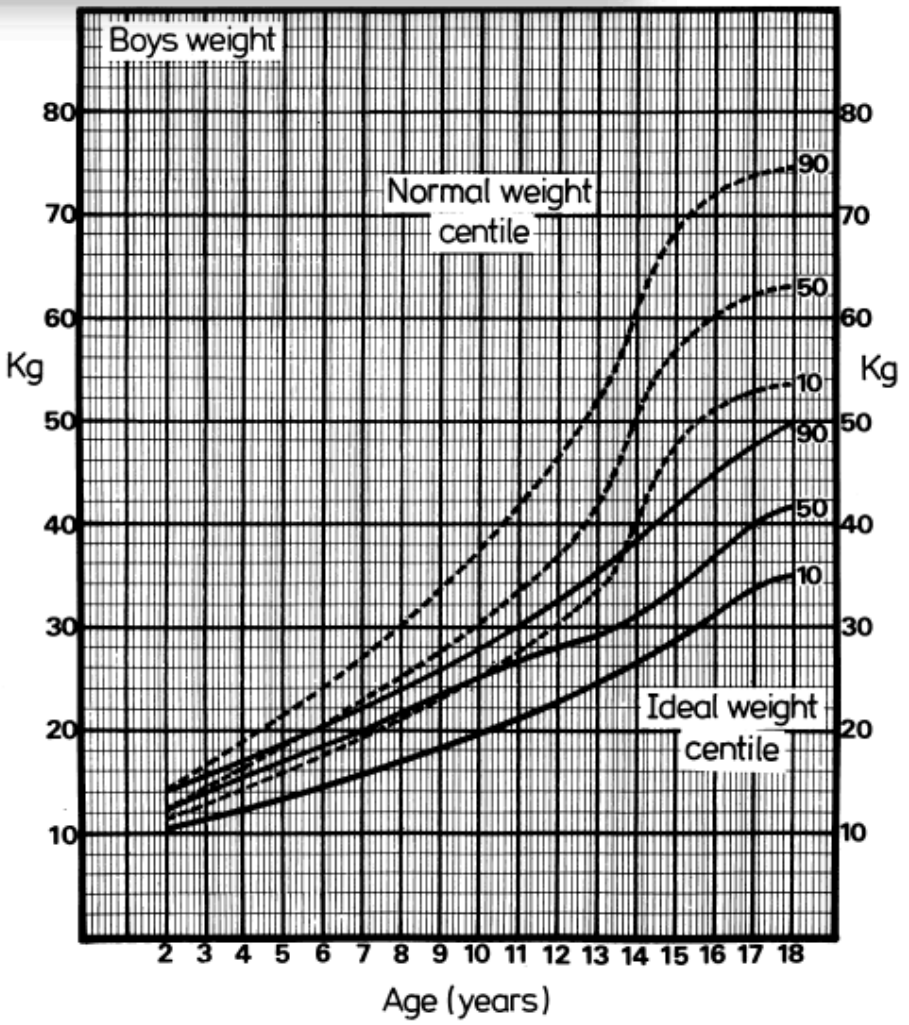


Figure Ideal weight centile chart for boys with Duchenne muscular dystrophy. Based on data from Edwards et al,[1] Edmonds et al,[4] and Tanner and Whitehouse.[3] Assumes a 4% per year decline in muscle bulk.

Nutrition assessment and monitoring

- **Short stature** is a characteristic of boys with DMD (Grade B)
- **BMI** as a screen for obesity is **not accurate** in boys with DMD (Grade C)

CURVE CRESCITA (NO STEROIDI)

J Pediatr. 2013 Dec;163(6):1759-1763.e1. doi: 10.1016/j.jpeds.2013.08.004. Epub 2013 Oct 6.

Patterns of growth in ambulatory males with Duchenne muscular dystrophy.

West NA[1], Yang ML, Weitzenkamp DA, Andrews J, Meaney FJ, Oleszek J, Miller LA, Matthews D, DiGuiseppi C.

Author information

Abstract

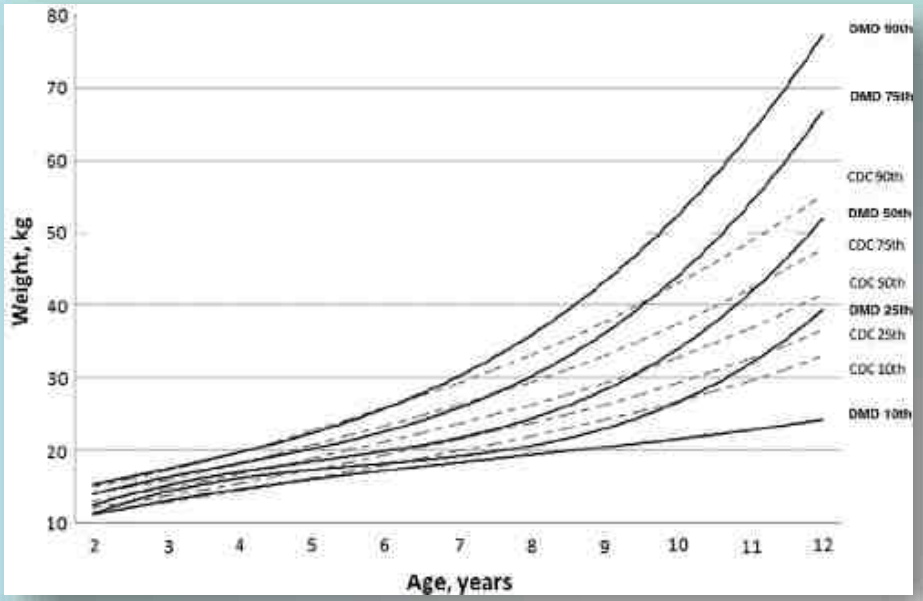
OBJECTIVE: To provide weight-for-age, height-for-age, and body mass index-for-age growth reference standards for ambulatory, steroid-naïve males, ages 2-12 years, with Duchenne muscular dystrophy (DMD) and to compare these growth curves to the 2000 Centers for Disease Control and Prevention growth charts for boys, which serve as references of physical size and growth for the general male pediatric population in the US.

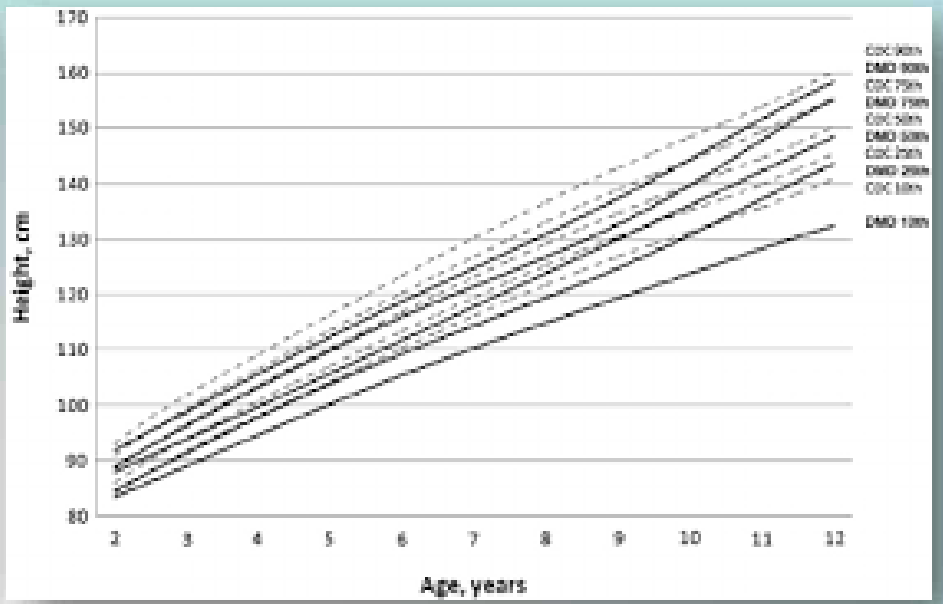
STUDY DESIGN: Through a multi-state population-based surveillance of individuals with muscular dystrophy, a total of 1877 weight and 1544 height measurements ascertained during 1985-2010 from 513 males with DMD were obtained retrospectively from medical record review. Cases were classified as DMD if loss of ambulation occurred before the 12th birthday or, if younger than 12 years and still ambulating, the earliest symptoms of dystrophinopathy occurred before the 6th birthday. Each growth chart was constructed using 5 percentiles: 10th, 25th, 50th, 75th, and 90th. Smoothing procedures were applied in 2 stages to the irregular plots of the empirical percentile values.

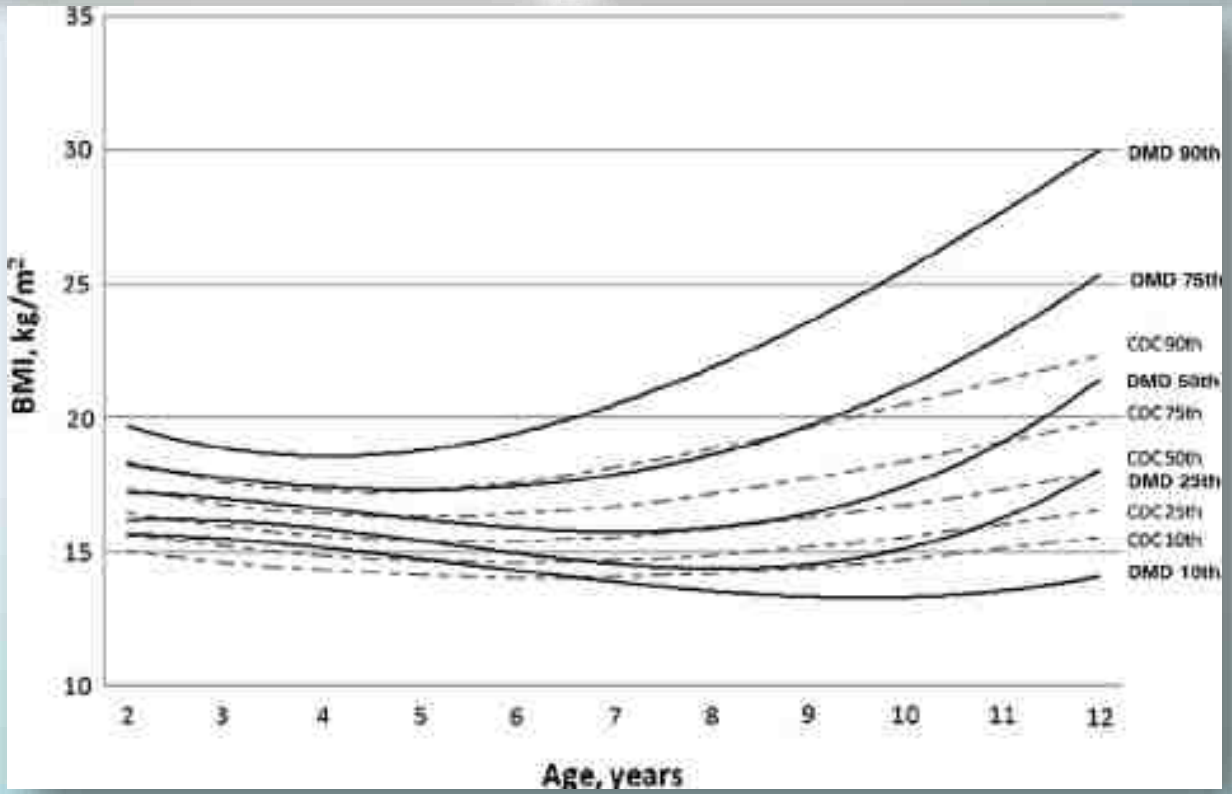
RESULTS: A set of growth curves, derived from a large cohort of male youth with DMD, are presented. These curves demonstrate that DMD males are shorter and tend to the extremes of weight and body mass index compared with the general male pediatric population in the US.

CONCLUSION: Charts representing the pattern of growth in ambulatory, steroid-naïve males with DMD can facilitate monitoring of growth and early detection of unusual growth patterns. Use of these growth standards also will assist in monitoring responses to corticosteroid treatment.

KEYWORDS: BMI; Body mass index; CDC; Centers for Disease Control and Prevention; DMD; DMD/BMD; Duchenne muscular dystrophy; Duchenne muscular dystrophy or Becker muscular dystrophy; MD STARnet; Muscular Dystrophy Surveillance, Tracking, and Research Network


Weight, kg
Age, years
DMD 90th
DMD 75th
CDC 90th
DMD 50th
CDC 75th
CDC 50th
DMD 25th
CDC 25th
CDC 10th
DMD 10th


Height, cm
Age, years


BMI, kg/m²
Age, years
DMD 90th
DMD 75th
CDC 90th
DMD 50th
CDC 75th
CDC 50th
DMD 25th
CDC 25th
CDC 10th
DMD 10th

2013

VALUTAZIONE STRUMENTALE

BIA

- Metodica di **valutazione della composizione corporea** agevole e non invasiva
- Validata come attendibilità generale da studi di composizione corporea isotopici e con pesata idrostatica
- Limiti:
  - Elettrodi non sempre su misura per pedriatici = posizionamento scorretto
  - Assenza di scale di riferimento pediatriche e per patologie
  - Errori di stima anche grossolani
- Valido l'aspetto legato a **cellularità** organismo

Estimating body composition in children with Duchenne muscular dystrophy: comparison of bioelectrical impedance analysis and skinfold-thickness measurement[1–3]

Elise Mok, Laurent Béghin, Pierre Gachon, Christel Daubrosse, Jean-Eudes Fontan, Jean-Marie Cuisset, Frédéric Gottrand, and Régis Hankard

The American Journal of Clinical Nutrition

Conclusions:

Body-composition estimates by BIA are closer to those by WD than are those by ST measurement.

Early detection of fat accumulation and longitudinal monitoring of nutritional care are 2 relevant applications of BIA to prevent obesity and hence lessen the burden of DMD

ASSESSING CHANGE IN BODY COMPOSITION IN CHILDREN WITH DUCHENNE MUSCULAR DYSTROPHY: ANTHROPOMETRY AND BIOELECTRICAL IMPEDANCE ANALYSIS VERSUS DUAL-ENERGY X-RAY ABSORPTIOMETRY.

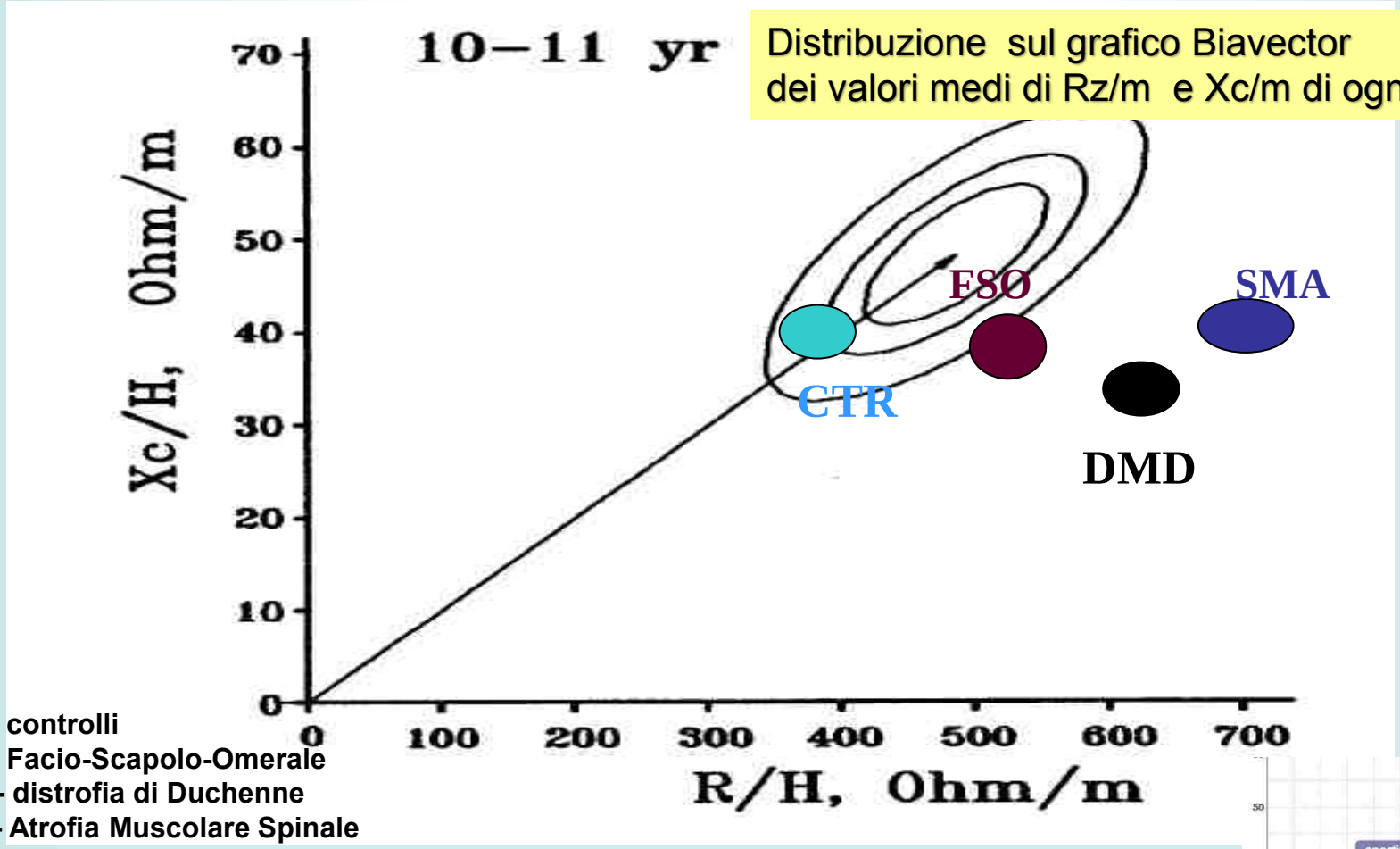
CONCLUSIONS:

BIA method can be used to follow changes in nutritional status of ambulatory DMD children or to evaluate treatment efficacy.

Clin Nutr. 2010

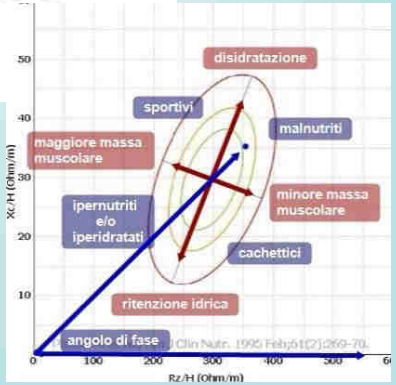
BIVA (ANALISI VETTORIALE)

De Palo et al. Nutrition Volume 16, Number 6, 2000



Distribuzione sul grafico Biavector
dei valori medi di Rz/m e Xc/m di ogni gruppo

CTR – controlli
FSO – Facio-Scapolo-Omerale
DMD – distrofia di Duchenne
SMA – Atrofia Muscolare Spinale



Pediatr Neurol. 2015 Jan;52(1):82-7. doi: 10.1016/j.pediatrneurol.2014.08.008. Epub 2014 Aug 27.

A bedside measure of body composition in Duchenne muscular dystrophy.

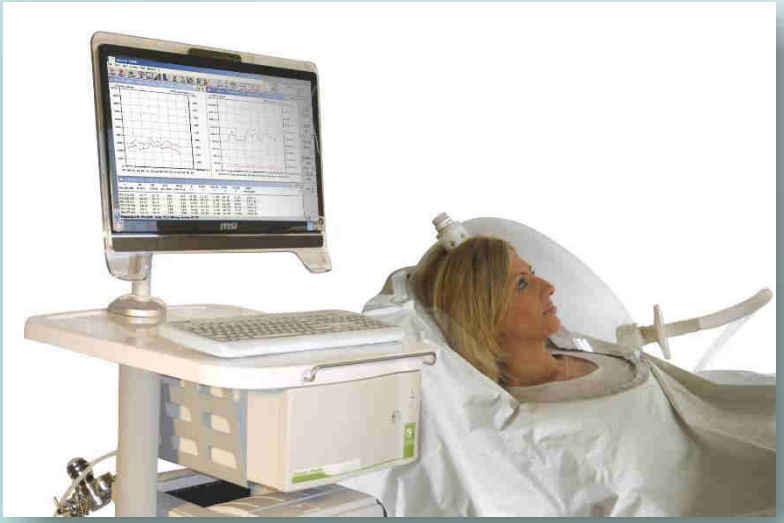
Elliott SA[1], Davidson ZE[2], Davies PS[1], Truby H[3].

BACKGROUND:
In clinical practice, monitoring body composition is a critical component of nutritional assessment and weight management in boys with Duchenne muscular dystrophy. We aimed to evaluate the accuracy of a simple bedside measurement tool for body composition, namely bioelectrical impedance analysis, in boys with Duchenne muscular dystrophy.
METHODS:
Measures of fat-free mass were determined using a bioelectrical impedance analysis machine and compared against estimations obtained from a reference body composition model. Additionally, the use of raw impedance values was analyzed using three existing predictive equations for the estimation of fat-free mass. Accuracy of bioelectrical impedance analysis was assessed by comparison against the reference model by calculation of biases and limits of agreement.
RESULTS:
Body composition was measured in 10 boys with Duchenne muscular dystrophy, mean age 9.01 ± 2.34 years. The bioelectrical impedance analysis machine values of fat-free mass were on average 2.3 ± 14.1 kg higher than reference values. Limits of agreement (based on 95% confidence interval of the mean) were -7.4 to 2.9 kg. There was a significant correlation between the mean fat-free mass and difference in fat-free mass between the bioelectrical impedance analysis machine and the reference model (r = -0.86; P = 0.02) suggesting that the bias was not consistent across the range of measurements. The most accurate predictive equation for the estimation of fat-free mass using raw impedance values was the equation by Pietrobelli et al. (mean difference, -0.7 kg; 95% limits of agreement, -3.5 to 2.0 kg).
CONCLUSIONS:
In a clinical setting, where a rapid assessment of body composition is advantageous, the use of raw impedance values, combined with the equation by Pietrobelli et al., is recommended for the accurate estimation of fat-free mass, in boys with Duchenne muscular dystrophy

CALORIMETRIA

- Valuta i **fabbisogni in funzione della situazione del momento** (risente di peso e composizione corporea, tipo di metabolismo in atto, situazione clinica).

  Non invasiva ma non disponibile in tutti i centri.



European Journal of Clinical Nutrition (2003) 57, 273–278

0954–3007/03 $25.00
www.nature.com/ejcn

ORIGINAL COMMUNICATION

Body composition and energy expenditure in Duchenne muscular dystrophy

MC Zanardi[1], A Tagliabue[1]*, S Orcesi[2], A Berardinelli[2], C Uggetti[3] and A Pichiecchio[3]

[1]*Applied Health Sciences Department, University of Pavia, Pavia, Italy;* [2]*Child Neurology Department, Regional Referring Centre for Neuromuscular Disorders in Childhood and Adolescence, Neurological Foundation C. Mondino, Pavia, Italy; and* [3]*Neuroradiology Department, Neurological Foundation C. Mondino, Pavia, Italy*

***CONCLUSIONS*: Our results do not demonstrate a low REE in DMD boys; on the contrary REE per kg of FFM is higher than normal, probably due to the altered FFM composition.**

We suggest that the development of obesity in DMD children is not primarily due to a low REE but to other causes such as a reduction in physical activity and or overfeeding.

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AGING NEUROSCIENCE

ORIGINAL RESEARCH ARTICLE
published: 17 November 2014
doi: 10.3389/fnagi.2014.00315

Nutritional status evaluation in patients affected by Bethlem myopathy and Ullrich congenital muscular dystrophy

Silvia Toni*[1], *Riccardo Morandi*[1], *Marcello Busacchi*[1], *Lucia Tardini*[1], *Luciano Merlini*[2], *Nino Carlo Battistini*[1] *and Massimo Pellegrini*[1]

[1] *Laboratory of Nutrition and Lifestyle, Department of Diagnostic, Clinical and Public Health Medicine, Modena, Italy*
[2] *Laboratory of Musculoskeletal Cell Biology, Istituto Ortopedico Rizzoli, Bologna, Italy*

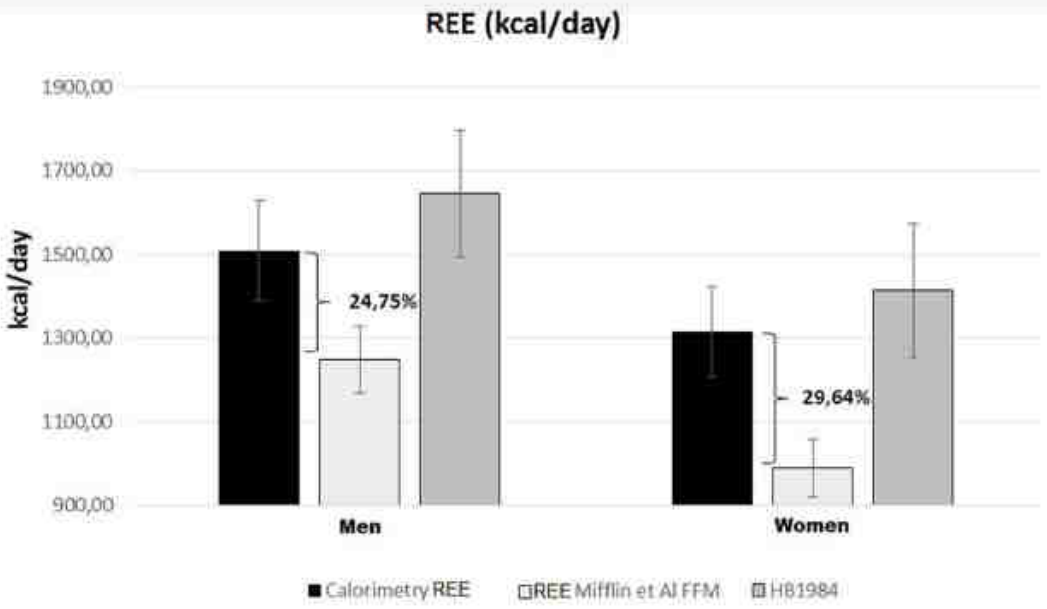
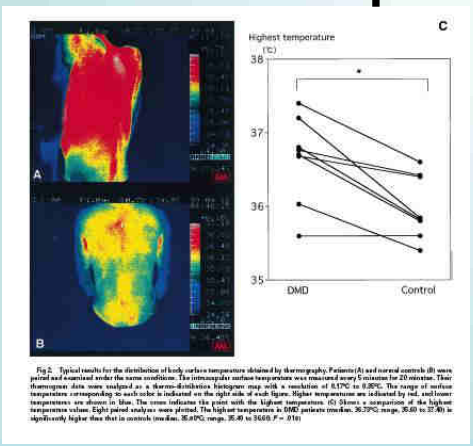


FIGURE 1 | Predicted and measured REE in UCMD and BM patients grouped by sex. Mean REE measured with indirect calorimetry (black rectangles); REE estimated with FFM-based Mifflin equation (Mifflin et al., 1990) (white rectangles) or Harris-Benedict) revised equation (Roza and Shizgal, 1984) (gray rectangles).

PARADOXICAL WEIGHT LOSS WITH EXTRA ENERGY EXPENDITURE AT BROWN ADIPOSE TISSUE IN ADOLESCENT PATIENTS WITH DUCHENNE MUSCULAR DYSTROPHY

Shigeko Satomura, Ichiro Yokota, Katsunori Tatara, Etsuo Naito, Michinori Ito, and Yasuhiro Kuroda

These results suggest that paradoxical activation of the sympathetic nervous system may accelerate the production of heat in brown adipose tissue (BAT) and increase the level of energy consumption in patients, and that adolescent DMD patients may require greater caloric intake than expected to maintain body weight, which is important to improve the prognosis of their respiratory function.



COMMENTARY
Adipocyte 3:3, 221–223; July/August/September 2014; © 2014 Landes Bioscience

Brown fat and vascular heat dissipation

The new cautionary tail

Amy Warner and Jens Mittag
Department of Cell & Molecular Biology; Karolinska Institutet; Stockholm, Sweden

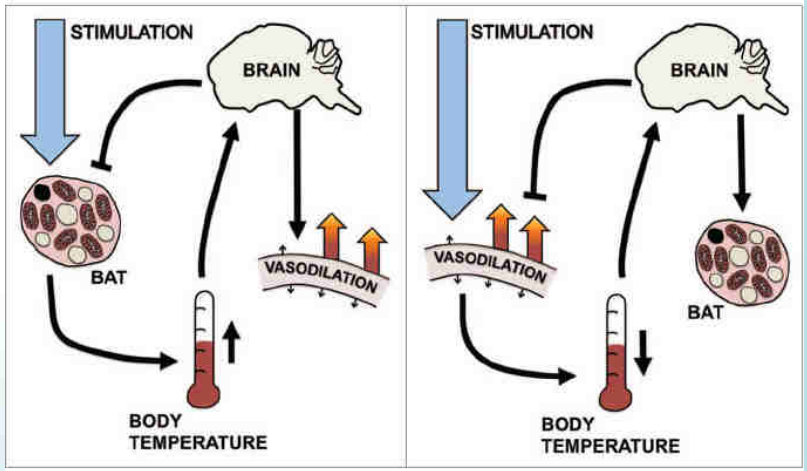
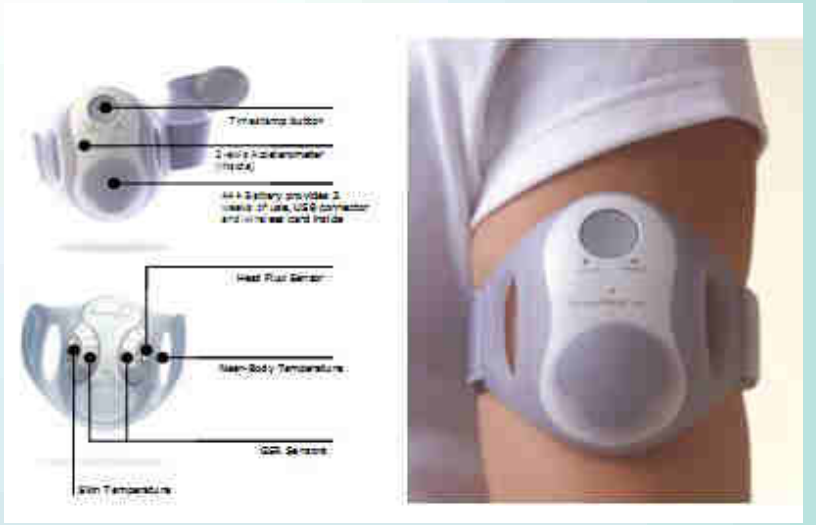


Figure 1. Manipulation of thermoregulation to increase energy expenditure. Current strategies predominantly center on the direct stimulation of brown adipose tissue (left panel). This will cause an increase in body temperature, which in turn triggers temperature sensitive circuits in the brain to initiate compensatory cooling mechanisms, including vasodilation in skin and tail as well as a reduction in the sympathetic activation of BAT. To trigger energy expenditure through thermoregulation, an alternative strategy could be the stimulation of heat dissipation, e.g., by the use of α1-adrenergic antagonists enforcing vasodilation (right panel). This will cause a decline in body temperature, which in turn via central temperature sensing circuits will elicit sympathetic activation of BAT, thereby increasing energy expenditure.

ARMBAND

- **Rilevazione con device applicato al corpo in grado di effettuare multiple rilevazioni fisiologiche:**
  - **Dispendio energetico,**
  - **n° di passi**
  - **Temperatura corporea**
  - **Ore di sonno**
  - **……**



- **Validato scientificamente**

INDAGINI BIOUMORALI

INDICI NUTRIZONALI

- Insieme di misurazioni che valutano lo stato proteico ed ematologico circolante:
  - **Albumina** (emivita 21 gg)
  - **Prealbumina** (emivita 1,9 gg)
  - **Linfociti** (emivita lunga)
  - Emoglobina (se c'è stato anemico)
  - Rapporto AAE/AANE ↓
  - Urea e creatinina ↓ o =
  - Vitamine = o ↓
  - Macro e micro minerali = o ↓
  - **Transferrina** (emivita 8 gg)
  - **RBP** (emivita 12 h)
  - T-linfociti ↓
  - 3-metil-istidina escreta ↑
  - Na ↓ (ma raro sotto soglia)
  - AGE ↓ o =
- **Valori NN non escludono malnutrizione** (specie marasma a lenta insorgenza) per conservazione PT viscerali a scapito di quelle muscolari/strutturali

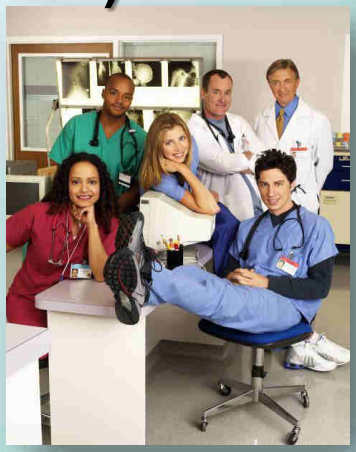
VALUTAZIONE INTAKE ALIMENTARE

INDAGINE ALIMENTARE

- **Diario alimentare (3-7 gg)** → valutazione di tipo, entità, frazionamento dei cibi assunti; **quesito: i fabbisogni vengono coperti (macro e micronutrienti)?**
- **Modalità di preparazione dei pasti** → valuta texture somministrata, densità calorica, problemi connessi con la fornitura; **quesito: la forma del cibo è adeguata?**
- **Modalità di somministrazione dei pasti** → sede, postura, interazione con ambiente, movimenti, rumori (tosse, gorgoglii, alterazione voce), rigurgiti o vomito, **durata** del pasto; **quesito: il pasto crea problemi?**
- **Apporto di liquidi** → valutazione di quantità, tipo, momento di somministrazione e problemi connessi con l'idratazione; **quesito: beve a sufficienza?**

MANAGEMENT

- Prevent excess weight gain by providing anticipatory guidance around energy balance prior to commencement of steroid therapy (Grade D)
- Advise reduction in energy intake with caution.
- Follow up with regular measurement of fat free mass to monitor progress where available (Grade D)
- Dietary texture modifications may be required to accommodate eating difficulties (Grade D)
- Support patients and families in decisions regarding enteral feeding and/or gastrostomy tube placement (Grade D)
- A MULTIDISCIPLINARY TEAM should be involved in the management of feeding difficulties (Grade D)



A new chart for weight control in Duchenne muscular dystrophy

R D GRIFFITHS AND R H T EDWARDS

Muscle Research Centre, Department of Medicine, University of Liverpool, Liverpool

Prevention of excess weight gain is preferable to severe restriction in the already obese.

The modern manifestation of this advice would be provide anticipatory guidance to boys commencing corticosteroids, and to follow up with regular monitoring of growth, in particular with respect to weight gain.

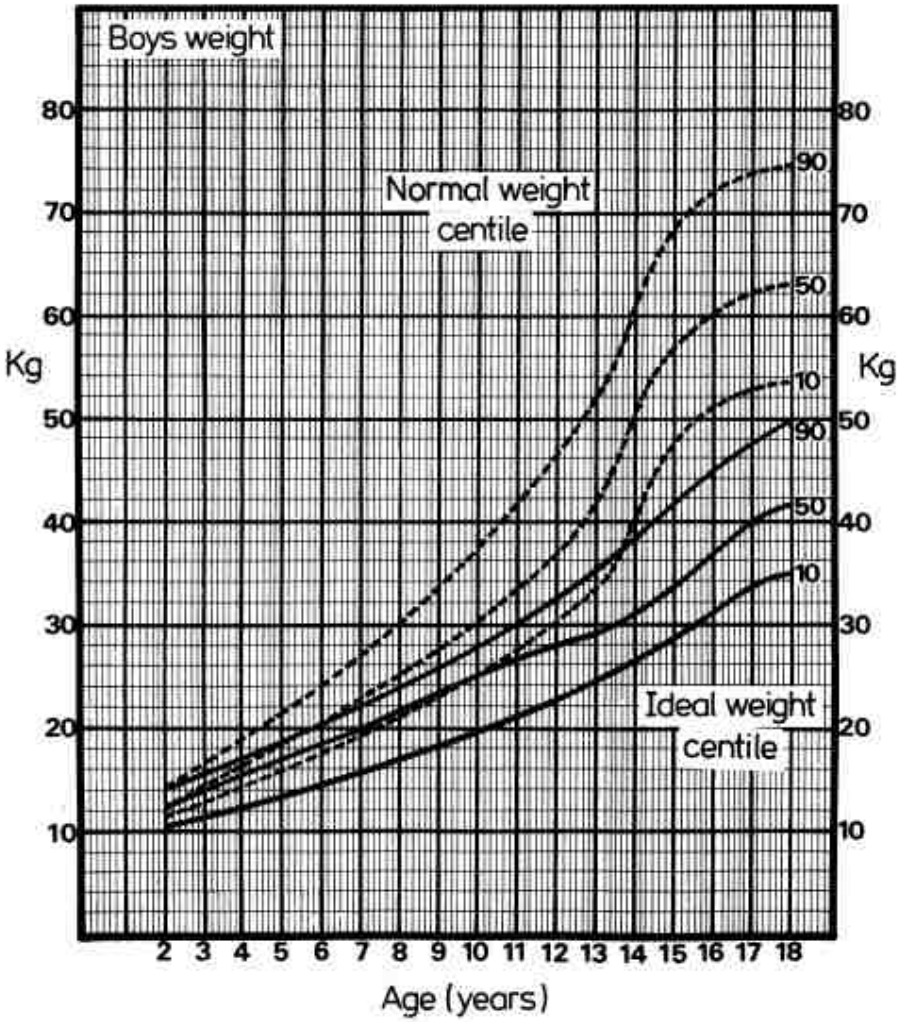


Figure Ideal weight centile chart for boys with Duchenne muscular dystrophy. Based on data from Edwards et al,[3] Edmonds et al,[4] and Tanner and Whitehouse.[5] Assumes a 4% per year decline in muscle bulk.

APPORTI

INTERVENTI

- **Valutazione fabbisogni <u>personalizzata</u>**:
  - Apporto calorico basale + aggiunte per la situazione clinica (tono muscolare, grado di attività, patologie concomitanti) + definizione di incremento pondero-staturale atteso (formule di Krick e di Culley)
- **Indice di adeguatezza alimentare**:
  (intake alimentare die / LARN) x 100
- **LARN per età sovrastimano** fabbisogni di bambini con deficit di sviluppo
- Fondamentale il **monitoraggio degli effetti** di ogni tipo di intervento

APPORTI (DMD)

- Calcolo dei fabbisogni basali seguito da correzione per dispendio fisico e per perdita muscolare:
  - TEE = BEE (HB) x LAF x MWF (muscle wasting factor)
- Se non deambulanti o con ventilazione:
  - TEE = 0.54[66.47 + 13,75 x peso in kg + 5 x altezza cm - 6.76 x età] HB
  - 0.54 valore medio dei limiti maggiore e minore del MWF (0.47<= MWF <= 0.52)
  - per i pazienti con ventilatore: moltiplicato per 1.1 LAF.
  - TEE = Total Energy Exp; BEE = Basal En Exp; LAF = Livello Att Fisica

GENERAL NUTRITION GUIDELINES SMA

(K. SWOBODA 2007)

- **Calorie: 8-11kcal x altezza in cm**
  l'apporto calorico dovrebbe essere monitorato e regolato per mantenere il peso per altezza intorno al 10 th percentile perché i soggetti SMA hanno ridotto in modo significativo la massa magra

- **Proteine: 1-2g/kg di peso/die (ma: 2g/kgPC/die solo x periodi limitati!)**

- **Grassi: non eccedere il 30%/die max**

GENERAL NUTRITION GUIDELINES SMA

(K. SWOBODA 2007)

- **Fibre**: buona regola generale è l'**età più 5** (3 anni avrebbe bisogno di 3 + 5 = 8 g fibra/die)
- **Fluidi**: **115 – 135 ml/kg**; in caso di febbre o eccessiva sudorazione le quantità possono essere aumentate
- **Micronutrienti**: evitare apporti superiori ai limiti tollerabili, alcuni micronutrienti possono avere effetti avversi: es: Piridossina (B6) neurotossica, vit A neurolesiva

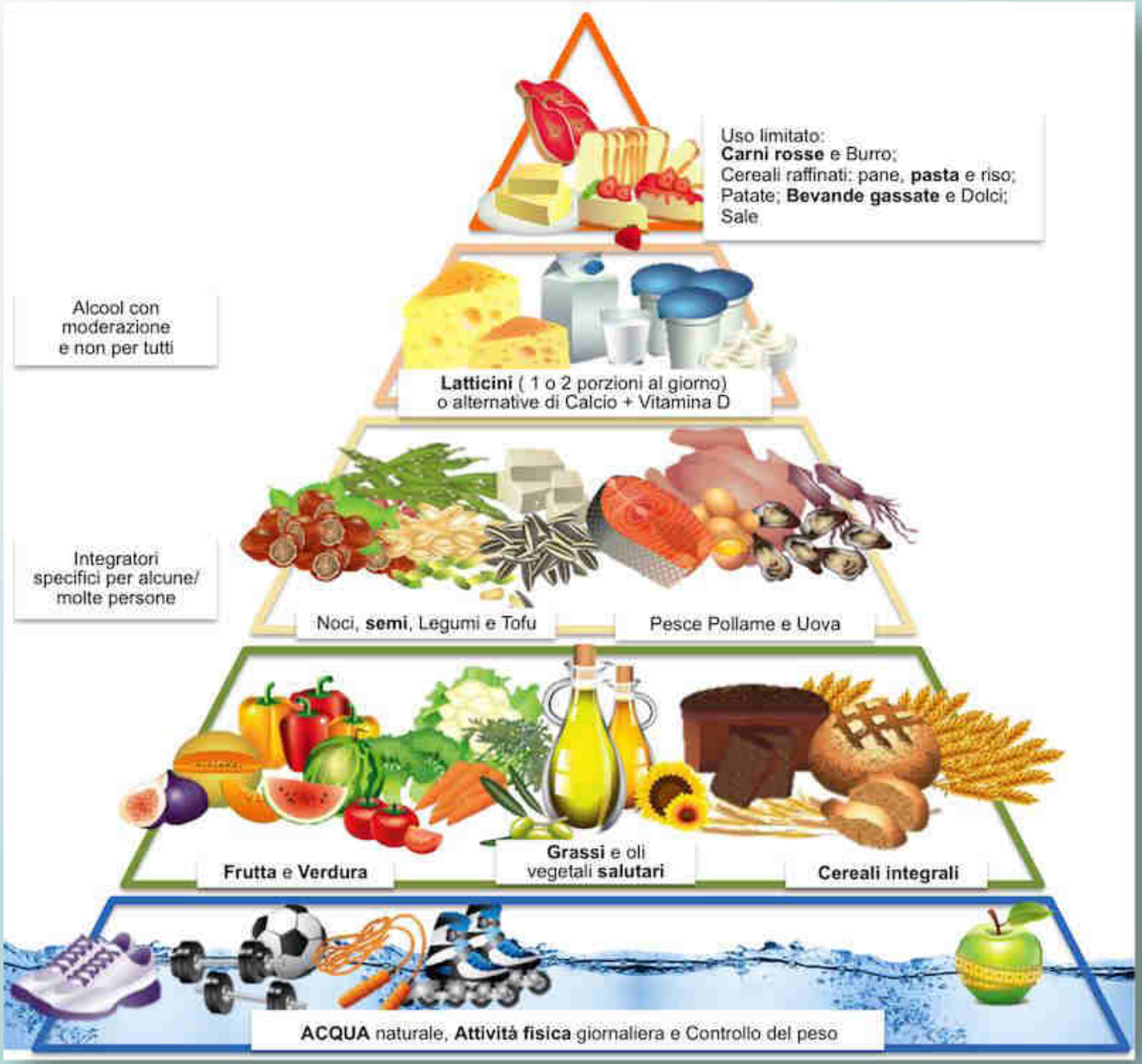
QUINDI INTERVENTO NUTRIZIONALE

- **BASALE**: prevenzione e/o cura di malnutrizione (x eccesso = obesità; x difetto = sottopeso, scarsa crescita)
- **IN ITINERE**: interventi nutrizionali delle complicanze: disfagia, decubiti, NA
- **INOLTRE**: gestione nutrizionale di patologie concomitanti: Cushing iatrogeno, ridotta tolleranza glucidica, ipertensione arteriosa, MRGE ed ulcere peptiche, stipsi

Tabella: Sequele metabolico-nutrizionali delle MNINM

	SLA	Distrofia Muscolare	Locked-in syndrome	Sclerosi Multipla	Atrofia muscolare spinale
Disfagia	++	+	++	+	+
Malnutrizione	++	+	++	+	+
Ipermetabolismo	++	/	++	/	/
Stipsi	+	+/-	+	+	+
Ipertrigliceridemia	+	+	+	+	+
Iperinsulinismo	+	+	+	+	+
Insulinoresistenza	+	+	+	+	+
Sindrome metabolica	-	+	+	+	+
Aumento massa grassa	-	+	+	+	+
Deficit vitaminici	+	+	+	+	+

MODELLO MEDITERRANEO



FATTORI NUTRIZIONALI

- Coenzima Q10
- Creatina
- Taurina
- Glutamina
- Arginina
- Leucina
- Epigallocatechina
- Curcumina
- Antiossidanti dai vegetali

RAZIONALE

- Migliorare la performance del muscolo
- Ridurre l'infiammazione

(ritenuta il fattore di danno più importante; i miociti distrofici sono più sensibili alle specie reattive dell'ossigeno – ROS – rispetto al muscolo sano; i radicali liberi – FR – inoltre danneggiano DNA, nucleotidi, proteine, lipidi, carboidrati e le strutture di membrana cellulare)

NEURODEGENERAZIONE

Neurodegenerazione → stress ossidativo → radicali liberi → neurodegenerazione

ANTIOSSIDANTI

- **Endogeni : (SOD, glutatione perossidasi..)**

- **Esogeni: assunti con la dieta (vitamine A, C ed E, selenio, licopene, polifenoli…)**

ANTIOSSIDANTI

ALIMENTI	POLIFENOLI g/100g	ORAC /100g
Cannella	9,7	131420
Chiodi di garofano	16,047	314446
Zenzero	Secco 0,47 Fresco 0,20	
Noce moscata	1,905	69640
Cumino		50372
Pepe nero		34054
Broccoli	0,19	1510
Uva nera	1,84	1746

Int J Mol Sci. Oct 2014; 15(10): 19183–19202

MICRONUTRIENTI

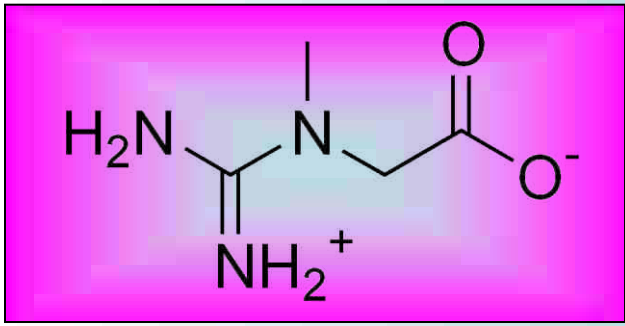
- **The use of steroid therapy combined with decreased mobility suggests that boys with DMD are at an increased risk of fractures and poor bone health (Soderpalm et al.,2007)**
- **Biggar et al. (2006) recommends daily calcium (750 mg) and vitamin D (1000 IU) supplementation in conjunction with corticosteroid therapy.**

Vitamin D3 (cholecalciferol) dosing

Age	RDA Dose, IU	Starting Dose, IU
0–6 mo	200	400
7–12 mo	260	500
1–8 y	1000	2000
9–18 y	1300	2500

Phys Med Rehabil Clin N Am. 2015 26, I1, 21–28

CREATINA

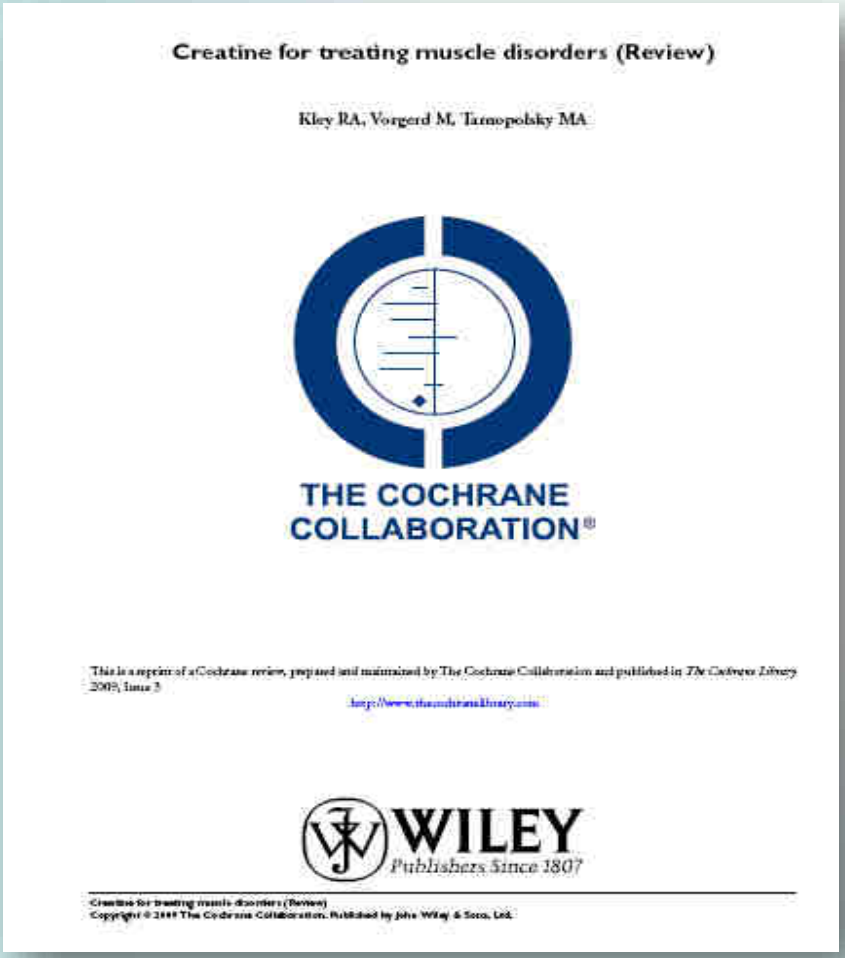


Composto intermedio del metabolismo energetico sintetizzato dal fegato (1 g/die) a partire da arginina, S-adenosil-metionina e glicina.

Utilizzato nei muscoli dei mammiferi per rigenerare ATP durante i primi secondi della contrazione muscolare.

Immagazzinamento max 0,3 g/Kg peso.

CREATINA



Creatine for treating muscle disorders (Review)

Kley RA, Vorgerd M, Tarnopolsky MA

THE COCHRANE COLLABORATION®

This is a reprint of a Cochrane review, prepared and maintained by The Cochrane Collaboration and published in *The Cochrane Library* 2009, Issue 3

http://www.thecochranelibrary.com

WILEY
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Creatine for treating muscle disorders (Review)
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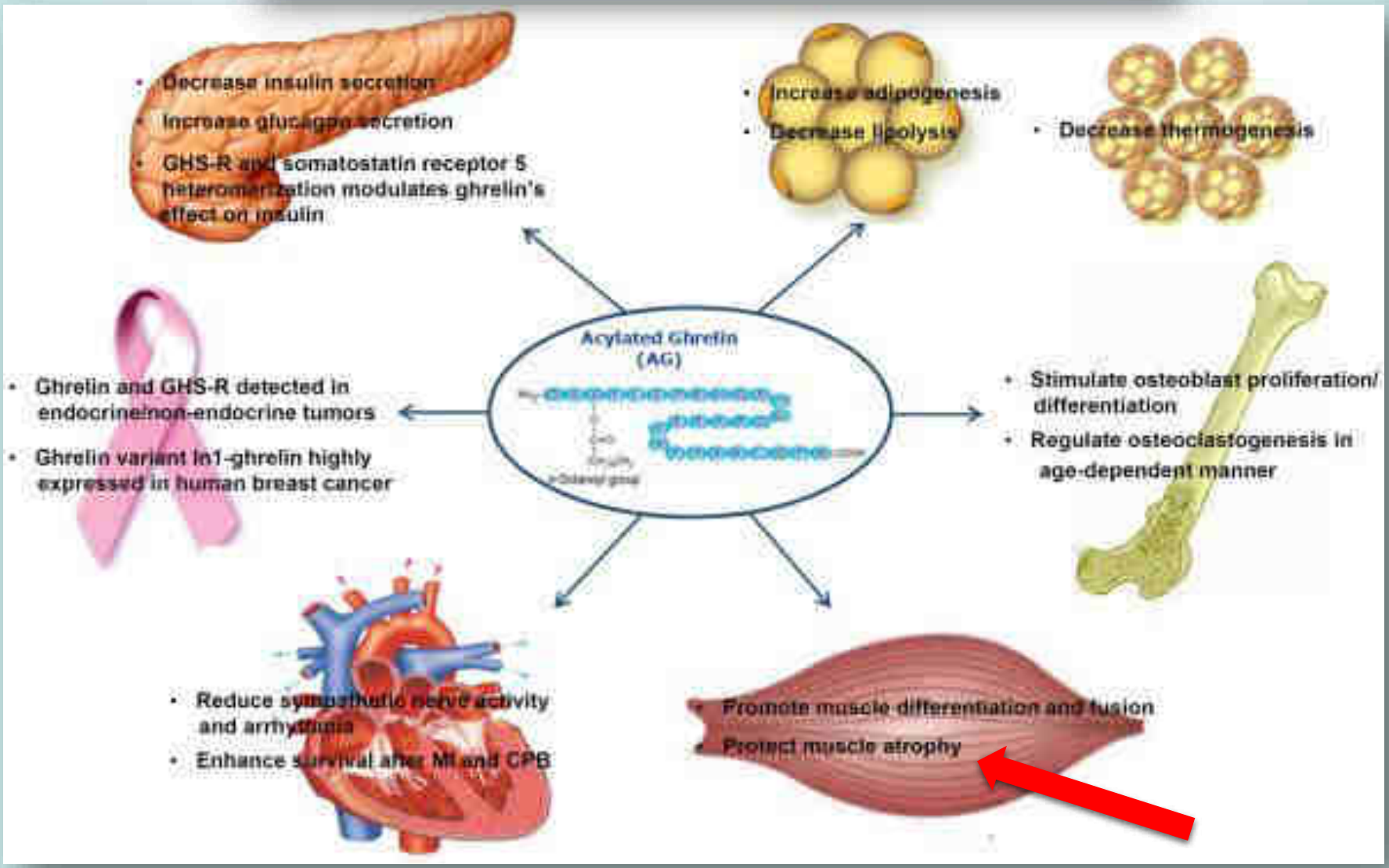
AUTHORS' CONCLUSIONS

Evidence from randomised controlled trials shows that short- and medium-term creatine treatment improves muscle strength in people with muscular dystrophies, and is well-tolerated.

12 studi randomizzati – 266 pts

Published in final edited form as:
Curr Opin Clin Nutr Metab Care. 2013 November ; 16(6): 619–624. doi:10.1097/MCO.0b013e328365b9be.

Ghrelin: much more than a hunger hormone



LA PEG

- **Rifiutata** o **posticipata** dai genitori nella maggioranza dei casi (vissuto di fallimento personale, di non naturalezza, di “punto di non ritorno”)
- Consente **rapide e buone riprese** di peso, stato nutrizionale ed aumento di statura (x ↑ apporti e ↓ consumi da stato infiammatorio da aspirazioni/ab-ingestis)
- **Non esclude apporti x os** quando consentiti

Efficacy and tolerance of gastrostomy feeding in Duchenne muscular dystrophy

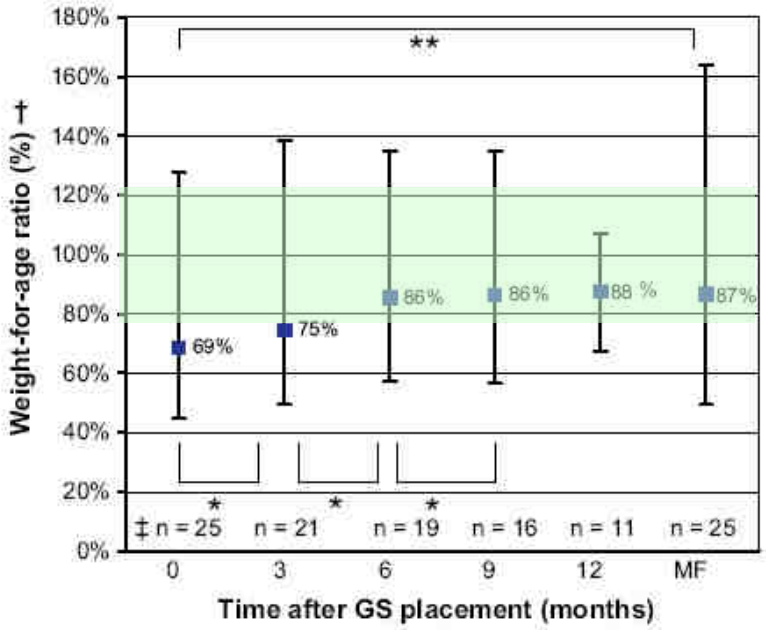


Fig. 1. Weight-for-age ratio evolution after gastrostomy placement. GS, gastrostomy; MF, maximal follow-up. *$P < 0.05$ (Wilcoxon test). **$P < 0.05$ (Friedman test). † Normal ranges for W/A ratio is 85–120%. ‡ Note that number of patients decreased at follow-up.

Clinical Nutrition

Gastrostomy is well tolerated by, and effective for improving the nutritional status of individuals with DMD.

Complicanze possibili

INTOLLERANZA A NE

- Disagio legato all'alimentazione per PEG
- Escludere **transito rallentato**, **reflusso g-e**, **stipsi** o **alvo alterno** (± diarrea)
- Valutare **modifica di schema di somministrazione** (velocità, bolo vs continuo, tipo miscela, densità o diluizione)
- Se disturbi persistono inserire **terapia farmacologica** ed eventualmente usare una **miscela idrolisata**
- Valutare **cibi frullati/omogeneizzati** x PEG
- Utilizzare **enzimi digestivi**

Journal of Pediatric Gastroenterology and Nutrition
43:123–135 © July 2006 Lippincott Williams & Wilkins, Philadelphia

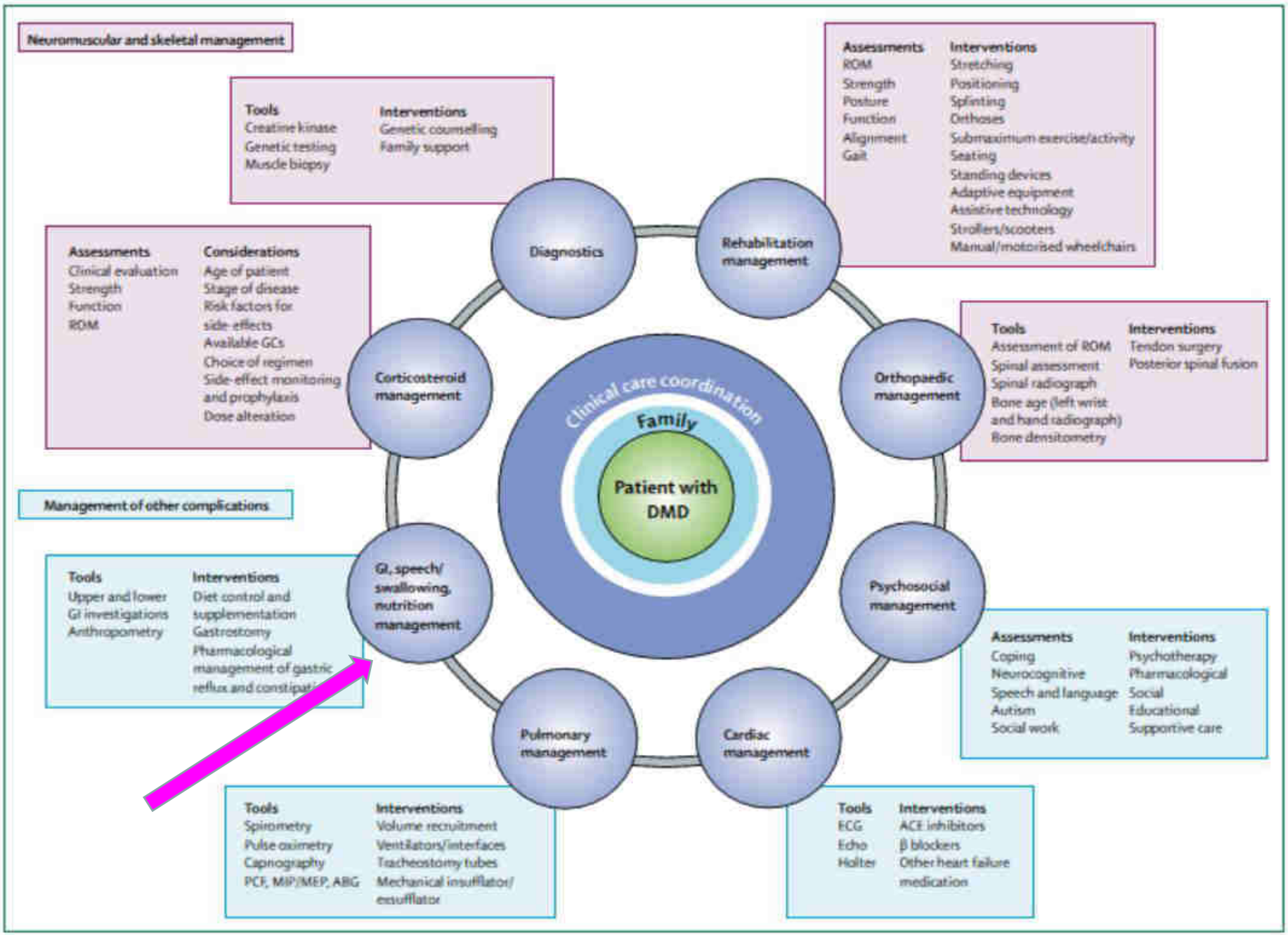
Medical Position Paper

Nutrition Support for Neurologically Impaired Children: A Clinical Report of the North American Society for Pediatric Gastroenterology, Hepatology, and Nutrition

*Valerie Marchand, †Kathleen J. Motil, and the NASPGHAN Committee on Nutrition

CONCLUSION

A multidisciplinary team of pediatric specialists, including physicians, nurses, dietitians, occupational and speech therapists, psychologists, and social workers, has the responsibility to monitor the nutritional status of neurologically impaired children and provide early, efficient nutritional intervention to ensure normal growth, optimal functional status, and quality of life. The decision to initiate enteral nutrition may be difficult for the family who perceive this approach as a failure on their part to feed their child. Involvement of the child and the family in the decision-making process is important to ensure successful nutritional intervention. Weight gain, ease of feeding the child, and the stress involved with meals usually improve with nutritional intervention. The multidisciplinary team is obligated to provide ongoing support and education to the family throughout the long-term relationship.



www.thelancet.com/neurology Published online November 30, 2009 DOI:10.1016/S1474-4422(09)70271-6

GRAZIE PER L'ATTENZIONE