

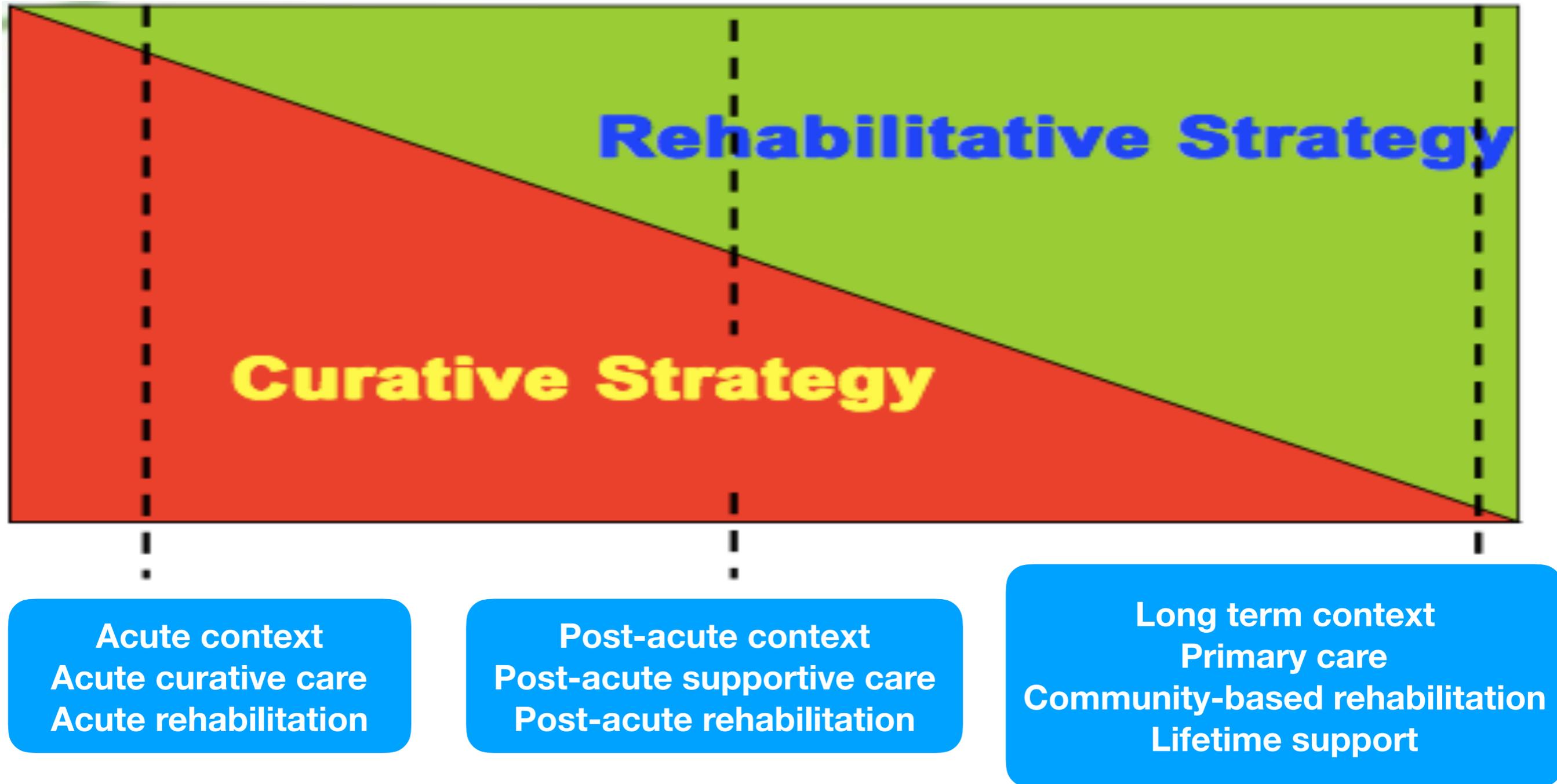
Il progetto riabilitativo nelle persone affette da Malattia di Pompe

Giovanni Iolascon

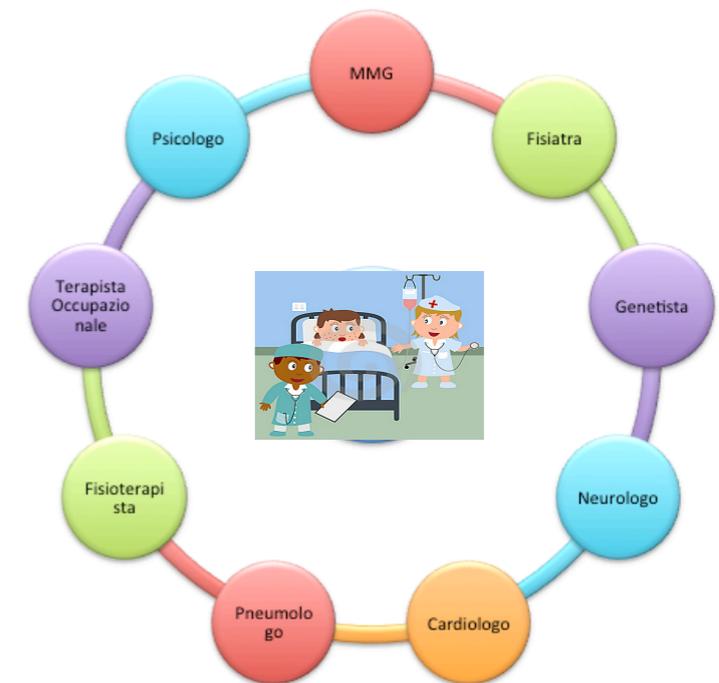
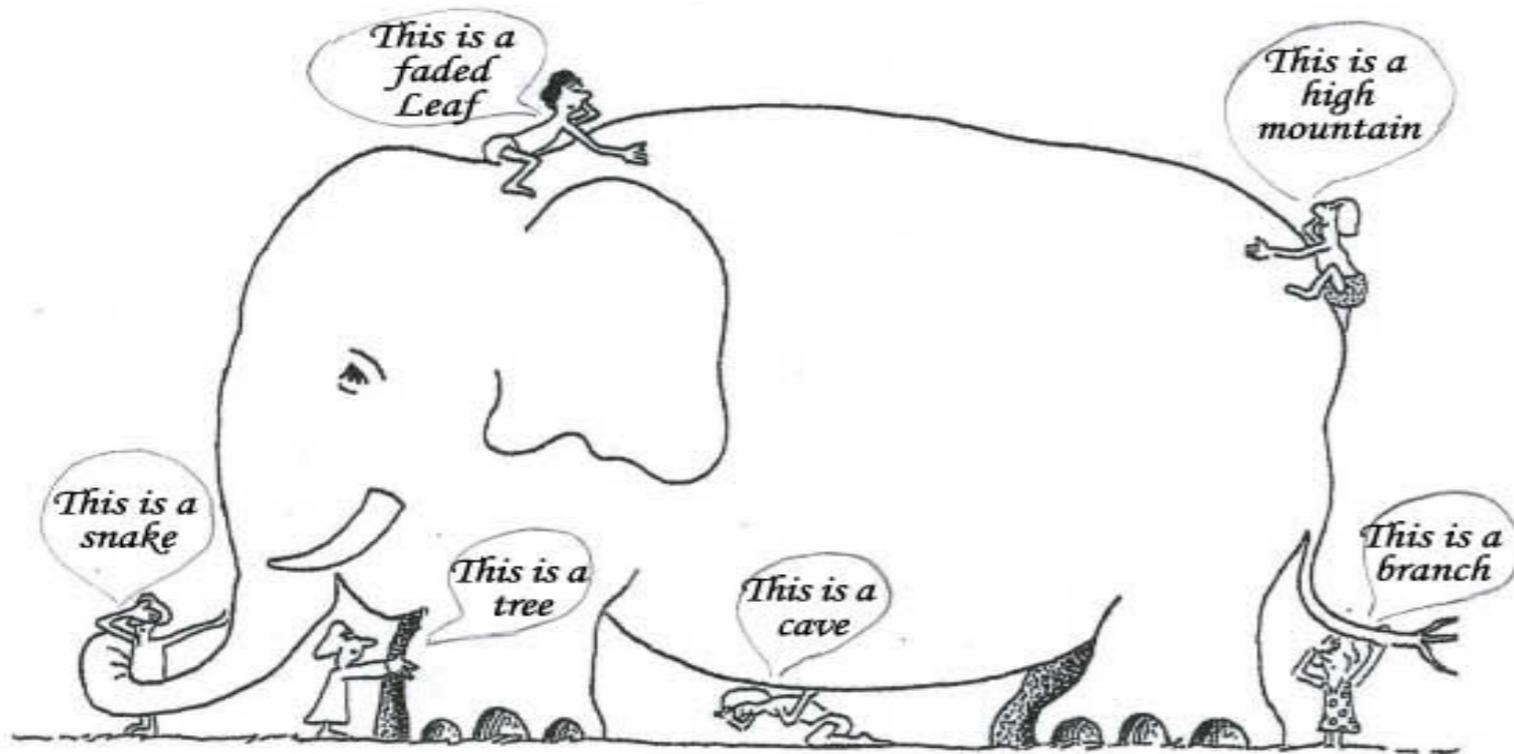
criticità

- le miopatie metaboliche si manifestano in maniera molto variegata
- variabilità fenotipica anche con lo stesso corredo genetico
- le basi razionali dell'approccio riabilitativo e dei relativi outcome non sono ben definiti
- non chiaro il giusto rapporto rischio / beneficio della maggior parte delle metodiche riabilitative
- definizione di un'attività fisica adattata al paziente affetto da LOPD

Health and Rehabilitation Services Continuum



Approccio multidisciplinare al paziente affetto da miopatie metaboliche



la complessità del paziente affetto da miopatia metabolica

- 1. Complessità sanitaria:** il paziente presenta una patologia fisica o mentale grave o è affetto da polipatologia
- 2. Complessità assistenziale:** il paziente non è autonomo nelle varie attività (mobilità, cura della persona, vita domestica, sociale...) e richiede aiuto da parte di altri, anche con l'uso di dispositivi fisici
- 3. Complessità ambientale:** il paziente presenta criticità legate alla abitazione, al reddito, al nucleo familiare e alle persone che forniscono aiuto, alle relazioni familiari, non ha ancora accesso a servizi, presidi, ausili e facilitazioni economiche

Paziente complesso



Piano d'indirizzo per la Riabilitazione

Definizione del grado di “necessità” della persona da riabilitare

1. **COMPLESSITÀ CLINICA:** assessment e stratificazione dell'alto rischio clinico. La complessità clinica si correla all'insieme della complessità diagnostica, assistenziale, organizzativa e dei differenti interventi terapeutici proporzionalmente graduati per complessità e per consumo di risorse.
2. **DISABILITÀ:** perdita delle capacità funzionali nell'ambito delle attività fisiche, motorie, cognitive, comportamentali che nella più attuale concezione bio-psico-sociale impattano con i fattori ambientali riducendo il livello di partecipazione dell'individuo allo svolgimento delle attività della vita quotidiana e di relazione. La disabilità viene usualmente misurata con scale di disabilità sia di tipo bio-psicometrico sia funzionali che indagano la possibilità di eseguire le diverse attività e che consentono il monitoraggio dell'evoluzione del quadro funzionale nel tempo.
3. **MULTIMORBIDITÀ:** insieme di patologie e condizioni classificate secondo scale a punteggi crescenti. Tali comorbidità possono rappresentare un mero elenco per una stratificazione prognostica più accurata o attivi cofattori che influenzano la clinica, il trattamento e la prognosi.



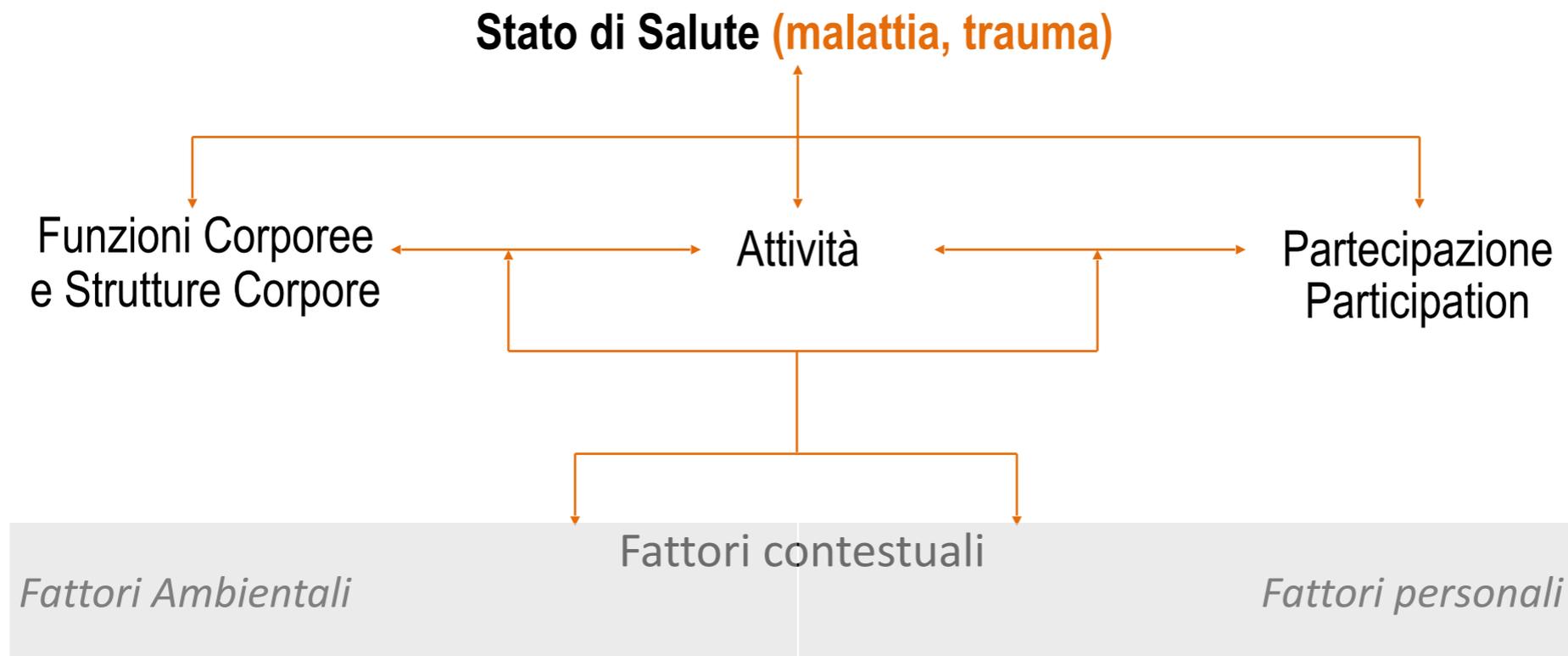
Ministero della Salute

Il “percorso assistenziale integrato” è il riferimento complessivo che rende sinergiche le componenti sanitarie e non sanitarie dell'intervento riabilitativo. In tale ambito **il Progetto Riabilitativo Individuale (PRI)** rappresenta lo strumento specifico, sintetico ed organico per tutto ciò, unico per ciascuna persona, definito dal medico specialista in riabilitazione¹ in condivisione con gli altri professionisti coinvolti. Elementi essenziali sono sempre rappresentati dalla piena informazione e dalla partecipazione consapevole ed attiva alle scelte ed agli interventi da parte della persona che ne è al centro, della famiglia e del suo contesto di vita.

Gli interventi derivanti dal progetto riabilitativo, incentrati sui diversi problemi rilevati, necessitano di una valutazione sistematica della *performance* e della definizione di obiettivi ed indicatori di processo, al fine della verifica del raggiungimento del risultato atteso.

Il PRI, applicando i parametri di menomazione, attività e partecipazione sociale elencati nella ***International Classification of Function (ICF)***, definisce la prognosi, le aspettative e le priorità del paziente e dei suoi familiari; viene condiviso con il paziente, quando possibile, con la famiglia ed i *caregiver*, definisce le caratteristiche di congruità ed appropriatezza dei diversi interventi, nonché la conclusione della presa in cura sanitaria in relazione agli esiti raggiunti.

ICF

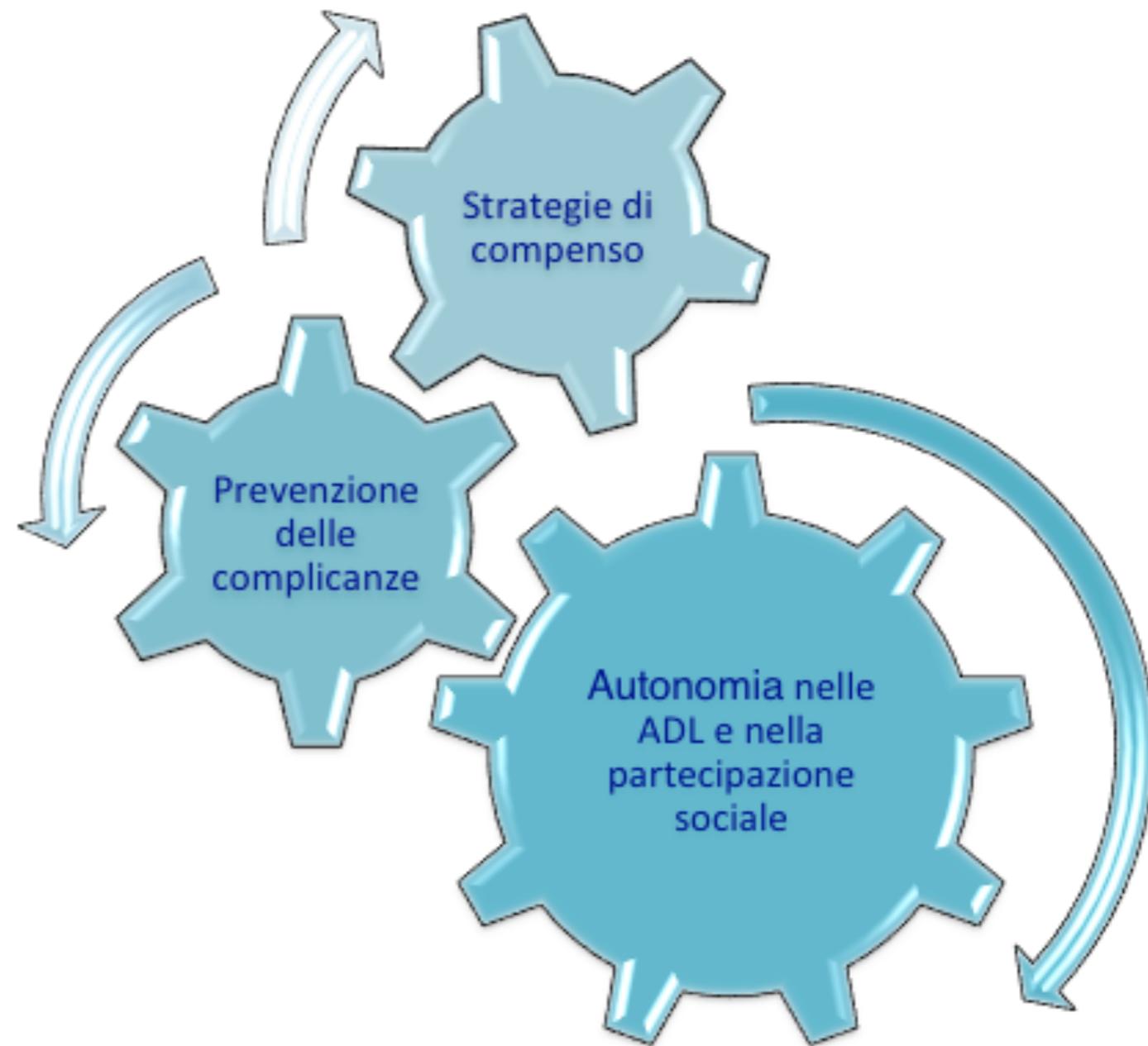
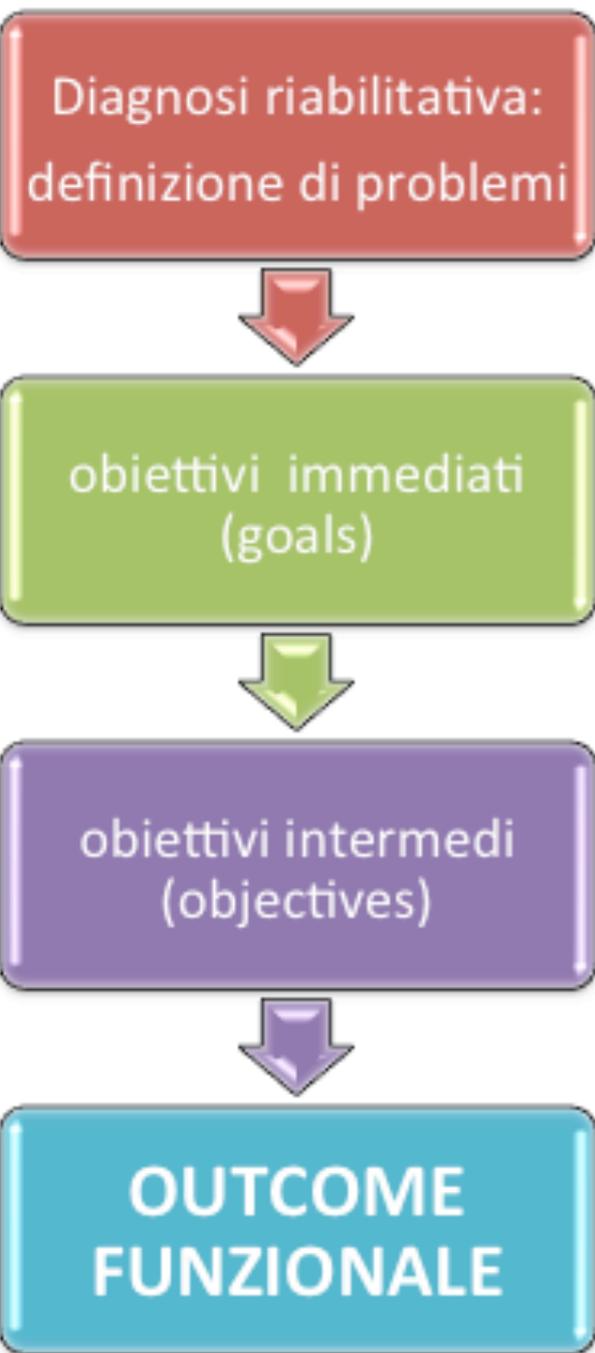


I Fattori Ambientali comprendono i fattori dell'ambiente fisico sociale ed attitudinale in cui vivono le persone. Sono i fattori esterni all'individuo che possono influenzare positivamente o negativamente la performance dell'individuo come membro della società, la sua capacità di eseguire azioni o compiti, o le sue funzioni e strutture.

I Fattori Personali comprendono particolari aspetti della vita del paziente, incluse caratteristiche dell'individuo che non fanno parte della condizione di salute o dello stato di salute. Sono inclusi il sesso, la razza, l'età, altre condizioni di salute, lo stile di vita, il background sociale, l'istruzione, la professione, le esperienze passate e presenti.

The ICF has proven to be suitable and feasible to be implemented at the level of clinical and rehabilitation practice, at the level of service provision and payment, as well as on the level of policy and program planning

Approccio riabilitativo problem-related



le linee guida internazionali sull'approccio riabilitativo del paziente con malattia di Pompe

- American College of Medical Genetics : Pompe disease diagnosis and management guideline
- American Association of Neuromuscular & Electrodiagnostic Medicine (AANEM) : CONSENSUS TREATMENT RECOMMENDATIONS FOR LATE- ONSET POMPE DISEASE
- Spanish Society of Internal Medicine (SEMI), Spanish Society of Neurology (SEN) and Spanish Society of Pneumology and Thoracic Surgery (SEPAR).
- Brazilian Network for Studies of PD. Guidelines for the diagnosis, treatment and clinical monitoring of patients with juvenile and adult Pompe Disease
- Pompe Disease: Diagnosis and Management Evidence-Based Guidelines from a Canadian Expert Panel
- Practical Recommendations for Diagnosis and Management of Respiratory Muscle Weakness in Late-Onset Pompe Disease

American College of Medical Genetics

Pompe disease diagnosis and management guideline

Motor and functional assessments are recommended to establish a baseline with repeat testing at 3 – 6 month intervals for children under age five years, and annually in older children and adults, except where additional testing is clinically indicated by change in function or failure to make expected progress. The following list includes most of the major assessments but is not intended to be an exhaustive listing of all possible assessments or tools used in assessments.

Musculoskeletal

- passive ranges of motion and measures of muscle extensibility (goniometry)
- posture and alignment in all positions
- **Spine:**
 - flexibility, stability, posture and alignment
 - monitoring for kyphosis, scoliosis, lordosis
- hip joint stability/integrity
- DEXA
- radiographs as needed for monitoring hip and spine status

American College of Medical Genetics Pompe disease diagnosis and management guideline

Routine evaluation in Pompe:

Multidisciplinary evaluation should include assessment within each category of the World Health Organization (WHO) International Classification of Functioning, Disability, and Health (**ICF**).

MUSCULOSKELETAL/FUNCTIONAL/REHABILITATION RECOMMENDATIONS

- Monitor cardiorespiratory status and response to position and activity with pulse oximetry during evaluation and treatment initially and with changes in status or activity.
- Screen for osteopenia/osteoporosis with DEXA and follow-up as needed.
- Assess musculoskeletal impairments, functional deficits, levels of disability, and societal participation at regular intervals and as needed, including radiographs as needed for monitoring of scoliosis, hip stability, and long bone integrity.
- **Enhance Muscle Function:**
 - **increase biomechanical advantage for movement:**
 - **provide practice, movement, and gentle strengthening within limits of physiological stability.**
 - **provide rests as needed to avoid overexertion.**
 - **Follow Guidelines For Strengthening From Other Progressive Muscle Diseases:**
 - **submaximal, functional, and aerobic exercise recommended.**
 - **avoid excessive resistive and eccentric exercise.**
 - **avoid overwork weakness.**
 - **avoid disuse atrophy.**
 - allow compensatory movements necessary for function, but prevent negative results (contracture and deformity).
- **Prevent/Minimize/Correct 2° Musculoskeletal Impairment (Contracture/Deformity):**
 - **stretching/positioning.**
 - **orthotic intervention and splinting.**
 - **seating systems/standers.**
- Optimize function with adaptation and assistive technology as needed.
- Educate the patient and family about the natural history and recommendations for intervention.

**American Association of Neuromuscular & Electrodiagnostic Medicine (AANEM)
CONSENSUS TREATMENT RECOMMENDATIONS FOR LATE- ONSET POMPE DISEASE
Muscle Nerve. 2012**

Strengthening and Therapeutic Exercise—There are no established guidelines for muscle strengthening or therapeutic exercise for individuals with late-onset Pompe disease. Studies of the effects and treatment roles of exercise and nutrition in late-onset Pompe disease have been few and have had small sample sizes.^{34,39,40} Although more information is needed, these studies suggest that sub-maximal aerobic exercise may increase muscle strength and function through improved clearance of accumulated glycogen in the muscle cytosol.⁴¹ In contrast, there is insufficient evidence that resistance training improves strength in late-onset Pompe disease. Traditionally, excessively strenuous resistance exercises have been discouraged in muscle disorders because of the potential for exacerbating muscle degeneration,^{42–44} and in Pompe disease there is additional theoretical concern that excessive muscle contraction might lead to increased leakage of glycogen from lysosomes or cause lysosomal rupture, thereby hastening muscle damage.⁴⁵ The favorable pattern of response in the strength of proximal and respiratory muscles in ERT studies of late-onset Pompe disease is encouraging, but further studies are needed.^{18,36} Therefore, at this time the authors recommend that the general precautions regarding strengthening exercises that are followed for other degenerative muscle diseases should be applied to late-onset Pompe disease^{46,47} (Table 1). Therapeutic exercise in late-onset Pompe disease may pose a risk for cardiopulmonary compromise in this population. Consequently, a pulmonologist should evaluate the patient before initiation of an exercise program.²⁴

American Association of Neuromuscular & Electrodiagnostic Medicine (AANEM) CONSENSUS TREATMENT RECOMMENDATIONS FOR LATE- ONSET POMPE DISEASE Muscle Nerve. 2012

Therapeutic exercise should start slowly, allowing for periods of rest, followed by a gradual increase in exercise intensity from mild to moderate, reaching aerobic levels of about 60–70% of maximal effort at a frequency of 3–5 days per week.^{24,40,43} Optimally, an experienced physical therapist should develop a structured program, monitor the patient, and remain in contact with the prescribing physician. Pulse oximetry, heart rate, and perceived exertional effort (e.g., Borg scale) should be monitored initially to guide progression of the program. Moreover, the physical therapist should teach the patient how to monitor heart rate and correlate it with perceived exertional effort and oxygen saturation information to prepare the patient for a self-monitored, home-based exercise program. A preliminary report of one late-onset Pompe disease patient demonstrated that side-alternating vibration training produced sustained improvement in mobility at 1 year of follow-up.⁴⁸ Larger studies are needed before this form of training can be recommended.

American Association of Neuromuscular & Electrodiagnostic Medicine (AANEM) CONSENSUS TREATMENT RECOMMENDATIONS FOR LATE- ONSET POMPE DISEASE Muscle Nerve. 2012

Contracture and Limb Deformity—Because no established guidelines exist for management of secondary musculoskeletal impairments, including contracture and deformity, in late-onset Pompe disease, general principles established for the management of other neuromuscular disorders can be applied.⁴⁹ These general principles include limiting contracture and deformity by gentle daily stretching, correction of improper positioning, judicious and timely use of splints and orthotic interventions, and provision of adequate support in all positions, including sitting and supported standing.⁴⁹ Written instructional materials for the family are more effective than verbal instruction.⁴⁹ Prevention of contracture and deformity is critical to preserve function and limit other secondary complications, such as skin breakdown and chronic musculoskeletal pain. A preventive stretching regimen should be implemented early and be performed as part of the daily routine. Such regimens are better tolerated and accepted if they are initiated before muscle tendon tightness and contractures develop, a point at which stretching often becomes painful. Aggressive stretching should be approached cautiously because, at least in severely affected children, a tendency for pathologic fracture has been observed.⁵⁰

**American Association of Neuromuscular & Electrodiagnostic Medicine (AANEM)
Muscle Nerve. 2012
CONSENSUS TREATMENT RECOMMENDATIONS FOR LATE- ONSET POMPE
DISEASE**

Adaptive Equipment and Orthotic Interventions—A variety of orthotic interventions are available for contracture management, such as ankle–foot orthoses (AFOs) to prevent plantarflexion contractures, thigh binders to prevent iliotibial band contractures, knee splints to prevent knee flexion contractures, resting wrist/hand/finger splints to prevent wrist and finger flexor contractures over multiple joints,³⁴ and lumbar corsets for management of back pain. Nocturnal resting splints or AFOs to prevent plantarflexion contractures have not been adequately studied in Pompe disease.^{49,51} Wheeled walkers or quad canes may help with

American Association of Neuromuscular & Electrodiagnostic Medicine (AANEM) Muscle Nerve. 2012 CONSENSUS TREATMENT RECOMMENDATIONS FOR LATE- ONSET POMPE DISEASE

Physical/occupational therapy

A physical or occupational therapist should develop an exercise program that may include one or more of the following: walking, treadmill, cycling, pool-based program, swimming, submaximal aerobic exercise, or muscle strengthening, that follows the guidelines for other degenerative muscle diseases

Avoid overwork weakness, excessive fatigue, disuse, strenuous exercises, and eccentric contractions

Emphasize submaximal aerobic exercise

Incorporate functional activities when possible

Teach patient to monitor heart rate and breathing in relation to exertion

Integrate energy conservation techniques and biomechanical advantages

A preventive stretching regimen should be started early and performed as part of the daily routine to prevent or slow the development of muscle contractures and deformities

Management of contractures

Manage contractures by using orthotic devices, appropriate seating position in the wheelchair, and standing supports

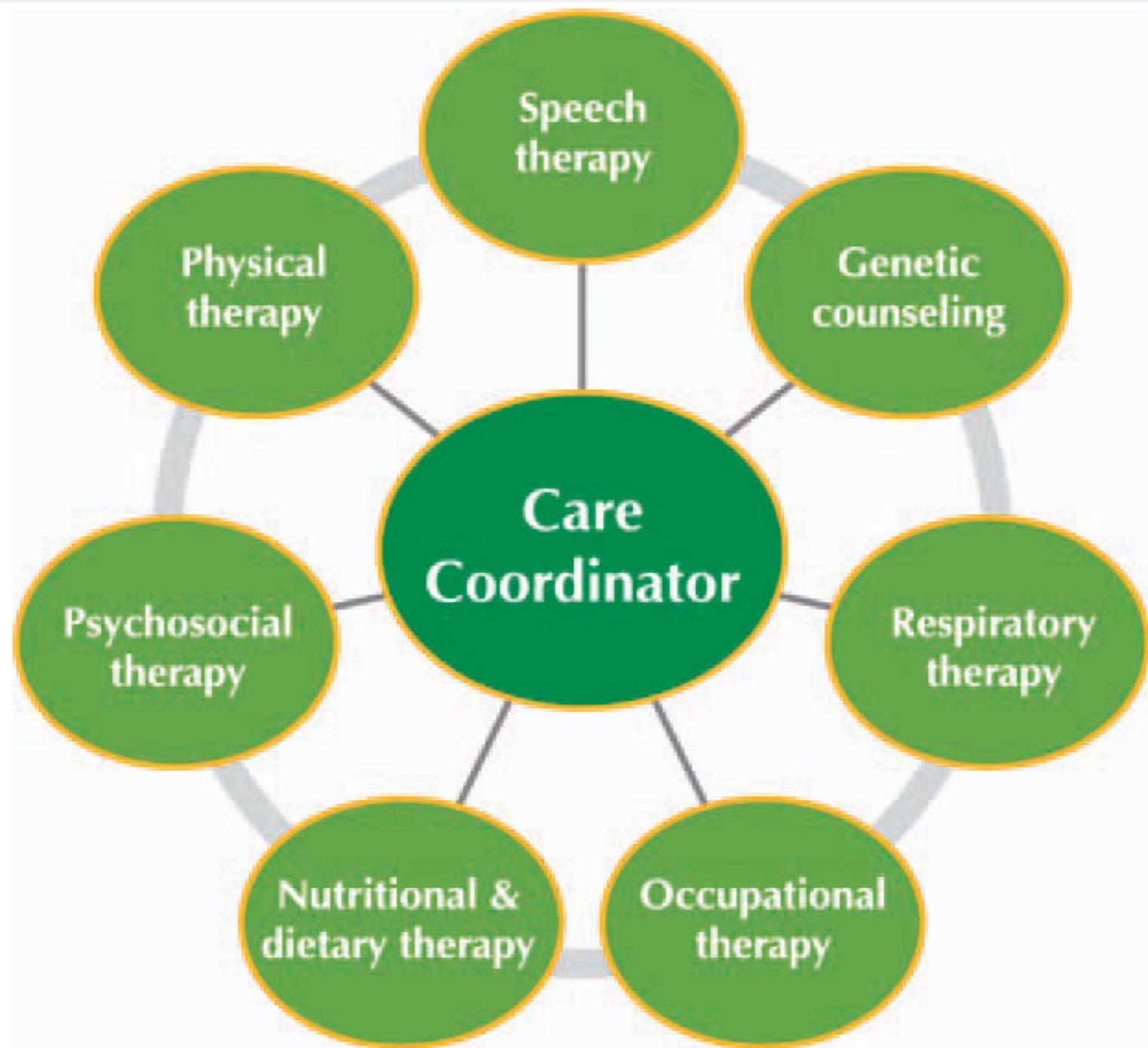
Surgical intervention

Surgical intervention should be considered for scoliosis when the Cobb angle is between 30° and 40°

**American Association of Neuromuscular & Electrodiagnostic Medicine (AANEM)
Muscle Nerve. 2012
CONSENSUS TREATMENT RECOMMENDATIONS FOR LATE- ONSET POMPE
DISEASE**

Osteopenia and Osteoporosis—Low bone mineral density (osteoporosis) is a common feature in patients with Pompe disease. A recent study demonstrated that 67% of the patients tested had a bone mineral density *z*-score of -1 and that the decrease in bone density was present in both the infantile- and late-onset forms of Pompe disease.⁶⁶ Therefore, the authors recommend that individuals with late-onset Pompe disease, including all children and adults who are wheelchair- or ventilator-dependent or who have decreasing muscle strength, be screened annually with dual-energy X-ray absorptiometry (DEXA).^{24,66} On the basis of a

American Association of Neuromuscular & Electrodiagnostic Medicine (AANEM) Muscle Nerve. 2012 CONSENSUS TREATMENT RECOMMENDATIONS FOR LATE- ONSET POMPE DISEASE



Guidelines for the diagnosis, treatment and clinical monitoring of patients with juvenile and adult Pompe disease

Diretriz para o diagnóstico, tratamento e acompanhamento clínico de pacientes com doença de Pompe juvenil e do adulto

Juan Clinton Llerena Junior¹, Osvaldo JM Nascimento², Acary Souza B Oliveira³, Mario Emilio T Dourado Junior⁴, Carlo D Marrone⁵, Heloise Helena Siqueira⁶, Cláudia FR Sobreira⁷, Elza Dias-Tosta⁸, Lineu Cesar Werneck⁹

Brazilian Network for Studies of PD. The meeting of these experts took place in October 2013, Sao Paulo, Brazil
Arq Neuropsiquiatr 2016;

Table 6. Classification of functioning, disability and health (ICF) for adult Pompe disease (2003). www.who.int/classification/icf

Code	Domain
b134	Sleep functions
b3100	Voice production
b445; s430	Respiratory muscle functions; structure of the respiratory tract
b455	Exercise tolerance functions
b510	Food intake functions
b525	Defecation functions
b530	Weight maintenance functions
b730	Muscle strength functions
d230	Performing daily routines
d450	Walking
d460	Moving through different locations
d598	Self-care
d640	Performing household chores
e120	Products and technologies to facilitate mobility and/or transport the person indoors and outdoors

Physiotherapy management in late-onset Pompe disease: Clinical practice in 88 patients

Marein M. Favejee ^{a,b,*}, Bionka M.A. Huisstede ^a, Johannes B.J. Busmann ^a, Michelle E. Kruijshaar ^{b,c}, Ans T. van der Ploeg ^{b,c}



Characteristics of Dutch adult Pompe patients (N=88).

	Total N=88	Ever PT N=52	Never PT N=36	P-value*
Gender, male (N (%))	42 (47.7)	23 (44.2)	19 (52.8)	0.430
Age in years (median (range))	53.0 (25–76)	53.0 (29–76)	51.5 (25–74)	0.842
Disease duration in yrs (median (range))	16.5 (3–53)	19.5 (6–45)	13.0 (3–53)	0.030
Mobility (N (%)):				0.02
- no walking aids	50 (56.8)	23 (44.2)	27 (75.0)	
- use of walking aids	14 (15.9)	10 (19.2)	4 (11.1)	
- partial wheelchair use	7 (8.0)	6 (11.5)	1 (2.8)	
- permanent wheelchair use	17 (19.3)	13 (25.0)	4 (11.1)	
Ventilator use (N (%)):				0.170
- no ventilator use	64 (72.7)	35 (67.3)	29 (80.6)	
- non-invasive use	18 (20.5)	12(23.1)	6 (16.7)	
- invasive use	6 (6.8)	5 (9.6)	1 (2.8)	
Physiotherapy (N (%)):				Na
- never	36 (40.9)	Na	36	
- at present	32 (36.4)	32	Na	
- stopped with treatment	20 (22.7)	20	Na	

Table 2

Reasons for referral to physiotherapy for Dutch adult Pompe patients.

	Total N (%) N=52	Mild N (%) N=30	Moderate–severe N (%) N=22
Muscle weakness	27 (51.9)	14 (46.7)	13 (59.1)
Loss of general condition	21 (40.4)	13 (43.3)	8 (36.4)
Pain	13 (25.0)	8 (26.7)	5 (22.7)
Problems with ADL	9 (17.3)	3 (10.0)	6 (27.3)
Stiffness	7 (13.5)	4 (13.3)	3 (13.6)
Other	4 (7.7)	2 (6.7)	2 (9.1)
Pulmonary problems	3 (5.8)	1 (3.3)	2 (9.1)

ADL: activities of daily living.

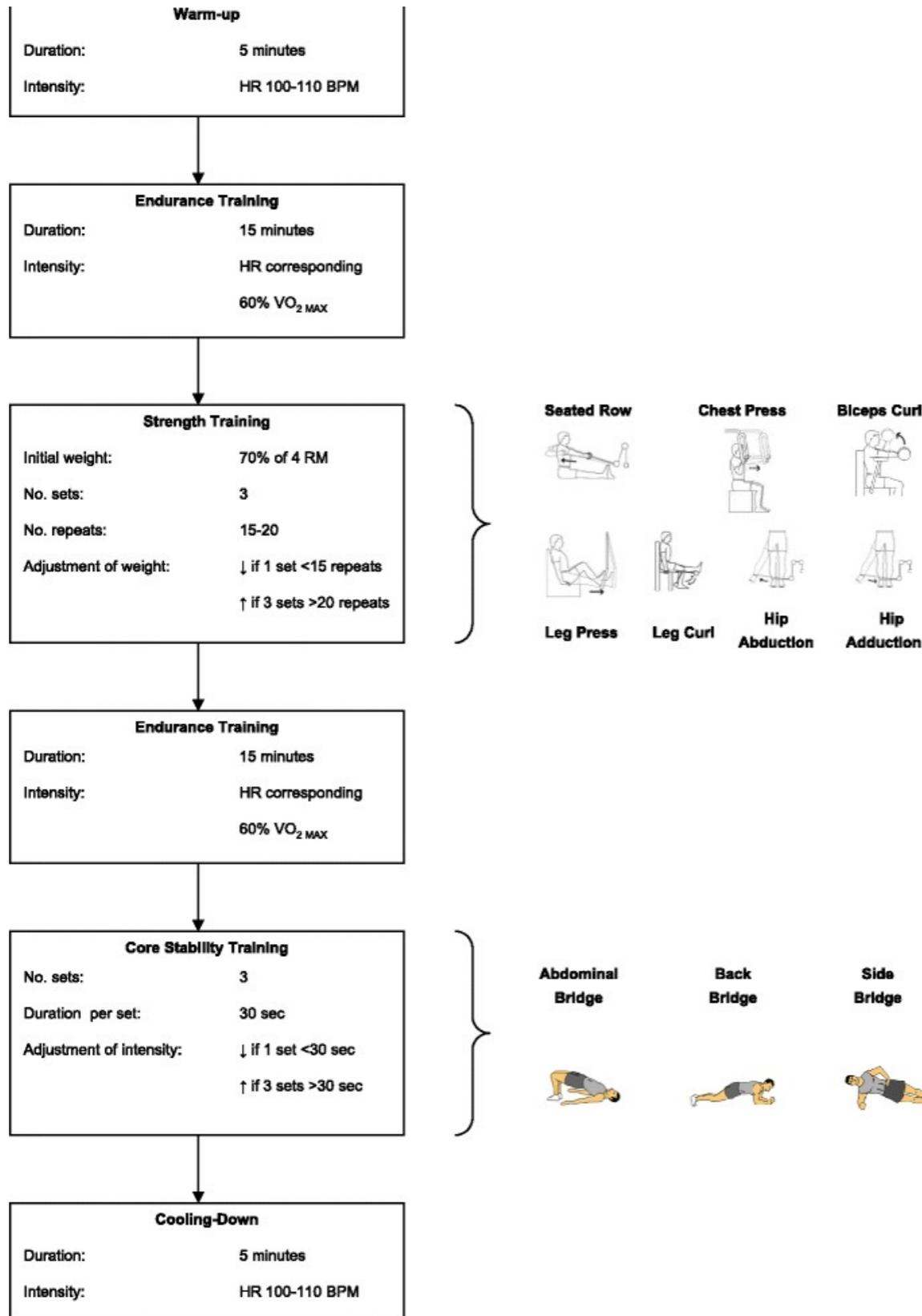
More than one reason can be stated so percentages add up to more than 100%.

Highlights

- **41% of patients with Pompe disease were never referred for physiotherapy.**
- More patients could be referred; evidence and guidelines are needed to support this.
- Treatment objective and intervention are often not in line with the referral reason.
- Tailoring intervention to referral reason and objective may improve physiotherapy care.

Safety and efficacy of exercise training in adults with Pompe disease: evaluation of endurance, muscle strength and core stability before and after a 12 week training program

Linda E. M. van den Berg¹, Marein M. Favejee^{1,2}, Stephan C. A. Wens³, Michelle E. Kruijshaar¹, Stephan F. E. Praet², Arnold J. J. Reuser⁴, Johannes B. J. Bussmann², Pieter A. van Doorn³ and Ans T. van der Ploeg^{1*}

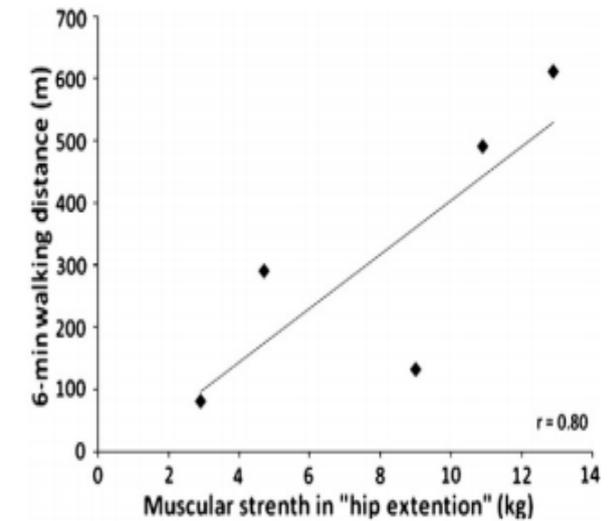
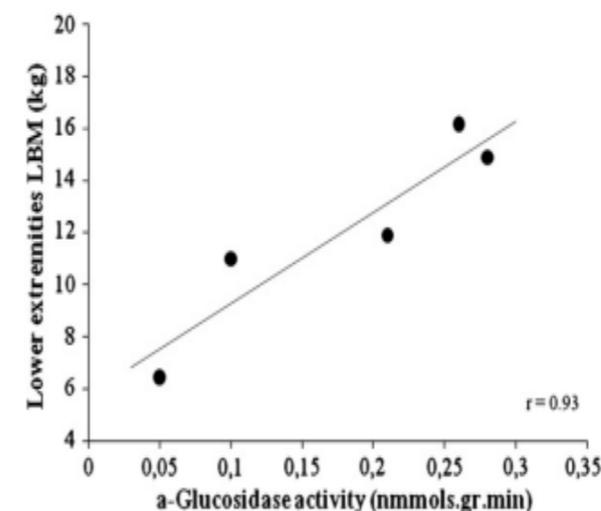
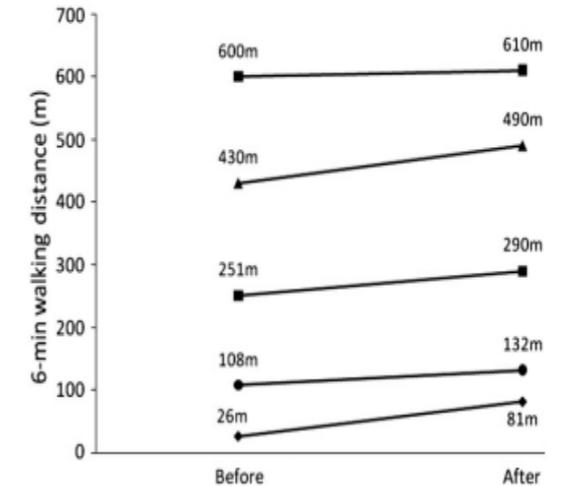
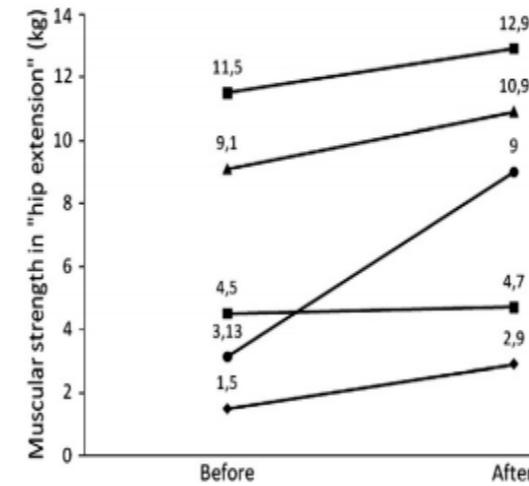


Our study shows that a combination of endurance, strength and core stability training is feasible and can be performed safely in patients with Pompe disease. Such training helps to improve endurance, muscle strength, muscle function and core stability.

This training program thus seems to offer added value for Pompe patients to those of ERT.

Management of LOPD: Exercise

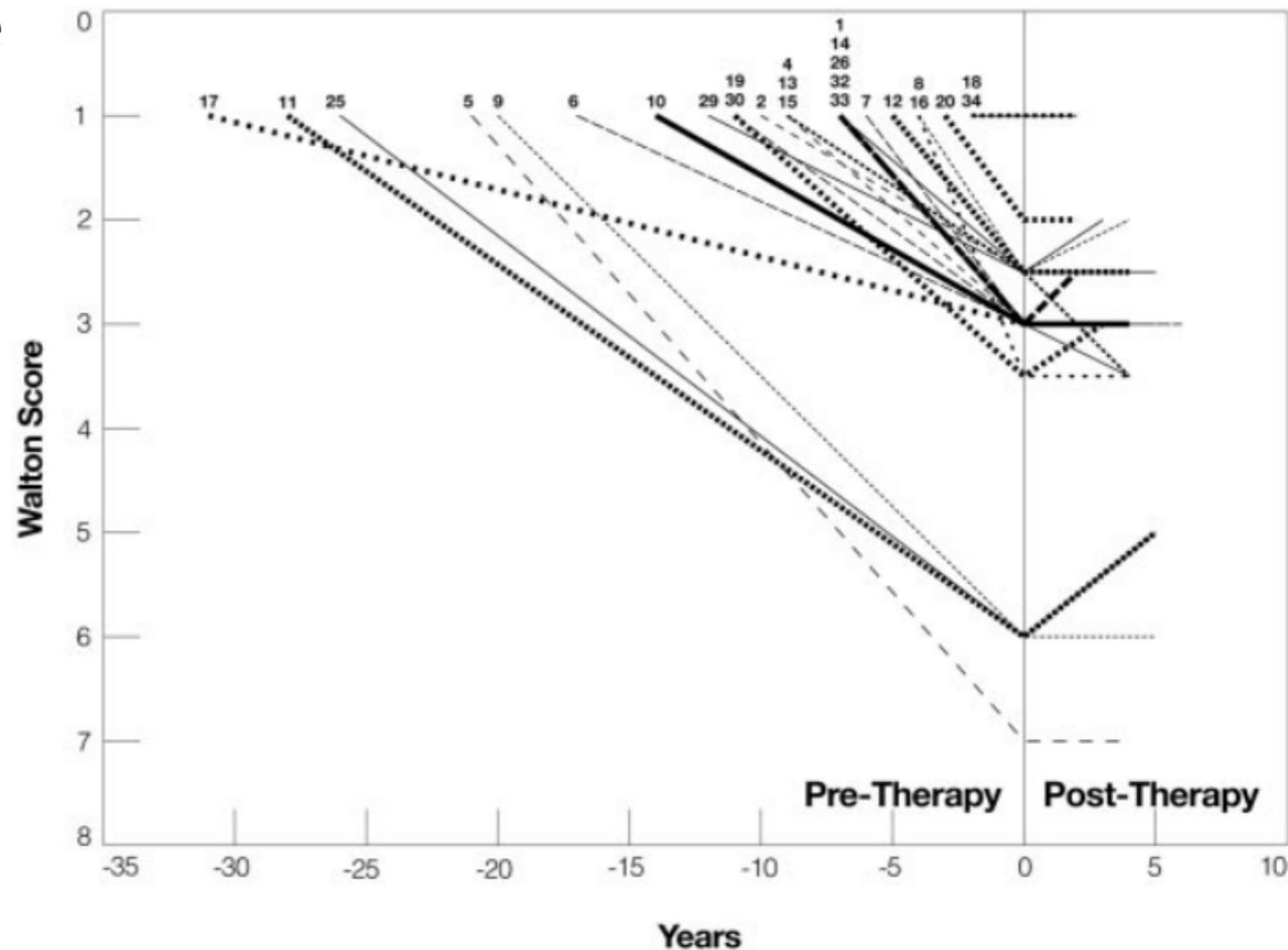
- **Disuse atrophy and muscle weakness** are universal in LOPD
- **Effect of exercise on muscle function** in patients with LOPD has limited evidences
- **Resistance exercise** may enhance **mitochondrial capacity** and **lower oxidative stress**.
- **Endurance exercise during ERT** may enhance **GAA uptake** by the working muscle by increasing blood flow, but evidence from a mouse model revealed no independent or synergistic effects on GAA activity or glycogen clearance in skeletal or cardiac muscle
 - However, improvements in running speed, endurance, balance and manual dexterity were noted in GAA-deficient mice receiving exercise alone or exercise + ERT
- Terzis et al. used a PRT program for 20 wks in 5 patients who received ERT for 1 year.
 - Improvements in muscle strength along with a significant increase in arm FFM
 - Increase in 6MWT was strongly associated with hip extension strength



Tarnopolsky M et al. Can J Neurol Sci. 2016
 Strothotte S et al. J Neurol. 2010
 Nilsson MI et al. Mol Genet Metab. 2012
 Terzis G et al. Mol Genet Metab. 2011

Management of LOPD: Exercise

- Overall, data from clinical studies and animal models suggest a possible benefit of endurance and resistance exercise in patients with Pompe disease that is independent of reducing muscle glycogen storage.
- **Low-impact or submaximal aerobic exercise, targeted training of specific muscles and exercises that improve balance may be prescribed.**
- **Exercise must be tailored to the patient's level of ability; however, the patient may be required to exercise beyond his or her level of perceived tolerance to obtain a benefit.**
- **There is no evidence to suggest that systematic activity restriction in Pompe disease is warranted.**



Management of LOPD: Functional Capacity, Fatigue and QOL

- Practical standpoint, at a minimum during clinic visits: inquiries about specific ADLs that reflect the underlying major aspects of the disease (limb and respiratory weakness).
 - **Getting in and out of bed**
 - **Meal preparation and eating,**
 - **Toileting, bathing**
 - **Ambulation (level walking and climbing stairs) and falls**
 - **Dressing**
 - **Shortness of breath on exertion, orthopnea, morning headaches**
 - **Sleep symptoms (e.g., apnoea, snoring, paroxysmal nocturnal dyspnea)**
 - **Muscle pain and fatigue**
 - **Speech and swallowing difficulties,**
 - **Heart palpitations.**

La progettualità riabilitativa

Definizione delle Aree di Problematicità

1. Area della stabilità internistica
2. Area del recupero delle funzioni vitali di base
3. Area delle funzioni sensomotorie
4. Area competenze comunicativo-relazionali
5. Area competenze cognitivo-comportamentali
6. Area autonomia e cura della persona
7. Area mobilità e trasferimenti
8. Area rieducazione e inserimento sociale

Problem-Related Approach

Diagnosi riabilitativa:
valutazione degli impairment, di attività e partecipazione

Identificazione degli outcome a breve e a lungo termine

Protocollo di valutazione funzionale

Pompe disease

Evaluation protocol

- ❖ Range of Motion (ROM)
- ❖ Manual Muscle Testing (MMT)
- ❖ Hand Grip Strength Test
- ❖ Unipedal Stance Time Test
- ❖ Functional Ambulation Category (FAC)
- ❖ Fatigue Severity Scale (FSS)
- ❖ Functional tests
- ❖ Pain
Brief Pain Inventory (BPI short form)
- ❖ Posture and Balance
Baropodometric examination
Tinetti Scale
- ❖ Disability
Barthel Index and Rankin Scale
- ❖ Bone Mass Density
DXA
- ❖ Quality of life
Short-Form 12 items Health Survey (SF-12)

stato cognitivo

Fuzioni esecutive	Memoria e apprendimento verbale	Velocità psicomotoria e attenzione
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<input type="checkbox"/> TMB	<input type="checkbox"/> Test di Rey	<input type="checkbox"/> TMA
<input type="checkbox"/> Matrici di Raven	<input type="checkbox"/> Test di Ricognizione	
<input type="checkbox"/> Stroop	<input type="checkbox"/> Span verbale e spaziale	
<input type="checkbox"/> D. Barrage		
<input type="checkbox"/> Copia Disegni		

+
Fluenza verbale
e MMSE

Danno cognitivo presente se 2 o più prove sono compromesse

Area funzioni vitali / stabilità internistica

Alterazioni della funzione respiratoria:

1. Alterazione della pompa ventilatoria (sistema nervoso, gabbia toracica, muscoli respiratori)
2. Ridotta espansione toraco-polmonare
3. Riduzione della frequenza e della profondità del respiro (prima durante il sonno)
4. Sindrome ostruttiva con focolai broncopneumonici e/o atelettasici
5. Tosse ipovalida e conseguente ristagno di secrezioni nelle vie aeree

Obiettivo del trattamento riabilitativo:

Ripristino della funzionalità respiratoria

Interventi riabilitativi:

- Fisioterapia respiratoria disostruttiva (in caso di complicanze ostruttive)
- Sollecitazioni meccaniche sull'addome
- Aspirazione delle secrezioni bronchiali
- Ventilazione meccanica

area senso-motoria

Alterazioni delle funzioni muscolari e correlate al movimento:

1. **Debolezza muscolare: 93% dei casi di Malattia di Pompe late-onset**

Muscoli del tronco

Muscoli del cingolo pelvico (flessori ed estensori, abduzioni e adduttori dell'anca)

2. **Riduzione del Range of Motion**
3. **Deformità scheletriche (contratture, scoliosi)**
4. **Affaticabilità**
5. **Dolore**

Limitazione delle attività:

1. **Difficoltà nel camminare, salire e scendere le scale, alzarsi dal pavimento**

Obiettivi del trattamento riabilitativo: Preservare il massimo livello di autonomia possibile attraverso:

- la prevenzione dell'estensione dei danni
- la prevenzione delle complicanze non necessarie
- il trattamento dei danni "disabilitanti"
- la ricerca di compensi interni
- lo sviluppo di supplenze

Interventi riabilitativi:

- Posizionamento corretto
- Mobilizzazione passiva
- Esercizio attivo, rinforzo selettivo
- Esercizi per il controllo posturale
- Ortesi e ausili

Muscle function, Activities and Pain in LOPD Patients

	SEX	AGE	BMI	MRC score	HGS test dx	HGS test sn	FSS	FAC score	BPI intensity	BPI interference	FIM
Vincenza	F	61	22,2	78	10	12	47	4	0	0	118
Maria	F	60	28	86	15	16	45	4	3,5	9,5	121
Anna	F	47	28,7	95	12	20	26	5	0,3	4,7	121
Concetta	F	54	33,1	97	21	22	19	5	0	0	124
Antonio	M	53	21,4	83	20,5	21	45	2	4	9,5	118
Mauro	M	56	28,1	93	22,5	22	12	4	1,3	0,6	122
Claudio	M	45	20,1	89	21	21	55	4	0	0	118
Olimpia	F	47	26,5	97	31,2	26,2	30	4	3.25	6.4	124
Anna Grazia	F	40	25,3	83	16	17	40	1	1.25	0	97
Morena	F	42	25	96	18.3	21	21	5	0	0	123
Diodato	M	51	31,8	100	66,1	52,6	18	5	0	0	126
Vincenzo	M	45	33,8	85	30,6	32,6	32	4	0	0	118

Bone Biochemical Parameters in LOPD patients

	SEX	AGE	BMI	Serum 25-OH D ₃ (ng/ml)	Serum PTH (pg/ml)	Calcium (mg/dl)	Phosphate (mg/dl)	Bone Alkaline Phosphatase (µg/l)
Vincenza	F	61	22,2	10,7	52	9,1	2,9	15
Maria	F	60	28	8,4	24	9	3,5	27
Anna	F	47	28,7	40,2	37	9,3	3,4	10
Concetta	F	54	33,1	34,6	40	9,5	4,5	22
Antonio	M	53	21,4	12,7	38	9	3,3	16
Mauro	M	56	28,1	28,4	26	9,2	5	17
Claudio	M	45	20,1	41,3	38	9,2	4,8	27
Olimpia	F	47	26,5	3,8	55	8,4	3,9	79
Anna Grazia	F	40	25,3	2,2	90	8,5	4	58
Morena*	F	42	25					
Diodato	M	51	31,8	8,9	48	8,7	3,3	80
Vincenzo*	M	45	33,8					

Esercizio fisico

- **Attività fisica:** qualsiasi movimento corporeo prodotto dai muscoli scheletrici che si traduce in un dispendio energetico superiore al livello basale
- **Esercizio terapeutico:** prescrizione di contrazioni muscolari e di movimenti corporei al fine di migliorare la funzionalità generale e specifica di un individuo per aiutarlo a rispondere al meglio alle esigenze della vita quotidiana , nell'ambito di un processo di recupero delle capacità di esecuzione di attività indispensabili alla sua partecipazione sociale.
- **Dimensioni dell'esercizio** – frequenza, intensità, tempo e tipologia – *“type is the most important for bone”*



International
Osteoporosis
Foundation

Prevenzione primaria del danno da ipomobilità

- La sedentarietà è minimo comune denominatore che determina e accelera il processo di disabilità nelle malattie croniche
- Attività Fisica Adattata (AFA) contrasta l'ipomobilità
- Sono esclusi dai programmi AFA i soggetti con instabilità clinica per i quali la risposta è esclusivamente sanitaria



Ministero della Salute

Piano d'indirizzo per la Riabilitazione

Gruppo di Lavoro sulla Riabilitazione

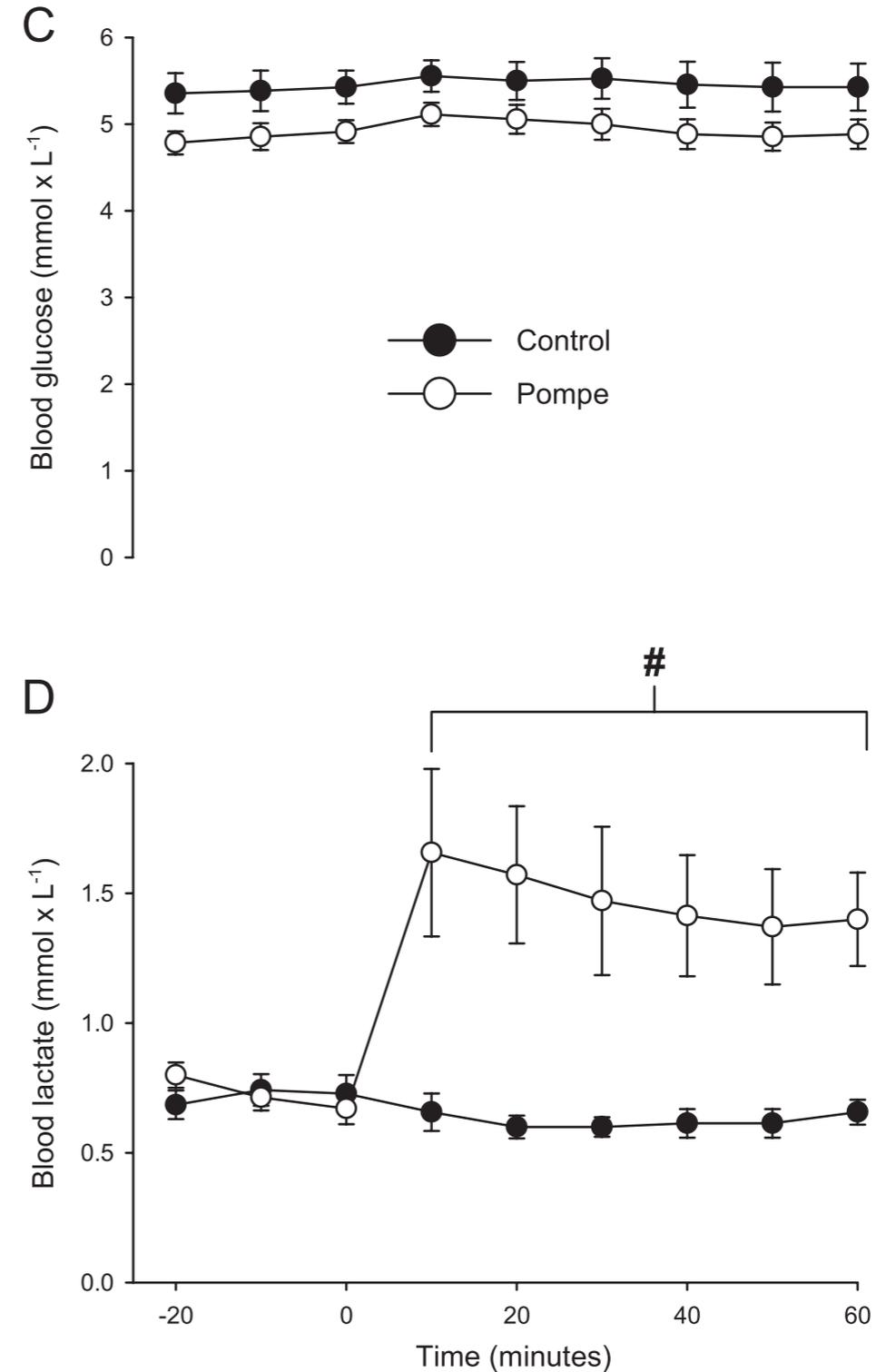
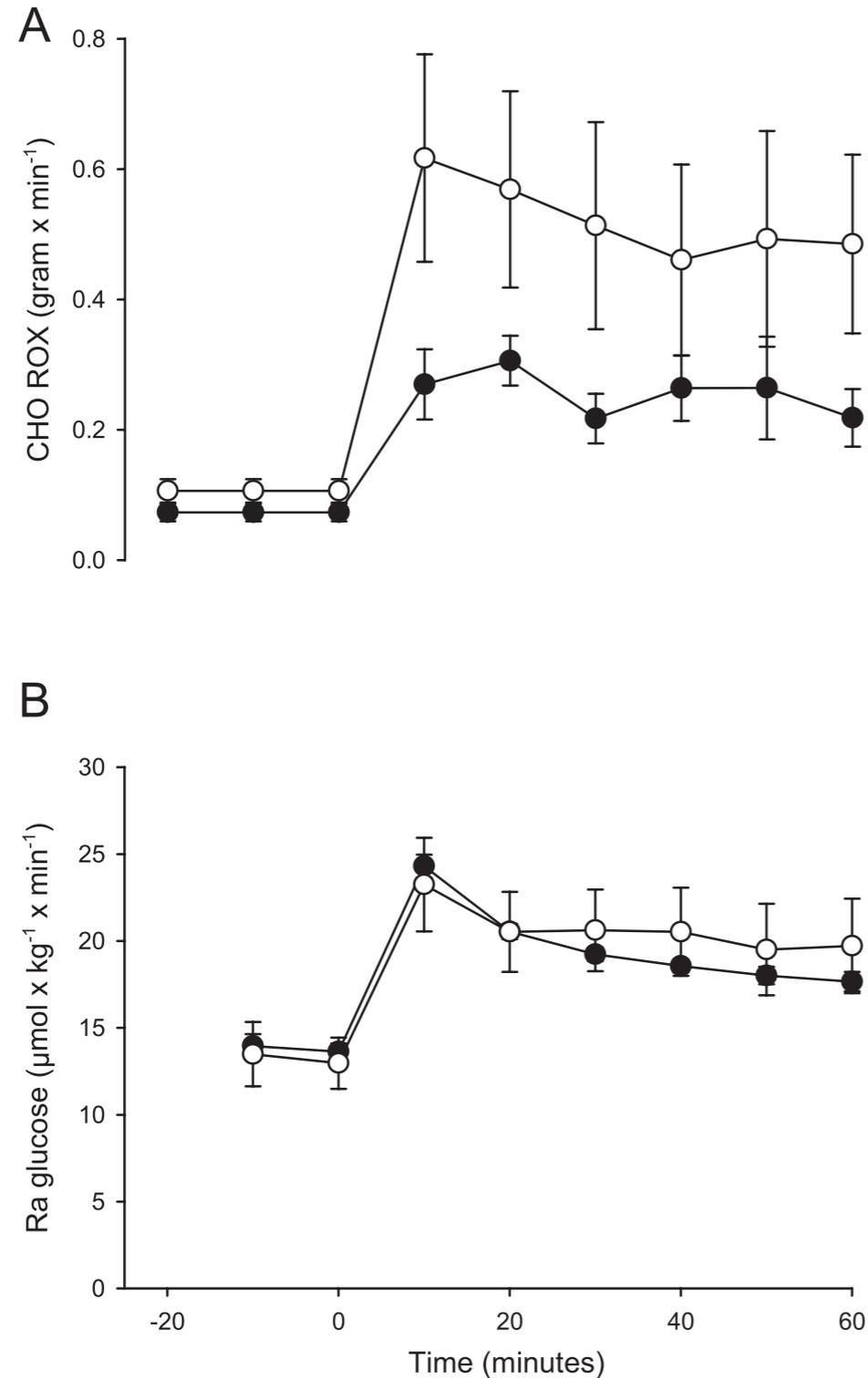
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L'Attività Fisica Adattata

- AFA è un termine "ombrello" per individuare un'area interdisciplinare, includente le attività d'educazione fisica, tempo libero, danza, sport, fitness e riabilitazione per individui con impairment
- è quindi rivolta a quelle persone che non sono in grado di partecipare con successo o in condizioni di sicurezza alle normali attività d'educazione fisica.
- Attraverso l'AFA si cerca di individuare percorsi formativi e d'attività "modificate", stabili o transitorie, che possano permettere alle persone con disabilità di condurre attività motorie.

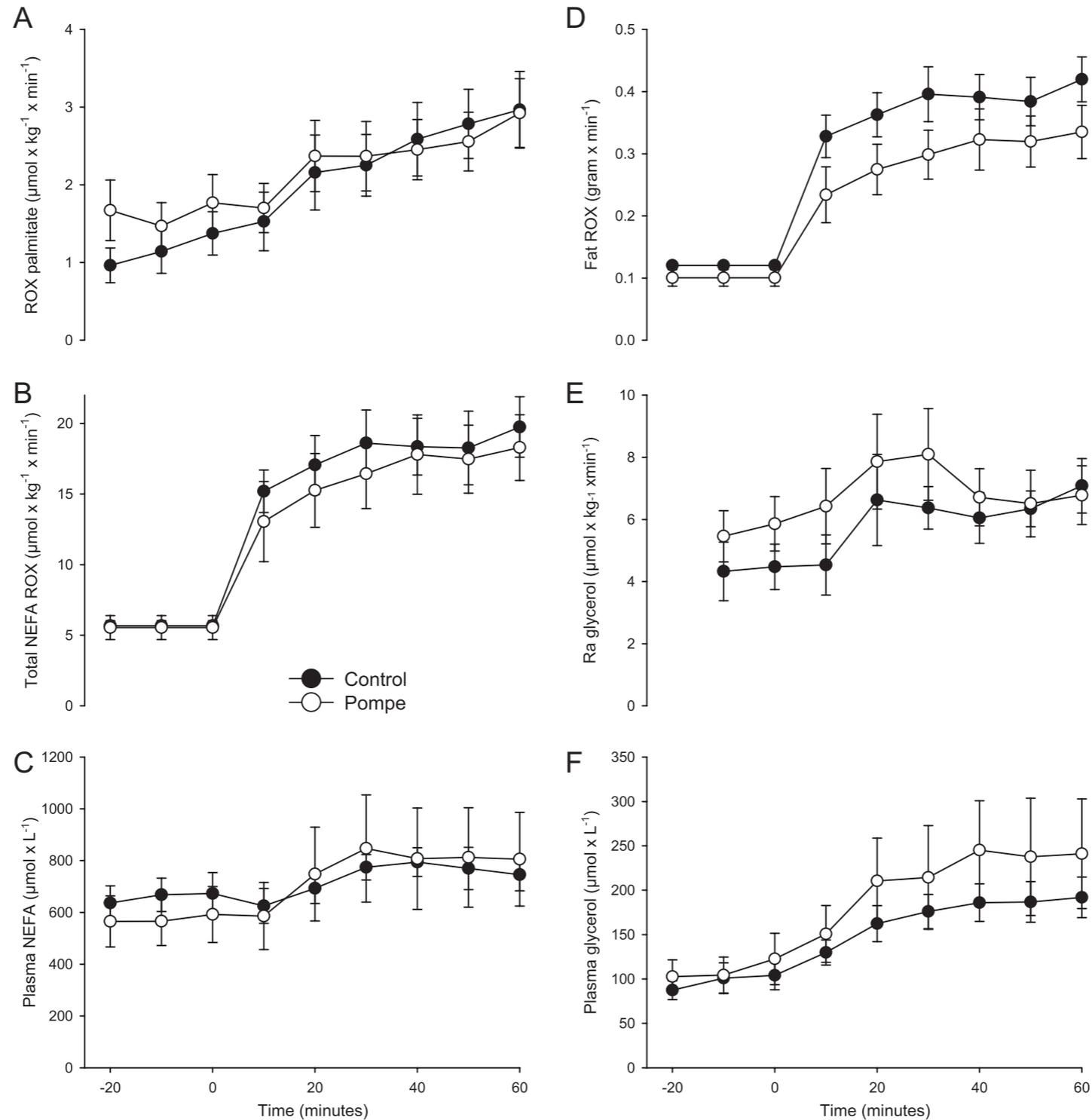
Skeletal muscle metabolism during prolonged exercise in Pompe disease

Nicolai Preisler¹, Pascal Laforêt², Karen Lindhardt Madsen¹, Edith Husu¹,
Christoffer Rasmus Vissing¹, Gitte Hedermann¹, Henrik Galbo³,
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In conclusion, **patients with LOPD** have a markedly **impaired maximal aerobic exercise capacity**.

On the other hand, the **patients' ability to endure prolonged submaximal exercise at a moderate intensity is normal**, because, unlike in other metabolic myopathies, in Pompe disease **skeletal muscle substrate use is normal during exercise**.

This supports the view that during exercise glycogen phosphorylase rather than alpha-glucosidase is decisive for stimulation of glycogen breakdown in liver and muscle.

Our findings **encourage use of regular exercise** in Pompe disease for recreational purposes as well as for improving physical function and health.

Conclusioni

- il paziente con m. di Pompe è un paziente complesso con variabilità genotipica e fenotipica
- L'approccio riabilitativo è mandatorio in tutte le fasi di malattia e va attuato mediante la formulazione di un Progetto Riabilitativo Individuale
- L'esercizio terapeutico anche di rinforzo contro resistenza o di endurance è sicuro, ma va modulato sul paziente
- La terapia enzimatica sostitutiva e l'esercizio terapeutico sono attualmente il gold standard per il management della malattia di Pompe
- Lo scopo ultimo è sempre l'empowerment del paziente facilitandone le attività e la partecipazione sociale, e migliorandone la qualità di vita