75° CONGRESSO NAZIONALE

FIMG Italiana Medici Con Professioni

Metis

Potenziare la medicina generale per migliorare l'Active Ageing

1-6 ottobre 2018

Complesso Chia Laguna - Domus de Maria (CA)

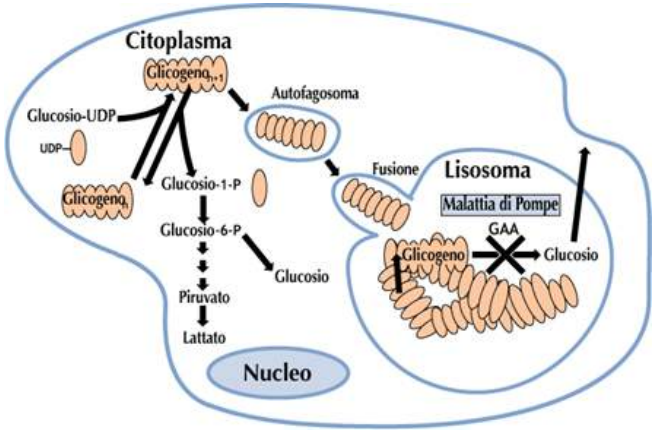
La Malattia di Pompe

Massimiliano Filosto

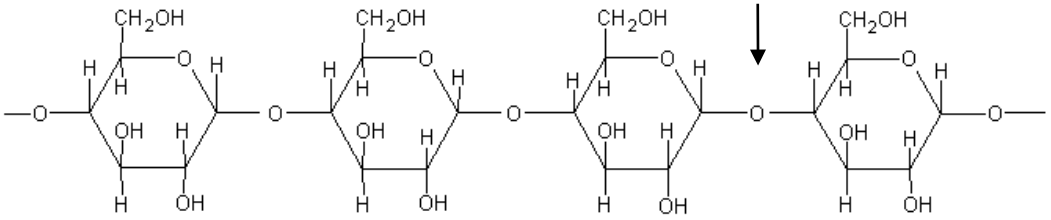
U.O. Neurologia
Centro per lo Studio delle Malattie Neuromuscolari
ASST “Spedali Civili” ed Università degli Studi di Brescia

Maltasi acida (alfa-1,4-glucosidasi acida)

- Enzima lisosomiale
- Dotata di attività alfa-glucosidasica → idrolizza il glicogeno in glucosio
- E' il solo enzima degradativo del glicogeno presente nei lisosomi
- E' codificato dal gene *GAA*
- Il deficit causa accumulo di glicogeno e dilatazione

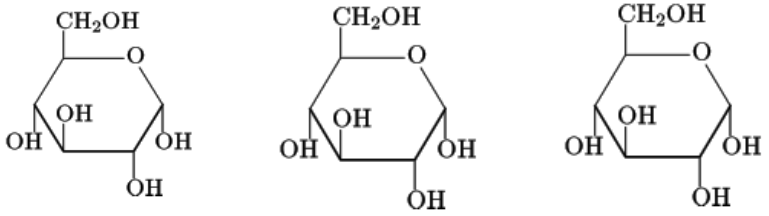


Legame α-1,4-glicosilico



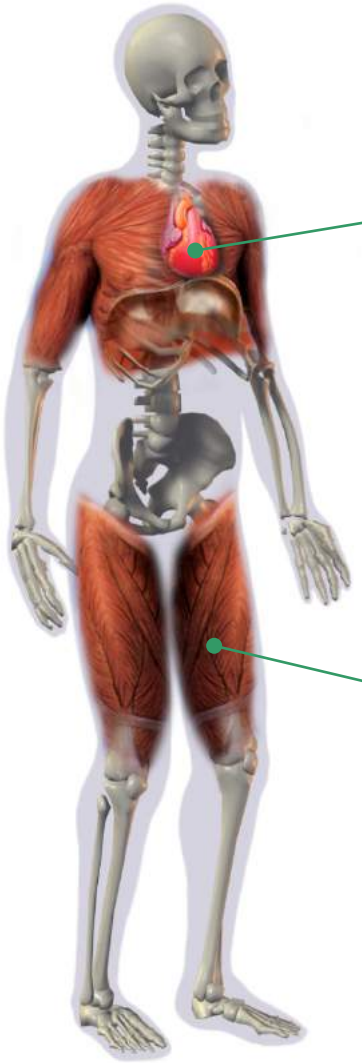
Glicogeno

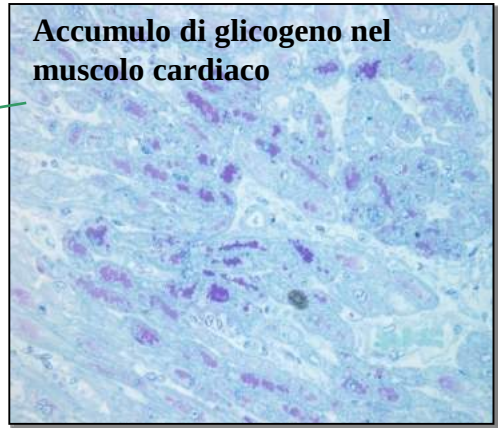
MALTASI ACIDA



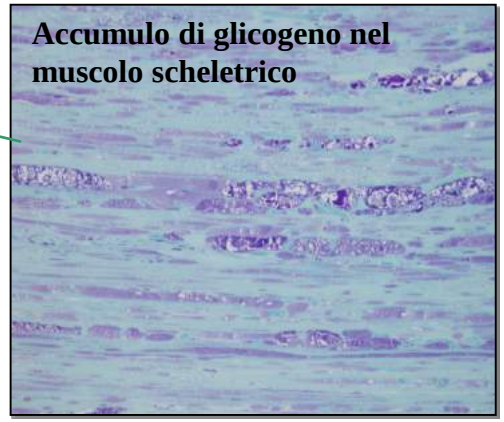
Molecole di glucosio

Coinvolgimento di organi chiave





Data on file, Genzyme Corporation.

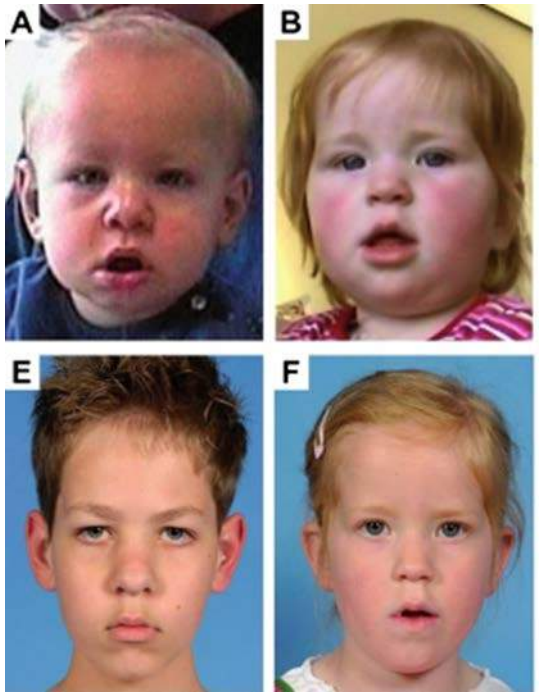


- L'accumulo di glicogeno **interessa principalmente il tessuto muscolare**
- Principali gruppi muscolari:
  - Muscolo cardiaco (infantile)
  - Muscolo scheletrico prossimale (in particolare arti inferiori e tronco)
  - Muscoli respiratori

Hirschhorn R, Reuser AJJ. Glycogen storage disease type II: acid alpha-glucosidase (acid maltase) deficiency.*The Metabolic and Molecular Bases of Inherited Disease.* 8th ed. New York: McGraw-Hill; 2001:3389-3420.

Clinical forms

- **INFANTILE FORM** - death within the first year of life due to progressive cardiac and respiratory involvement with macroglossia
- **LATE-ONSET FORM**
  - **Juvenile form** – it occurs between the two and the twenty years of age. Liver and cardiac involvement is possible
  - **Adult-onset form** - slowly-progressive; it affects mobility with proximal limb weakness and respiratory function (predominant diaphragmatic dysfunction); absence or mild involvement of cardiac muscle
- **POST-ERT FORM** - long-term survivors present improvement in cardiac and motor functions. Patients present residual facial muscle weakness, hearing loss, risk for arrhythmias, hypernasal speech, dysphagia with risk for aspiration, and osteopenia



J Inherit Metab Dis (2012) 35:505–511
DOI 10.1007/s10545-011-9404-7

ORIGINAL ARTICLE

Facial-muscle weakness, speech disorders and dysphagia are common in patients with classic infantile Pompe disease treated with enzyme therapy

C. M. van Gelder · C. I. van Capelle · B. J. Ebbink · I. Moor-van Nugteren · J. M. P. van den Hout · M. M. Hakkesteegt · P. A. van Doorn · I. F. M. de Coo · A. J. J. Reuser · H. B. W. de Gier · A. T. van der Ploeg

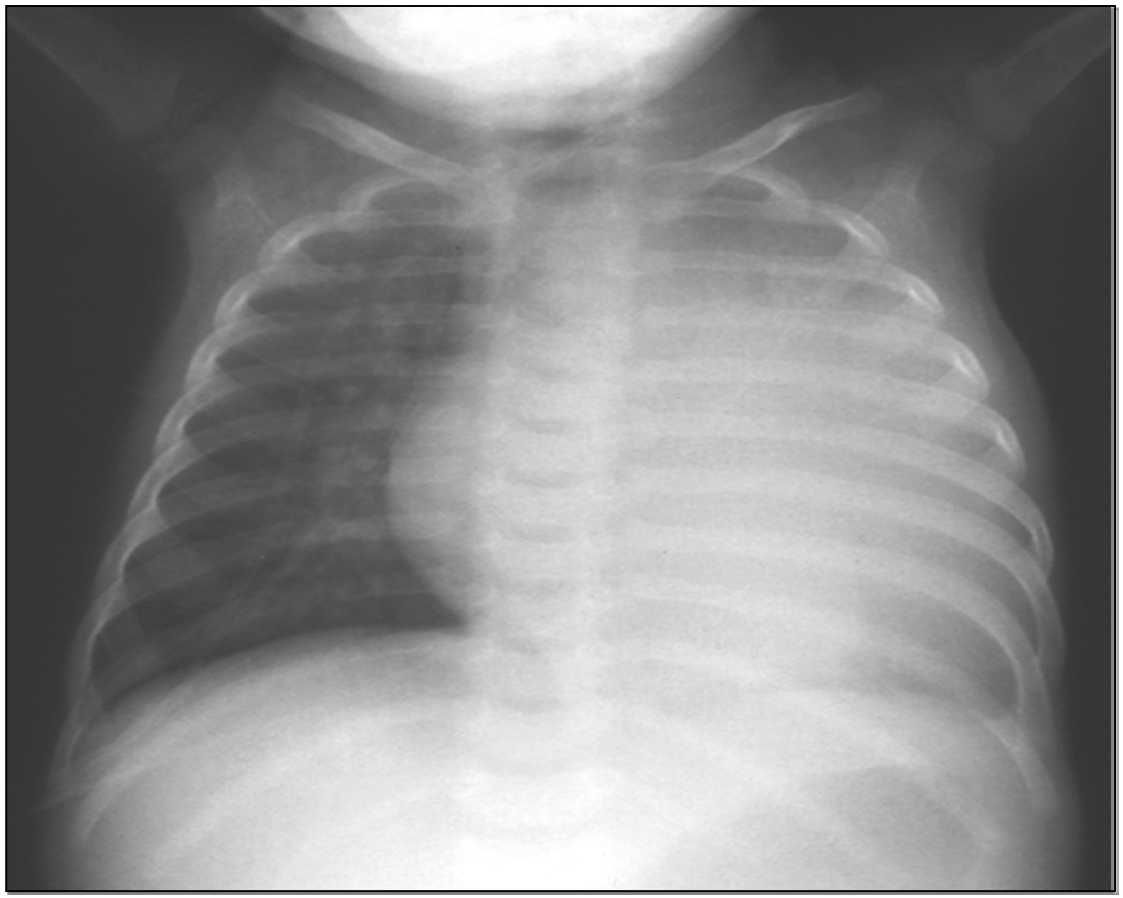
(Van Der Ploeg AT Lancet 2008)

Caratteristiche cliniche – forma infantile

Cuore	• Cardiomegalia/cardiomiopatia marcata • Progressione verso insufficienza cardiaca
Apparato muscoloscheletrico	• Debolezza muscolare profonda e a rapida progressione (ipotonia/bambino atonico/flessione all'indietro della testa) • Principali stadi di sviluppo motorio ritardati
Apparato respiratorio	• Infezioni respiratorie frequenti • Progressione verso insufficienza respiratoria • Morte dovuta a insufficienza cardio-respiratoria
Apparato gastrointestinale	• Difficoltà di alimentazione/mancanza di crescita • Organomegalia (epatomegalia/splenomegalia/macroglossia)

Hirschhorn R, Reuser AJJ. *Glycogen storage disease type II: acid alpha-glucosidase (acid maltase) deficiency. The Metabolic and Molecular Bases of Inherited Disease. 8th ed. New York: McGraw-Hill; 2001:3389-3420.*
Slonim AE, Bulone L, Ritz S, et al. *Identification of two subtypes of infantile acid maltase deficiency. J Pediatr. 2000;137:283-285.*

GSDII – forma infantile



- Cardiomegalia evidenziabile con RX torace
- Cardiomiopatia evidenziabile con Eco-cardio
- Progressione verso insufficienza cardiaca
- Anomalie ECG
  - Intervallo PR breve
  - Complessi QRS ampi

Hirschhorn R, Reuser AJJ. Glycogen storage disease type II: acid alpha-glucosidase (acid maltase) deficiency. *The Metabolic and*

Caratteristiche cliniche – forma late-onset

Apparato muscoloscheletrico	• Debolezza muscolare prossimale progressiva (in particolare arti inferiori e tronco) • Anomalie a livello di deambulazione • Dolore muscolare • Difficoltà a salire le scale • Cadute frequenti • Scapole alate
Apparato respiratorio	• Insufficienza respiratoria di vario grado • Ortopnea • Apnea notturna • Dispnea da sforzo • Infezioni respiratorie
Altro	• Sonnolenza durante il giorno • Cefalea mattutina • Osteopenia • Coinvolgimento delle funzioni cognitive superiori

Hirschhorn R, Reuser AJJ. *Glycogen storage disease type II: acid alpha-glucosidase (acid maltase) deficiency. The Metabolic and Molecular Bases of Inherited Disease. 8th ed. New York: McGraw-Hill; 2001:3389-3420.*

Debolezza prossimale

Debolezza nei muscoli del cingolo pelvico, dimostrata da manovra di Gower positiva

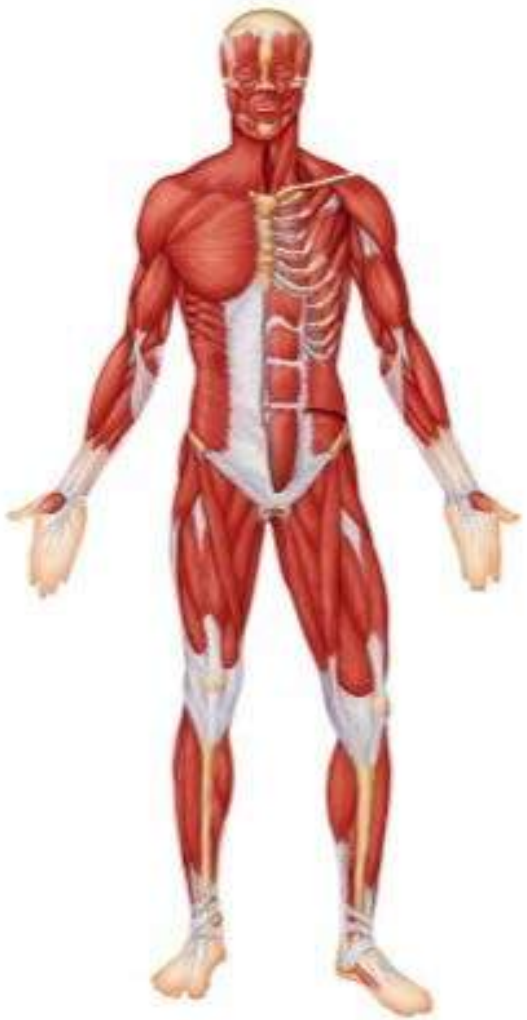
Hirschhorn R, Reuser AJJ. Glycogen storage disease type II: acid alpha-glucosidase (acid maltase) deficiency. *The Metabolic and Molecular Bases of Inherited Disease.* 8th ed. New York: McGraw-Hill; 2001:3389-3420.

GSD II – forma late-onset
Diagnosi differenziale

Diagnosi	Segni e sintomi in comune	Diagnosi differenziale
Distrofia muscolare progressiva scapolo-omerale e del cingolo pelvico	Debolezza muscolare prossimale progressiva a livello pelvico, di gambe o spalle	Muscoli del tronco (GSDII)
Distrofia muscolare Becker/Duchenne	Debolezza muscolare prossimale progressiva, distress respiratorio, difficoltà di deambulazione	Colpisce maschi e femmine (GSDII)
Polimiosite	Debolezza muscolare progressiva, spesso simmetrica	Possibile dolore muscolare, decorso relativ. rapido (PM)

Brown RH Jr, Mendell JR. Muscular dystrophies and other muscle diseases. In: Braunwald E, et al, eds. *Harrison's Principles of Internal Medicine.* 15th ed. New York: McGraw-Hill; 2001:2530-2539.
Dalakas MC Jr. Polymyositis, dermatomyositis, and inclusion body myositis. In: Braunwald E, et al, eds. *Harrison's Principles of Internal Medicine.* 15th ed. New York: McGraw-Hill; 2001:2524-2529.
Rosenkranz H. Polymyositis. Available at: www.emedicine.com. Accessed on August 25, 2005.
Hirschhorn R, Reuser AJJ. Glycogen storage disease type II: acid alpha-glucosidase (acid maltase) deficiency. In: Scriver CR, Beaudet AL, et al, eds. *The Metabolic and Molecular Bases of Inherited Disease.* 8th ed. New York: McGraw-Hill; 2001:3389-3420.
Chen Y-T. Glycogen storage diseases and other inherited disorders of carbohydrate metabolism. In: Braunwald E, et al, eds. *Harrison's Principles of Internal Medicine.* 15th ed. New York: McGraw-Hill; 2001:2281-2289.

Glicogenosi II - Eterogeneità



Eterogeneità Clinica

Eterogeneità Patologica

Eterogeneità Genetica

Eterogeneità clinica

Table 1. Clinical and laboratory data of 38 patients with GSD2

GSD2 phenotype	*Infantile*	*Juvenile*	*Adult/late-onset*
No. of patients	4	10	24
Age at onset (mean)	6.5 months	11.5 years	42.3 years
Brain			
Myelination defects	n.r./n.i.	n.r./n.i.	n.r/n.i.
Angiopathy	n.r.	n.r.	1
Eyes			
Retina	n.r./n.i.	n.r./n.i.	n.r./n.i.
Hearing			
Impaired	0	2	3
Heart			
Cardiac arrhythmia	0	1 (WPW)	2 (WPW)
Cardiomyopathy	4	1	0
Scoliosis	0	8	16
Muscle			
Hypotonia	4	0	0
Scapular winging	4	5	17
Trendelenburg's sign	–	3	21
Tongue enlarged	2	1	1
Proximal	4	10	24
Distal	4	5	10
Diaphragm	4	8	11
Axial	4	8	17
Liver			
Enlarged	4	1	1
Laboratory findings			
CK elevation	4 (1–10x)	10 (1–10x)	24 (1–10x)
LDH elevation	4	5	10
ASAT, ALAT elevation	4	4	4

GSD2, glycogen storage disease type 2; n.r., not reported; n.i., not identified; WPW, Wolf–Parkinson–White syndrome; CK, creatine kinase, LDH, lactate dehydrogenase; ASAT, aspartate aminotransferase; ALAT, alanine aminotransferase.

IperCKemia → **Mild muscle involvement** → **Respiratory involvement**

RESEARCH PAPER

LOPED study: looking for an early diagnosis in a late-onset Pompe disease high-risk population

O Musumeci,[1] G la Marca,[2] M Spada,[3] S Mondello,[1] C Danesino,[4] G P Comi,[5] E Pegoraro,[6] G Antonini,[7] G Marrosu,[8] R Liguori,[9] L Morandi,[10] M Moggio,[11] R Massa,[12] S Ravaglia,[4] A Di Muzio,[13] M Filosto,[14] P Tonin,[15] G Di Iorio,[16] S Servidei,[17] G Siciliano,[18] C Angelini,[6,19] T Mongini,[20] A Toscano,[1] the Italian GSD II group

Neuropathology and Applied Neurobiology (2007), 33, 544–559 doi: 10.1111/j.1365-2990.2007.00839.x

Adult-onset glycogen storage disease type 2: clinico-pathological phenotype revisited

B. G. H. Schoser*, J. Müller-Höcker†, R. Horvath*, K. Gempel‡, D. Pongratz*, H. Lochmüller* and W. Müller-Felber*

J Neurol (2015) 262:968–978
DOI 10.1007/s00415-015-7664-0

ORIGINAL COMMUNICATION

Clinical and molecular aspects of 30 patients with late-onset Pompe disease (LOPD): unusual features and response to treatment

Federica Montagnese · E. Barca · O. Musumeci · S. Mondello · A. Migliorato · A. Ciranni · C. Rodolico · P. De Filippi · C. Danesino · A. Toscano

Clinical Heterogeneity

- HEART
- RESPIRATORY
- MUSCLE
- GASTROINTESTINAL
- BONE
- BRAIN
- VASCULAR
- PERIPHERAL NERVES

Acta Myologica • 2013; XXXII: p. 91-94

Non-muscle involvement in late-onset Glycogenosis II

Massimiliano Filosto[1], Alice Todeschini[1], Maria Sofia Cotelli[1], Valentina Vielmi[1], Fabrizio Rinaldi[1], Silvia Rota[1], Mauro Scarpelli[2] and Alessandro Padovani[1]

Human Molecular Genetics, 2015, Vol. 24, No. 3 625–636
doi:10.1093/hmg/ddu476
Advance Access published on September 12, 2014

Peripheral nerve and neuromuscular junction pathology in Pompe disease

Darin J. Falk[1,2,*], Adrian Gary Todd[1,2], Sooyeon Lee[3], Meghan S. Soustek[1,2], Mai K. ElMallah[1], David D. Fuller[4], Lucia Notterpek[3] and Barry J. Byrne[1,2,*]

Small-Fiber Neuropathy in Pompe Disease: First Reported Cases and Prospective Screening of a Clinic Cohort

Lisa D. Hobson-Webb
Stephanie L. Austin
Sneha Jain
Laura E. Case
Karla Greene
Priya S. Kishnani

Bent Spine Syndrome as the Initial Symptom of Late-Onset Pompe Disease

Taisne Nicolas[1]; Desnuelle Claude, MD, PhD[2]; Juntas Morales Raul, MD[3]; Ferrer Monasterio Xavier, MD[4]; Sacconi Sabrina, MD, PhD[2]; Duval Fanny, MD[4]; Sole Guilhem, MD[4]; Flipo René Marc, MD, PhD[1]; Lacour Arnaud, MD[5]; Vermersch Patrick, MD, PhD[5]; Cardon Thierry, MD[1*]

Mauro Monforte
Institute of Neurology, Catholic University School of Medicine, Rome, Italy
Email: mauro.monforte@gmail.com

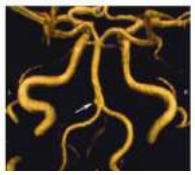
Serenella Servidei
Institute of Neurology, Catholic University School of Medicine, Rome, Italy

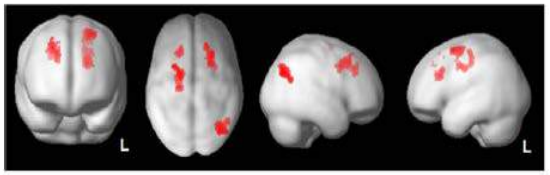
Enzo Ricci
Institute of Neurology, Catholic University School of Medicine, Rome, Italy

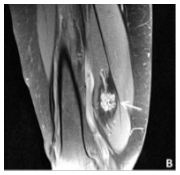
Giorgio Tasca
Institute of Neurology, Policlinico "A. Gemelli" Foundation University Hospital, Rome, Italy

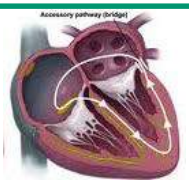
To the Editor

Fasciculations in Late-Onset Pompe Disease: A Sign of Motor Neuron Involvement?









EMG and MRI

EMG

Needle electromyography demonstrated spontaneous activity in 83% of adults.
Myotonic discharges were found in 72% of adults, often isolated to **the paraspinal muscles**.
Paraspinal examination is necessary in adults with symptoms suggestive of Pompe disease,
as abnormalities may be isolated to this region.

Clin Neurophysiol. 2011 Nov;122(11):2312-7
The clinical and electrodiagnostic characteristics of Pompe disease with post-enzyme replacement therapy findings.
Hobson-Webb LD, Dearmey S, Kishnani PS.

MRI

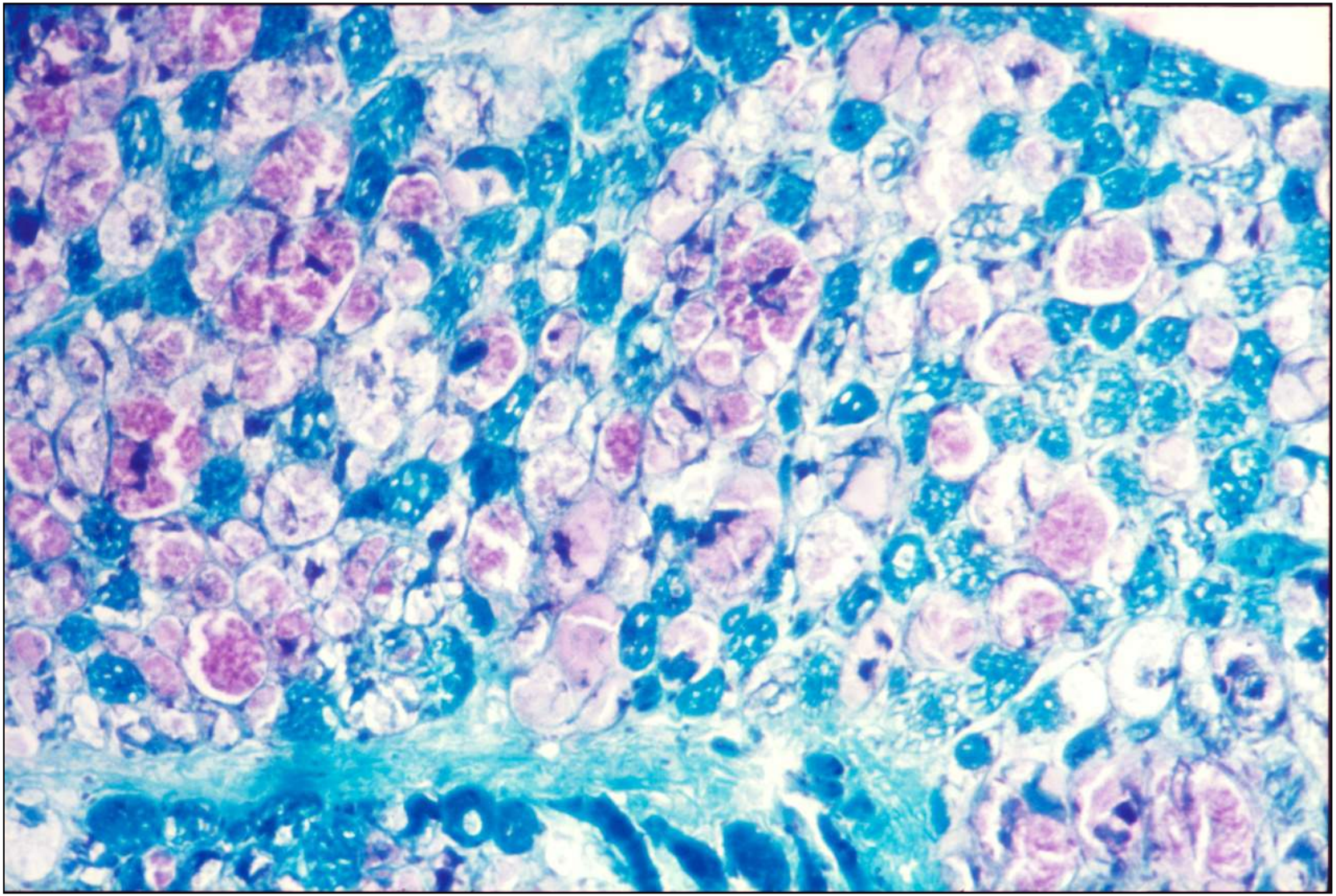
Muscle changes consisted of *internal bright signals of fatty replacement* without severe retraction of the muscles' corpus.
Spine extensors and pelvic girdle involvement.
Muscle changes also in the tongue and subscapularis muscle.
Thigh involvement is heterogeneous in terms of distribution across muscles as well as with respect to the overall clinical presentation.

The combination of paravertebral and abdominal muscle involvement may serve as a useful tool in the diagnostic work-up of patients with a clinical suspicion of Pompe disease;
Trunk abnormalities appear at very early stages of disease and even in asymptomatic patients, possibly "announcing" the onset of the disease and thus the need for a closer clinical follow-up.

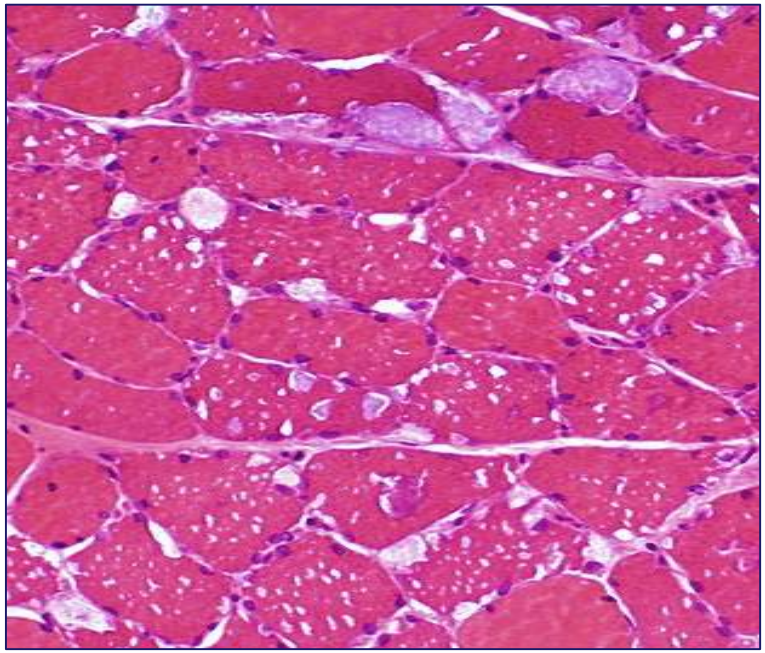
Whole-body muscle MRI in 20 patients suffering from late onset Pompe disease: Involvement patterns

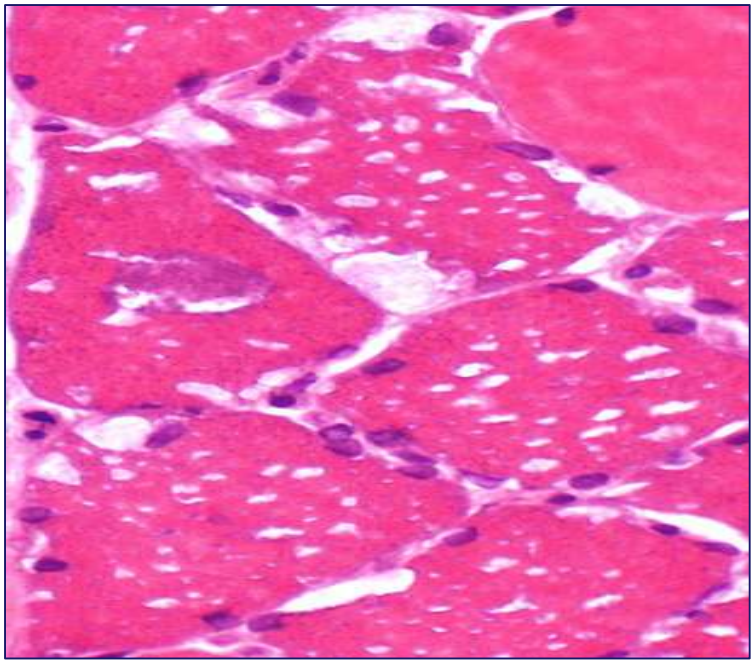
Robert-Yves Carlier[a,b,f,*], Pascal Laforet[c,d], Claire Wary[b], Dominique Mompoint[a], Kenza Laloui[c,d], Nadine Pellegrini[e], Djillali Annane[e,f], Pierre G. Carlier[b], David Orlikowski[e,f]

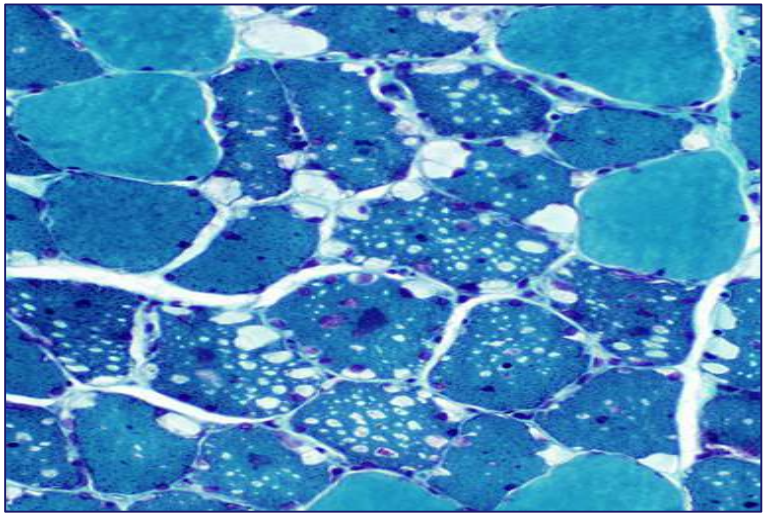
Neuromuscul Disord. 2012 Oct 1;22 Suppl 2:S148-54.
Trunk muscle involvement in late-onset Pompe disease: study of thirty patients.
Alejaldre A, et al

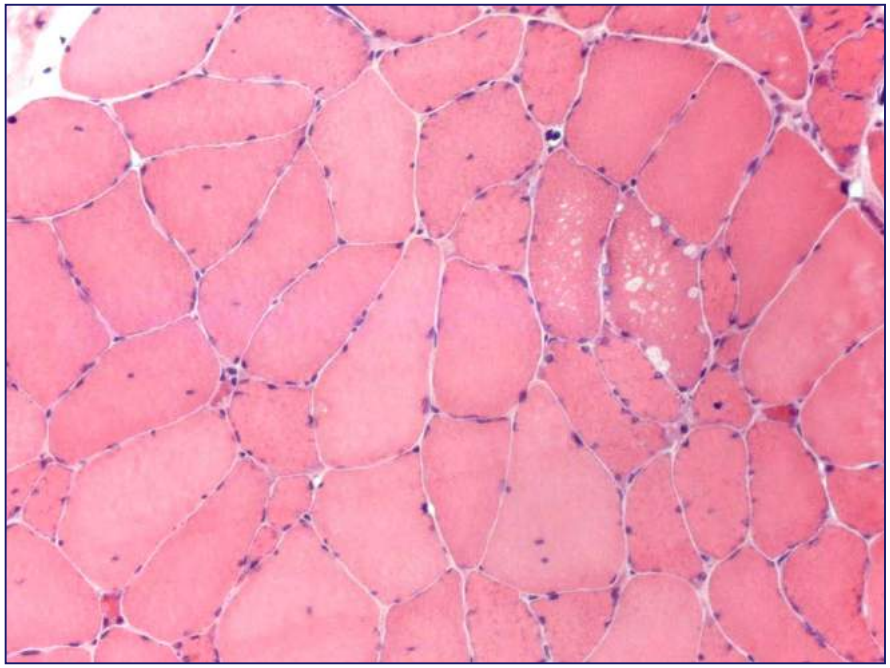


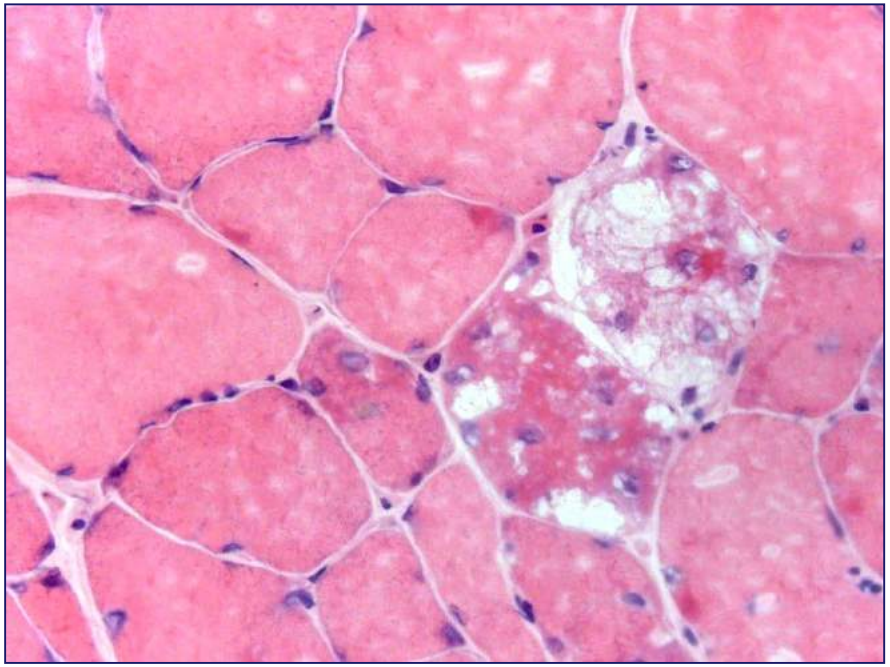
Late-onset form

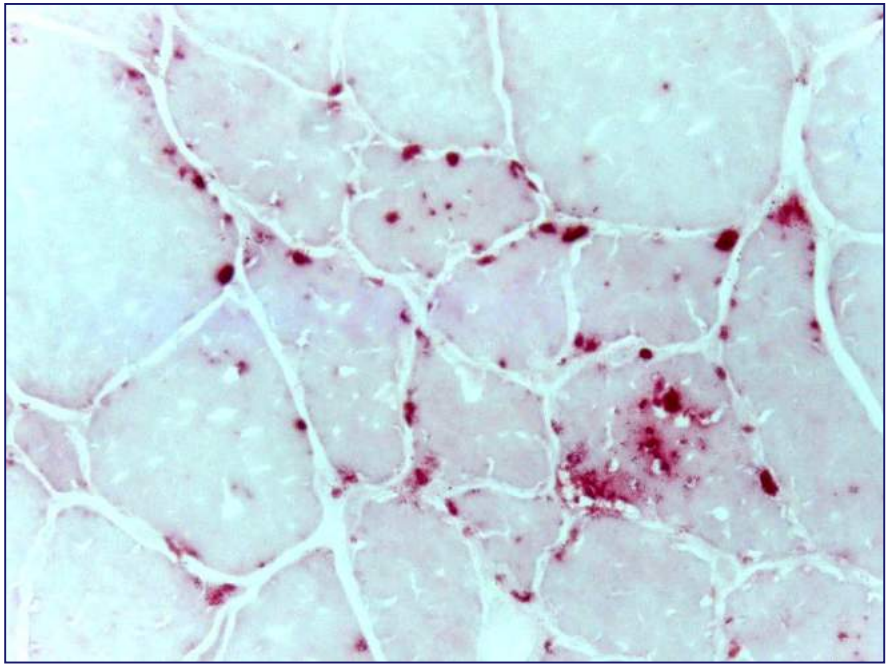


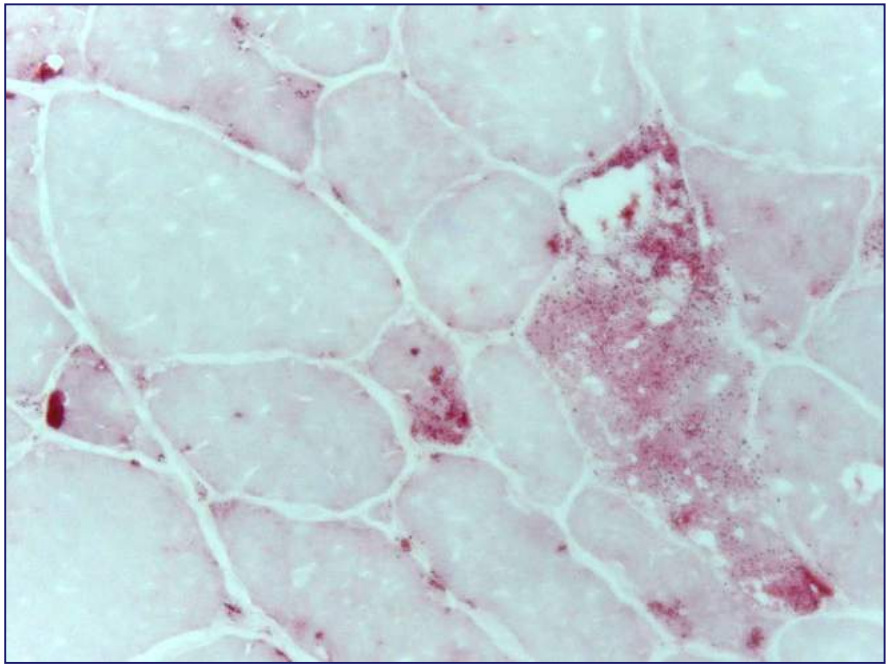












Biochemical diagnosis

SAMPLE	*ADVANTAGES*	*DISADVANTAGES*
LEUKOCYTES	Minimally invasive, a few steps to prepare.	Requires the use of acarbose to eliminate the interference of glucoamylase
PURIFIED LYMPHOCYTE	Minimally invasive	The preparation is more laborious compared to that required for the dosage on leukocytes
FIBROBLASTS	Availability of a cell line. Reliable assay with synthetic substrate	Results in 4-6 weeks. Laboratory equipped for cell culture. More invasive
MUSCLE BIOPSY	Histological evaluation and biochemical assay. Useful for the differential diagnosis with other myopathies	More invasive. The transport requires the use of liquid nitrogen. Frozen tissue
DRIED BLOOD SPOT (DBS)	Reliable first test for screening patients suspected of having Pompe disease	

Glycogenosis II: biochemical diagnosis

STUDIO LOPED

RESEARCH PAPER

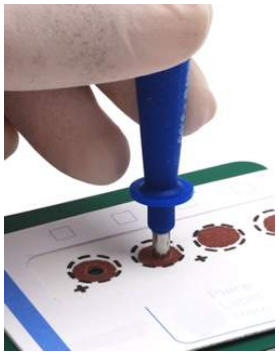
LOPED study: looking for an early diagnosis in a late-onset Pompe disease high-risk population

O Musumeci,[1] G la Marca,[2] M Spada,[3] S Mondello,[1] C Danesino,[4] G P Comi,[5] E Pegoraro,[6] G Antonini,[7] G Marrosu,[8] R Liguori,[9] L Morandi,[10] M Moggio,[11] R Massa,[12] S Ravaglia,[4] A Di Muzio,[13] M Filosto,[14] P Tonin,[15] G Di Iorio,[16] S Servidei,[17] G Siciliano,[18] C Angelini,[6,19] T Mongini,[20] A Toscano,[1] the Italian GSD II group

Multicenter observational study by the "Italian Group for the Study of Glycogen storage disease type II“ (Italian Association of Myology, AIM) designed to evaluate the prevalence of GSD II in a large population of patients at high risk for the disease, by means of different biochemical DBS methods:

-Fluorimetric technique

-Tandem mass spectrometry



Associazione Italiana di Miologia

17 pazienti positivi con deficit biochimico confermato

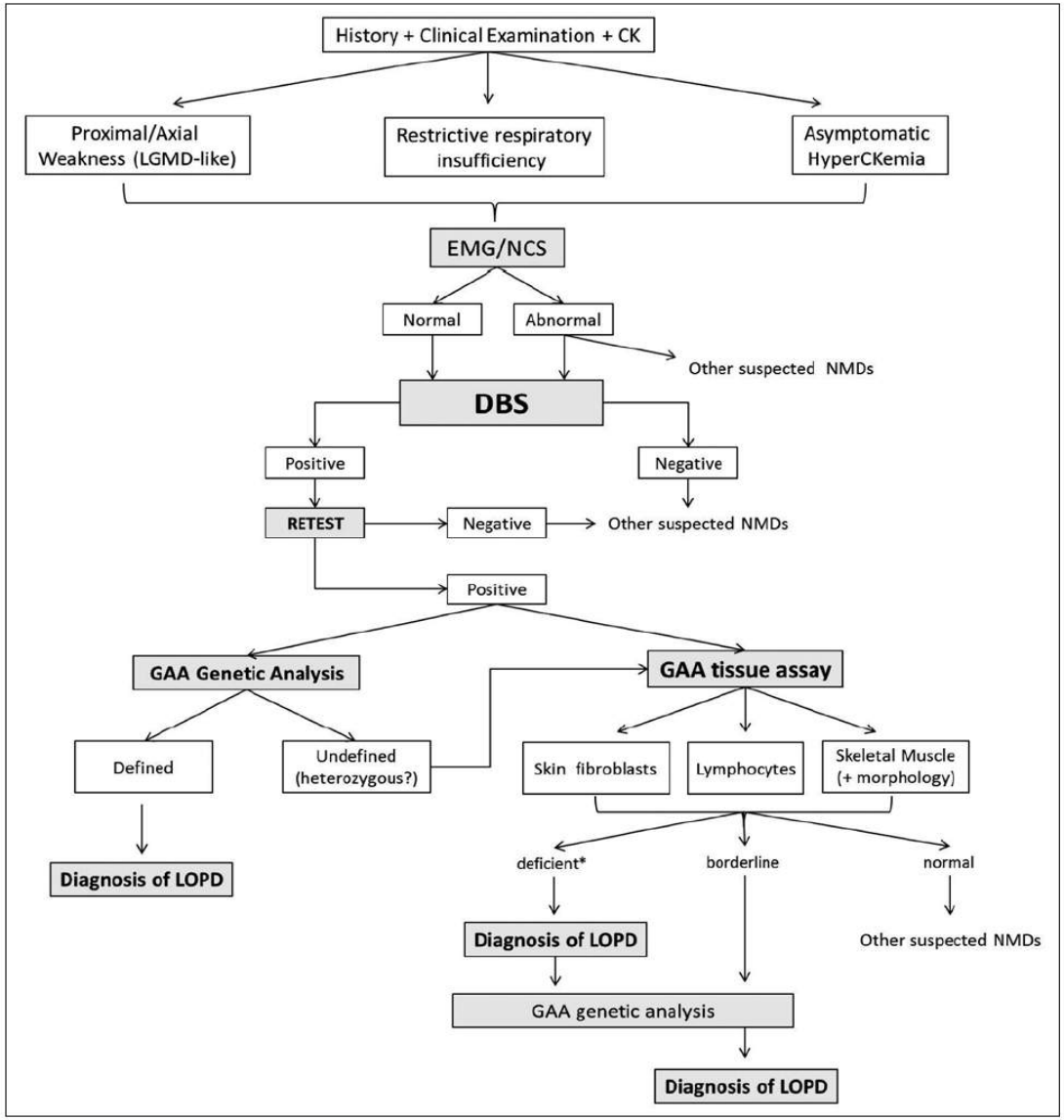


Figure 1. LOPD diagnostic algorithm.

(LOPD) Late-Onset Pompe Disease; (LGMD) Limb-Girdle Muscle Dystrophy; (EMG/NCS) electromyography and nerve conduction studies; (NMDs) neuromuscular disorders; (DBS) dried blood spot; (GAA) α-glucosidase A; *GAA activity <30%, borderline 30-40%.

Quando inviare il paziente?

History + Clinical examination + CK

- Proximal/axial weakness (LGMD-like)

- Restrictive Respiratory insufficiency

- Asymptomatic HyperCKemia

Centri di Riferimento

www. miologia.org
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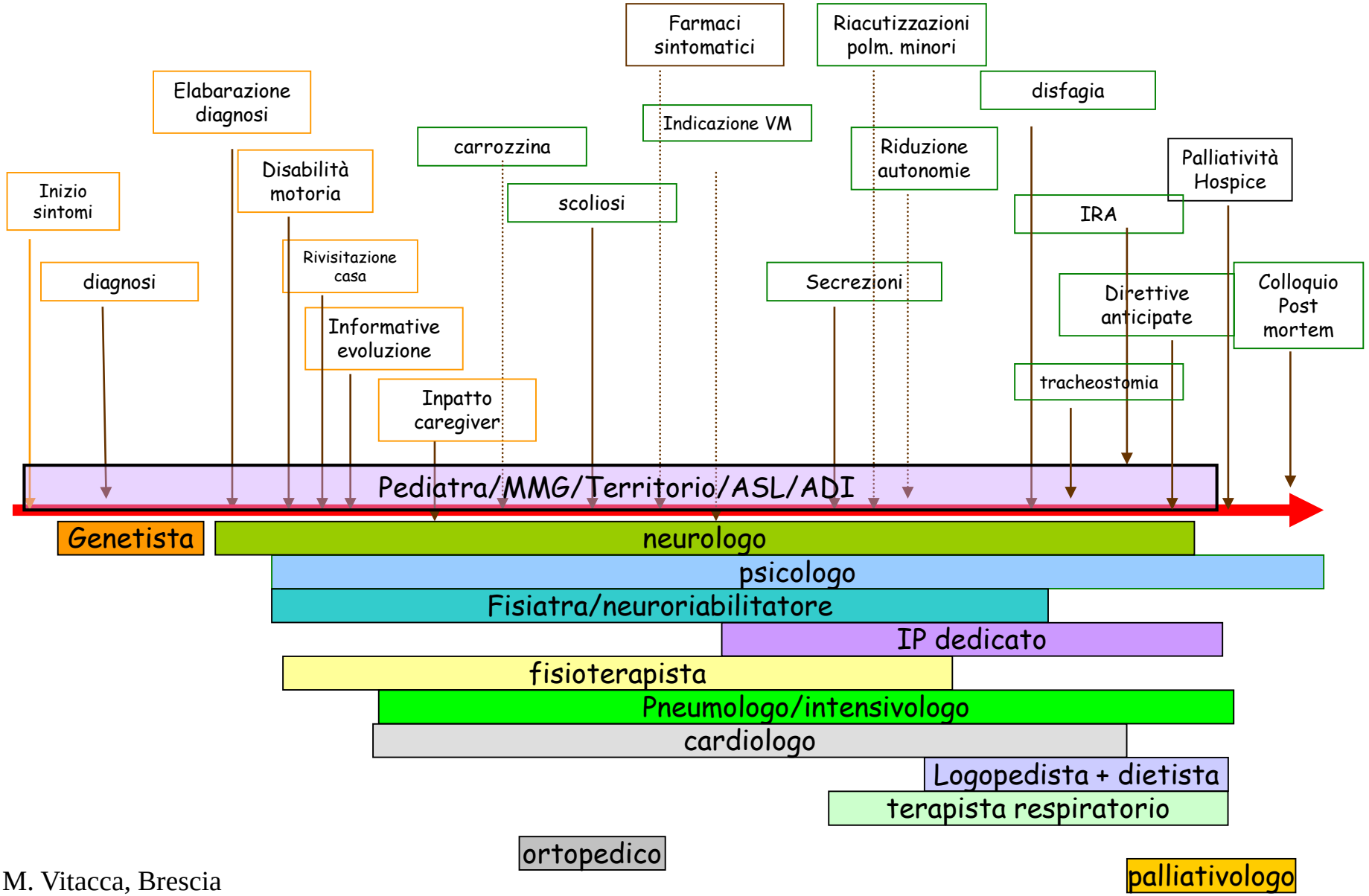
AIG:
ASSOCIAZIONE ITALIANA
GLICOGENOSI ONLUS

Malattie neuromuscolari



Dr. M. Vitacca, Brescia

Michele Vitacca
Massimiliano Filosto

Gestione integrata neurologica e pneumologica del paziente con glicogenosi tipo II

Multidisciplinary neurological and pneumological management of patients with glycogenosis type II

. Rassegna di Patologia dell'Apparato Respiratorio 2010; 25: 00

La necessità di una co-gestione pneumologico-neurologica dei pazienti adulti affetti dalla malattia ha aperto nuovi scenari nell'approccio a pazienti fino ad oggi sostanzialmente "sconosciuti".

L'insufficienza respiratoria polmonare è la principale causa di morbidità e mortalità tra i pazienti affetti dalla forma giovanile o adulta di GSDII.

Current Molecular Medicine 2014, *14*, 1-8

Late-Onset Glycogen Storage Disease Type 2

M. Filosto[*,1], M.S. Cotelli[1], V. Vielmi[1], A. Todeschini[1], F. Rinaldi[1], S. Rota[1], M. Scarpelli[2] and A. Padovani[1]

[1]*Clinical Neurology, Section for Neuromuscular Diseases and Neuropathies, University Hospital "Spedali Civili", Brescia, Italy*

[2]*Clinical Neurology, Department of Neurological, Neurosensorial, Morphological and Movement*

ERT: Myozyme (alglucosidasi alfa)

The recommended dose of MyozymeTM is 20 mg/kg of body weight administered every 2 weeks (14 days) with intravenous infusion.

Schema di Infusione di Myozyme (elaborato da10-Par. 4.2)
1 mg/Kg/h per 30 minuti
se Myozyme risulta ben tollerato passare a
3 mg/Kg/h per 30 minuti
se Myozyme risulta ben tollerato passare a
5 mg/Kg/h per 30 minuti
se Myozyme risulta ben tollerato passare a
7 mg/Kg/h per il restante periodo di infusione

Riferimenti bibliografici:
Elaborato dalla sezione 4.2. Myozyme^{TM} Riassunto Caratteristiche di Prodotto

ERT

The NEW ENGLAND JOURNAL of MEDICINE

ORIGINAL ARTICLE

A Randomized Study of Alglucosidase Alfa in Late-Onset Pompe's Disease

Ans T. van der Ploeg, M.D., Ph.D., Paula R. Clemens, M.D., Deyanira Corzo, M.D., Diana M. Escolar, M.D., Julaine Florence, P.T., D.P.T., Geert Jan Groeneveld, M.D., Ph.D., Serge Herson, M.D., Priya S. Kishnani, M.D., Pascal Laforet, M.D., Stephen L. Lake, Sc.D., Dale J. Lange, M.D., Robert T. Leshner, M.D., Jill E. Mayhew, P.T., Claire Morgan, M.D., M.P.H., Kenkichi Nozaki, M.D., Ph.D., Dorothy J. Park, M.D., Alan Pestronk, M.D., Barry Rosenbloom, M.D., Alison Skrinar, M.P.H., Carine I. van Capelle, M.D., Nadine A. van der Beek, M.D., Melissa Wasserstein, M.D., and Sasa A. Zivkovic, M.D., Ph.D.

Table 2. Results of Analysis of Covariance for Changes from Baseline to Week 78 for Primary and Secondary End Points.*

End Point	Alglucosidase Alfa Group (N = 60)	Placebo Group (N = 30)	Difference between Groups	P Value
Distance walked on 6-min walk test — m				
Baseline	332.2±126.7	317.9±132.3		
Week 78	357.9±141.3	313.1±144.7		
Change (95% CI)	25.13 (10.07 to 40.19)	−2.99 (−24.16 to 18.18)	28.12 (2.07 to 54.17)	0.03
Forced vital capacity — % of predicted				
Baseline	55.4±14.4	53.0±15.7		
Week 78	56.7±16.3	50.7±14.9		
Change (95% CI)	1.20 (−0.16 to 2.57)	−2.20 (−4.12 to −0.28)	3.40 (1.03 to 5.77)	0.006
Quantitative muscle testing, leg — % of predicted				
Baseline	37.7±18.9	32.5±18.2		
Week 78	39.1±21.8	30.4±20.5		
Change (95% CI)	1.18 (−1.07 to 3.42)	−2.00 (−5.16 to 1.17)	3.18 (−0.73 to 7.08)	0.11
Quantitative muscle testing, arm — % of predicted				
Baseline	55.9±20.4	56.9±18.2		
Week 78	60.9±21.7	58.3±20.9		
Change (95% CI)	5.05 (1.91 to 8.18)	1.47 (−2.92 to 5.87)	3.57 (−1.83 to 8.97)	0.19
Maximum inspiratory pressure — % of predicted				
Baseline	40.0±19.7	42.6±21.0		

Therapeutic Advances in Neurological Disorders *Original Research*

Progress in enzyme replacement therapy in glycogen storage disease type II

Corrado Angelini, Claudio Semplicini, Paola Tonin, Massimiliano Filosto, E. Pegoraro, Gianni Sorarù and Marina Fanin

Therapeutic Advances in Neurological Disorders
(2009) 2(3) 143–153
DOI: 10.1177/1756285609103324

J Neurol (2012) 259:952–958
DOI 10.1007/s00415-011-6293-5

ORIGINAL COMMUNICATION

Observational clinical study in juvenile-adult glycogenosis type 2 patients undergoing enzyme replacement therapy for up to 4 years

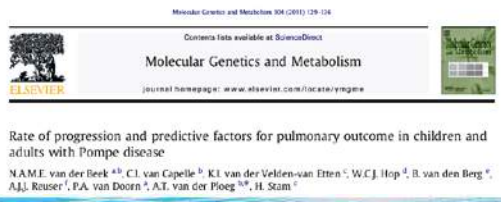
C. Angelini · C. Semplicini · S. Ravaglia · B. Bembi · S. Servidei · E. Pegoraro · M. Moggio · M. Filosto · E. Sette · G. Crescimanno · P. Tonin · R. Parini · L. Morandi · G. Marrosu · G. Greco · O. Musumeci · G. Di Iorio · G. Siciliano · M. A. Donati · F. Carubbi · M. Ermani · T. Mongini · A. Toscano · The Italian GSDII Group

Effects of the ERT

- **The muscle strength and respiratory functions deteriorate significantly before beginning therapy**

Following treatment:

- Significant increase in **muscle strength, pulmonary function and daily living activities,** followed by a stabilization of these parameters
- **Muscle function** improves in patients who are not dependent on a wheelchair and who have mild or moderate muscle weakness: it confirms the importance of the timely institution of therapy
- **Respiratory function** in the supine position improves in younger patients who are not dependent on artificial ventilation; Daily use of HMV and hospitalization rate are lower in ERT treated patients; **ERT reduces ventilatory dependence**


Molecular Genetics and Metabolism

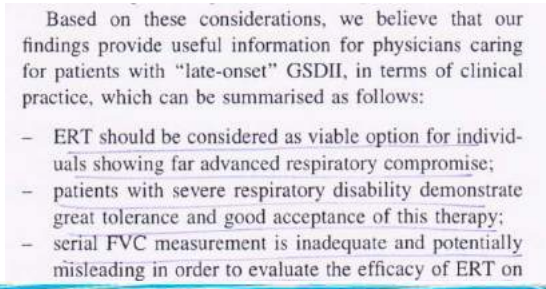
Rate of progression and predictive factors for pulmonary outcome in children and adults with Pompe disease


Lung (2013) 191:537–544
DOI 10.1007/s00408-013-9489-x

NEUROMUSCULAR DISEASE

Enzyme Replacement Therapy Improves Respiratory Outcomes in Patients with Late-Onset Type II Glycogenosis and High Ventilator Dependency

Andrea Vianello · Claudio Semplicini · Luciana Paladini · Alessandra Concas · Sabrina Ravaglia · Serenella Servidei · Antonio Toscano · Tiziana Mongini ·


Based on these considerations, we believe that our findings provide useful information for physicians caring for patients with "late-onset" GSDII, in terms of clinical practice, which can be summarised as follows:

- ERT should be considered as viable option for individuals showing far advanced respiratory compromise;
- patients with severe respiratory disability demonstrate great tolerance and good acceptance of this therapy;
- serial FVC measurement is inadequate and potentially misleading in order to evaluate the efficacy of ERT on

Available online at www.sciencedirect.com
ScienceDirect
Neuromuscular Disorders 25 (2015) 674–678
www.elsevier.com/locate/nmd

Workshop report

208th ENMC International Workshop: Formation of a European Network to develop a European data sharing model and treatment guidelines for Pompe disease Naarden, The Netherlands, 26–28 September 2014

Benedikt Schoser [a], Pascal Laforêt [b], Michelle E. Kruijshaar [c], Antonio Toscano [d], Pieter A. van Doorn [e], Ans T. van der Ploeg [c,*] on behalf of the European Pompe Consortium (EPOC)

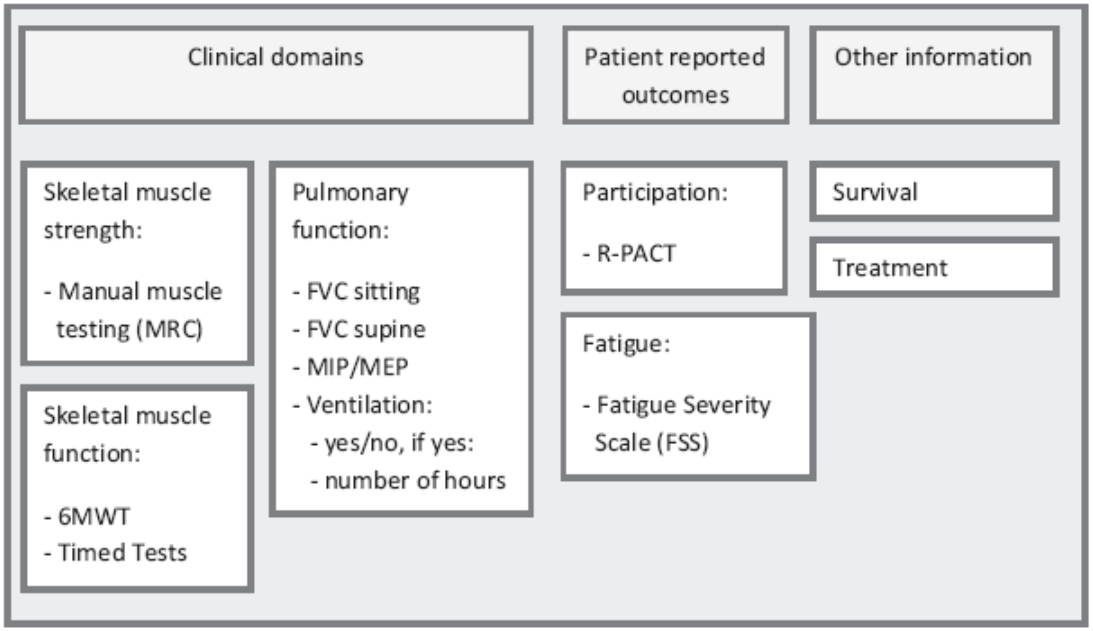


Fig. 1. Domains affected in adult Pompe patients and minimal set of outcome measures. MRC – the Medical Research Council grading scale (ranging from 0 to 5); 6MWT – six minute walk test; Timed tests: walking 10 metres, climbing four steps, standing up from supine position and standing up from a chair; MIP/MEP – maximum inspiratory/expiratory pressure, FVC – forced vital capacity; R-PACT – Rasch-built Pompe-specific activity scale, FSS – fatigue severity scale.

Transizione dalle cure pediatriche a quelle dell'adulto

La Società Americana di Medicina dell'adolescente ha definito la fase di transizione come

"un passaggio, programmato e finalizzato, di adolescenti e giovani adulti affetti da problemi fisici e medici di natura cronica da un sistema di cure centrato sul bambino ad uno orientato sull'adulto"

Malattia di Pompe in transizione - Rischi

Creazione di un gruppo di pazienti con problematiche cliniche diverse dai pazienti infantili e dai pazienti ad esordio tardivo

Difficoltà di gestione

Assenza di coordinazione gestionale

Creazione di “Malati orfani”

Aknowledgment

ASST «Spedali Civili» and University of Brescia
Irene Volonghi
Silvia Rota
Filomena Caria
Serena Gallo Cassarini
Enrico Baldelli
Mattia Marchesi
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Anna Galvagni
Alessandro Padovani

ASSOCIAZIONE ITALIANA
MIOLOGIA
ITALIAN ASSOCIATION OF MYOLOGY