eamda

European Alliance of Neuromuscular Disorders

35th Annual General Conference & Meeting

22. - 25. September
2005 Zagreb

Croatia

Dear representatives of neuro-muscular patient organizations in Europe,

The board of the European Alliance of neuro-Muscular Disorders Associations (EAMDA) is very happy to announce our plans and our conference in Zagreb.
As we decided during our AGM in 2003 EAMDA would deal with issues in the psychosocial area and the care area but also target cure, research and development of medicines for NMD. Our conference in 2004 was dedicated to research and cure. We are very happy to announce that Dr. Peter Streng from the Netherlands offered to take the lead in the field of cure, research and development of medicines. He will represent EAMDA in organizations like Epposi and Eurordis and in Brussels and Luxemburg (EU). EAMDA will become a member of both organizations. We realize that because of many developments in ENMC but more over in the research field multicentre trials will be an opportunity to realize in all the countries in Europe. Our members are hoping for a cure and little by little this is becoming a more realistic possibility. That is why we have to unite our common force and idealism.
In the upcoming years we are planning to organize our EAMDA conferences as parallel sessions during the WMS conferences once every two years when the WMS is in Europe, with topics about research and development. Dr. Peter Streng is offering to organize these sessions. The conference in the other years could have themes like we have know "respiratory care", physiotherapy, in other words subjects who are related to care and psycho social issues.
At this moment EAMDA has got 23 members. We are looking forward to have other muscular dystrophy associations to re-join EAMDA as Rusia is going to do in Zagreb.
We, the board of EAMDA, are very proud to announce these developments. Moreover we are very thankful to Mr Tomislav Goll and Ms Lilo Piquet who organized and sponsored our conference in Zagreb. We made the choice to move our conference to Zagreb to give the opportunity to other members who may find it easier to attend to Zagreb.
It is a very interesting program for all of you. That is why we invite you all members and non-members to come.

We hearty welcome you all!!!!

The EAMDA board

HOST OF EAMDA 2005. AGM MEETING

CROATIAN ALLIANCE OF MD SOCIETES

SDDH

Alliance of neuro-muscular societies has been established in 1970. Alliance is social-humanitarian association of voluntarily united non-party and non-profit societies-communities from municipal, town or county societies which gathers individuals which have muscular dystrophy and similar neuromuscular illnesses in the area of republic Croatia. Its purpose is realization and adjustment of common interests, in order to advance the rehabilitation and protect people with muscular dystrophy and similar neuromuscular illnesses.

Alliance gathers 30 basic societies. We will quote some of its tasks. Alliance accompanies, analyzes and considers rights regulated in the area of special protections of muscular dystrophy people, their life and work conditions. Converts and cooperates, propose, all factors that solve problematic of disabled persons in republic Croatia at all levels of state, regarding the legal and other regulations for the promotion of the legal, social and economic status of sick from the dystrophy and pawns for their consistent use. Cooperate with Government of the republic Croatia, Ministries, Parliament. Encourages and helps the foundation of neuromuscular society's in the area, these adjust and directs activities and cooperation of the county, town and municipal society of disabled persons. Also proposes and realized actions from common interests for members.

World Health Organization estimates that in Croatia lives around 4.500 individuals that have neuromuscular illnesses. On the initiative and pleasure of Alliances and people that have dystrophy we have started with the work of Referential centre for sick individuals by KBC "Rebro" in Zagreb. Diagnostics these illnesses bases on the clinic review, electromyography, testing of serum enzymes and other biochemical analyses, electronic-microscopic and light-microscopic analyses, biopsy of obtained tissue,

The real cause of muscular dystrophy is still unknown. The past researches have shown that were genetic disorders that cause changes in muscles and nerves. In the great number of cases illness is hereditary. **Illness is not contagious**.

Alliance is member of the world and Euro-associations such as: EAMDA - European Alliance of Muscular Dystrophy Associations (president of Alliance, Mr Tomislav Goll is also the vice-president of EAMDA at the moment), then International Alliance of ALS/MND Associations, and the member of WAMDA -

World Alliance of Muscular Dystrophy Associations.

Through its daily activity, among others, Alliance converts the public and electronic media with the aim of better informing about problems of members, as well as the promotion of quality of living of neuromuscular people, and also getting to know public with muscular dystrophy. For that purpose Alliance published its own magazine GEN and has started web portal on the address: www.sddh.hr

Alliance, traditionally, four years in a row organizes the Croatian Conference of Neuromuscular Person, and this year is the host of the largest European set of neuromuscular associations, 35th annual conferences and assemblies EAMDA from 22nd till 25th September in Zagreb. By chance or not, 35th conference corresponds with 35 year of work and existences of SDDH, so the reason for the celebration is larger.

With kind regards!

Tomislav Goll,
president

our GEN

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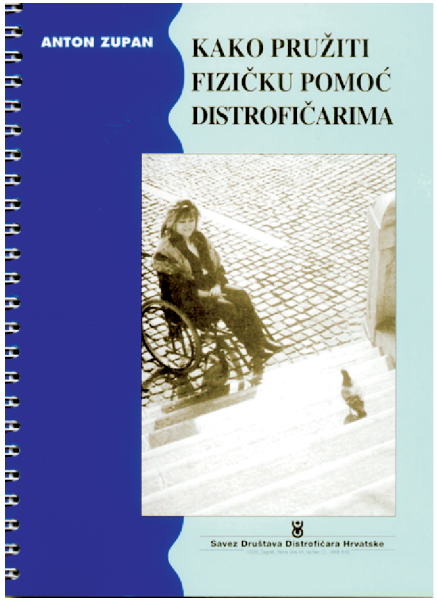
our publications



dr. Kovač, Ida
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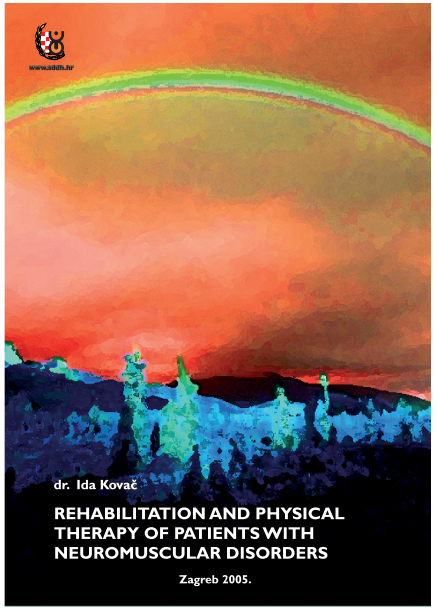
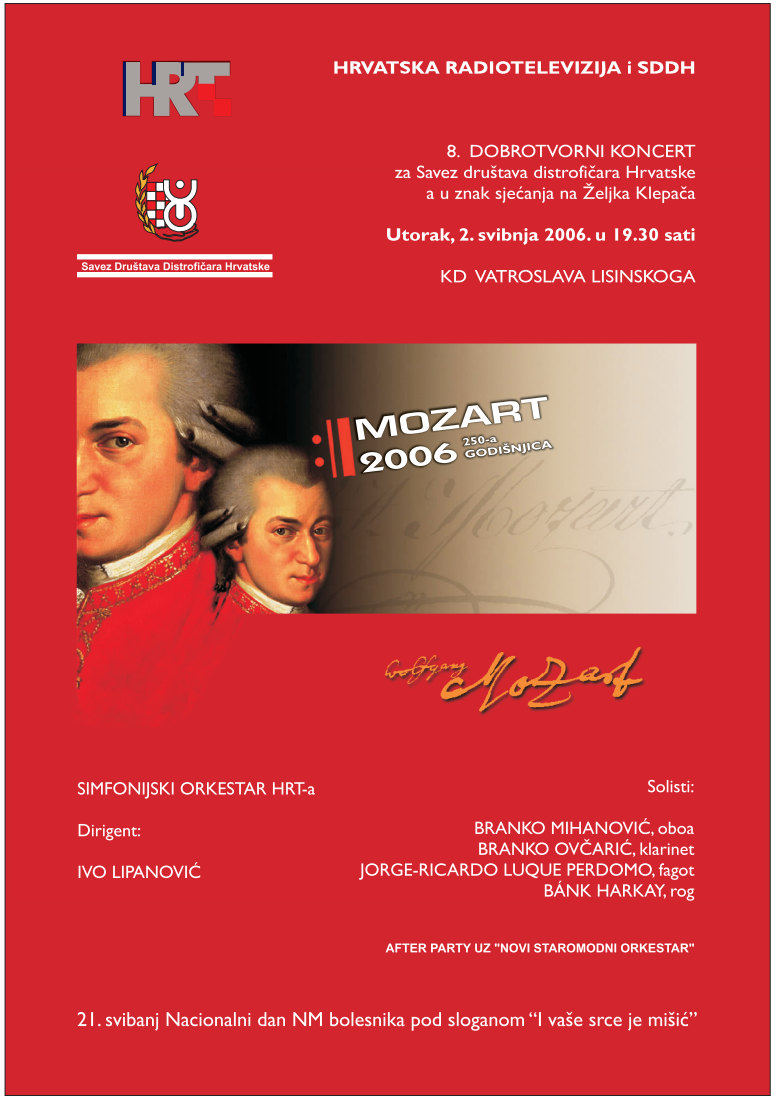


BOOK CONTAINS MULTIMEDIA CD WITH
VIDEO PRESENTATIONS OF ALL EXERCISES

Croatian Conference of Neuromuscular Patients

EXPERT AND SCIENCE CONFERENCES

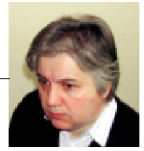
OUR CONCERTS OF CLASSICAL MUSIC



Alliance, traditionally, 8 years in a row organizes excellent concerts of classical music in concert hall Vatroslav Lisinski in Zagreb as major found raising event of the year. With money that we collect in this event we help our members or some of our activities, and concerts are excellent promotion of Alliance in public and media!

presidency of the Alliance

Mr Tomislav Goll President of Alliance
Slavonski Brod
brings more than 20 years of experience in policy advocacy, fundraising, coalition building, and organizational management to the presidency of the Alliance.



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Zagreb

Ms Marica Mirić Executive Vice President
Zagreb

Mr Goran Razum Secretary-General
Osijek

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Mr Zdravko Jakopović - Sisak
Ms Gordana Jurčević - Karlovac
Mr Davor Komar - Pula
Ms Lidija Muršić - Čakovec
Ms Mila Udiljak - Rijeka
Mr Nenad Vulas - Split
Ms Anđelka Zelenko - Slatina

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YOUR HEART IS A MUSCLE TOO!

21. May National Day of Neuromuscular patients

sddh@zg.t-com.hr

www.sddh.hr

eamda

European Alliance of Neuromuscular Disorders Associations

35. ANNUAL GENERAL MEETING

22.-25.09. 2005.

ZAGREB CROATIA

Home mechanical ventilation in Neuromuscular diseases Organizational aspects

EAMDA

35th Annual general meeting

September 2005, Zagreb

Patrick Leger MD

Introduction

- **Mechanical ventilation changes drastically the prognosis of NMD**
  - **Improving survival and quality of life especially for the less progressive diseases**
- **Non invasive ventilation is particularly interesting**
  - **Can be proposed as soon as hypoventilation appears**
  - **More affordable for patients and for family**
  - **Can be the first step to more complex care like tracheotomy and nutritional support**

DMD population in Denmark

(Jeppesen & al Neuromuscular Disorders 2003; 13: 804-812)

- Average age from 11 y (1977) to 17 y (2002)
- Oldest from 19 y (1977) to 40 y (2001)
- Rises in prevalence of DMD patients older than 19 y largely coincided with the establishment of the respiratory centers, but also with introduction of measures to prevent respiratory complications
- 80% of the DMD patients older than 19 are ventilator users

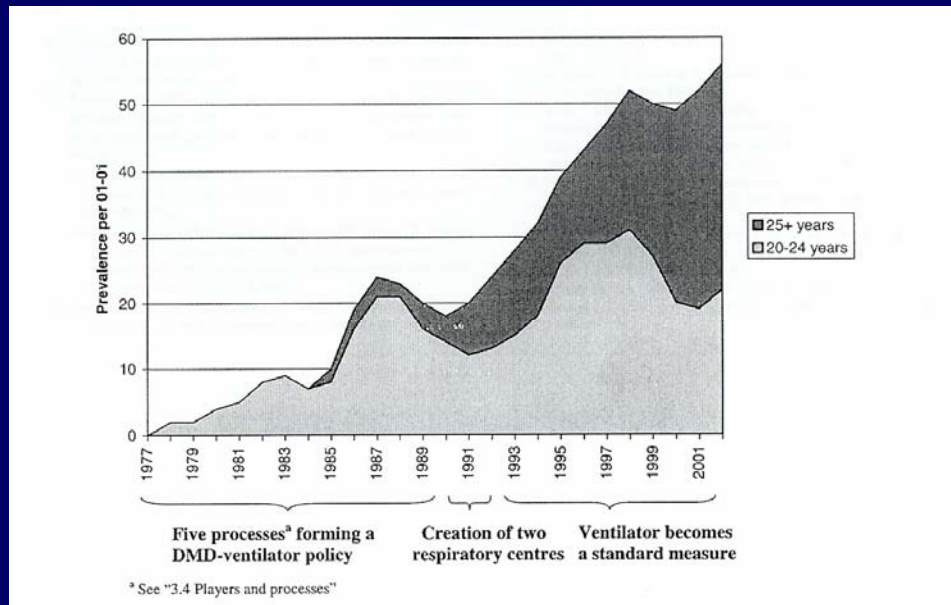


Fig. 4. Prevalence trends in relation to the introduction of ventilator use in the DMD population, absolute numbers Denmark 1977–2001. [a]See Section 3.4.

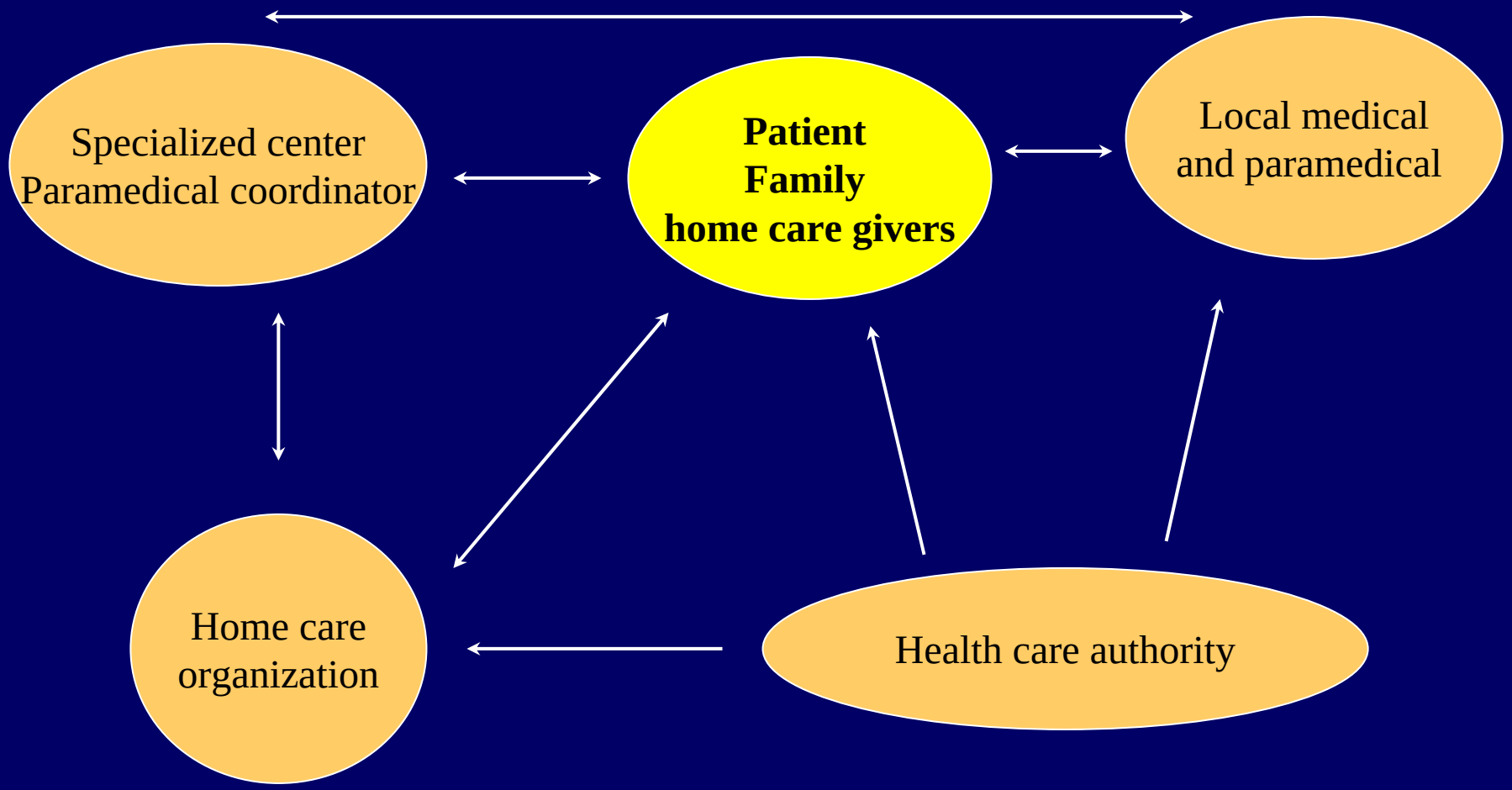
Ventilator users' Perspectives IVUN Survey (2000-2002)

- Patient point of view
  - Respiratory support: supplemental help like a Wheelchair
  - QOL doesn't change much
  - « I should have done it earlier »
  - Difficulties :
    - Ventilator noise and size (need more development based on input from users)
    - Health care professionals are not well informed
    - Funding and co-ordination of health care systems are insufficient

Goals of home mechanical ventilation

- Increase life expectancy
- Improve comfort and quality of life
- Facilitate independent living
- Give the tools to prevent hospitalization for acute respiratory problems (the worst place for a neuromuscular patient)

Returning and staying home: Participants



Home mechanical ventilation program : Key points

- Specialty centers hospital based
- Close relationship with health care authority
- Home ventilator maintenance program
  - Provide and maintenance of equipment
  - Patient, family and care givers information
  - Family physician and local paramedics information

Specialty centers

- Allows grouping of patients affected with rare diseases = larger numbers = greater expertise
- Provides a multidisciplinary approach
  - Diagnosis
  - Multiple impact of the disease and their interaction : Rehab physician, Orthopedic, pulmonary, nutrition, Cardiac, physical and psychological
- Provides a center to help coordinate the care with local medical caregivers i.e. family doctor, physiotherapist, palliative care,
  - Inform local practitioners about specific treatment options, indications and contra-indications
  - Reference center: maintain an on-call presence for questions and emergencies
- Provides regular follow-up
- Possible access to Critical care service and Step down pulmonary rehabilitation center

Close relationship with health care authority

- Regulation
- Legal aspects
  - Capability to transfer medical care to non professional care givers
- Financial aspects
  - Equipment
  - Home paramedical care
  - Care givers

Paramedical coordinator

- Paramedical from the referral center
- Improve relations with specialists
- Transfer and translate information to the local people (nurse, physiotherapist, family doctor, home care providers)
- Able to go home if necessary
- Prepare regular check-up
- Positive experience already for cystic fibrosis patients

Discharge from hospital to home

- Patient stability
- Adequate home environment
- Agreement from the health authority for payment or financial resources
- Home care organization
- Discharge planning

Problems of domiciliary ventilator dependency

- Patient are dependent on a machine:
  - Not a problem technically
- Patient will be more dependent on people
  - Rare
  - Expensive
  - Uncertain
  - Potential Conflict

Who supplies and maintains equipment ?

- Hospital
- Ventilator dealer
- Home care organization
  - Technicians
  - Paramedical staff

Equipment

- At least initially, NIV can be done efficiently with very simple device not too complicated or expensive i.e. simple bilevel device on ST mode
- Heated humidification if necessary
- O2 is usually unnecessary in NMD

Difficulty Doning and Doffing: Be creative

Complexity varies according to patient dependency

- 2 ventilators with internal and external batteries for dependant patients (> 12 h/day)
- Cough assist machine
- Tracheostomized patients need additional equipment, volume preset ventilator, 2 suction machines
- Wheelchair modification or adaptation to carry the ventilator

Alarms/Monitors

- Use depends on the level of motor and ventilator dependency
- Most important: Ventilator power alarm and disconnection
- Secondary Ventilation Alarms: Low pressure, expiratory tidal volume
- Additional Alarms:
  - Manual alarm to alert staff (smoke detector with head plate or sip and puff)
- Oxymeter/ EKG
  - Certainly for young children

Safe environment

- Continuous availability of electricity
  - Priority list with the electric company
  - Emergency compressor
- Emergency call system 24 hours hotline to home care organization and to respiratory physician from the specialized center
- Resuscitation bag is a very simple but major piece of equipment for ventilated patients, it should be provided to any NM ventilated patient

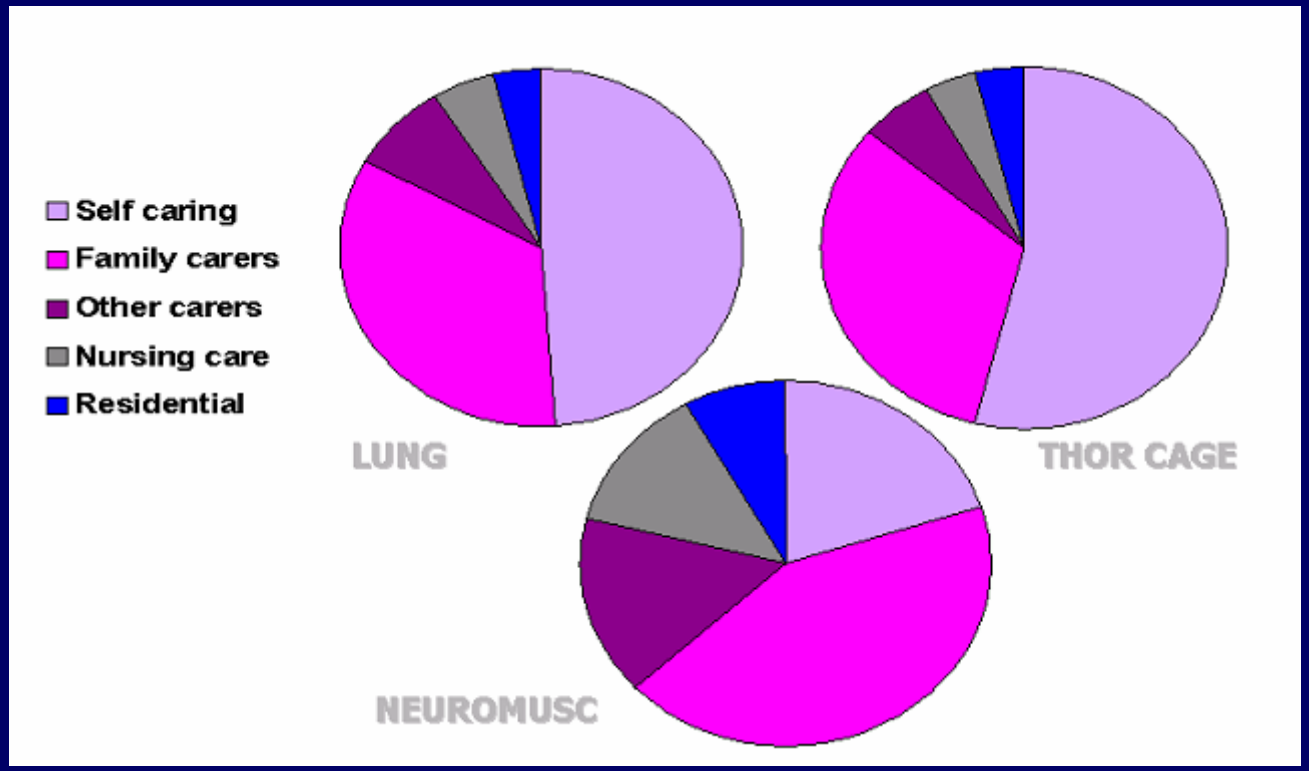
Resuscitation bag

and proper connecting tubes ALWAYS with the patient

Home supervision

- Regular visits according to the device and daily usage
- Emergency visits : 24h/24
- Patient / equipment interaction
- Patient and caregivers knowledgable about equipment
- Transmission of information to the physician who prescribed ventilation relative to
  - Equipment and
  - Patient using the equipment

Home mechanical ventilation in Europe: Caregivers



Data from the European survey: Eurovent Europ Resp J ; 2005, 25: 1025-1031

Human environment for ventilator dependent patient

- Best situation but rare: patient is the boss
  - Recruit, teach, pay the helpers
  - Scandinavian countries (6-7 full time non professional care givers are employed)
- Not so bad: patients group themselves in a community
- Least Desirable (the more frequent & the more difficult for long term): Family in charge
  - Family, parents, wife or husband change position they become medical professionals
    - Frequently unable to disassociate both positions
    - Refuse help from outside (not as good)
    - Anxiety for the future
  - Having a back-up plan is very important but rarely done ahead of time
    - Temporary rest or in case of emergencies…

Training of patient and care givers

- Patient training is always suitable but not always possible (very young children, some NM unable to cooperate)
- Family and care givers
- Training about:
  - Equipment : Machine, settings, resuscitation bag
  - Assisted cough and hyperinflation techniques
  - Emergency situations:
    - Power and device failure
    - Ability to recognize signs and symptoms requiring assistance
    - Advance directive in case of emergency

Regular evaluation of patient and helpers capabilities

- Capability to
  - Recognize signs and symptoms requiring assistance
- Capability to teach helpers to react adequately in an emergency situation
- Capability to manage his own equipment

Conclusion: Preventative attitudes

- Medical Team:
  - Objectively inform patient and/or family about
    - Potential disease risks
    - Need for regular follow-up
    - How to prevent worsening
    - Abnormal clinical signs
    - Treatment options
  - Listen carefully to the patient and family
    - Remember that they live daily with their disease and therefore become specialists especially about changes that may occur in their medical condition
- Patient and family:
  - Participate in decisions that will need to be made:
    - Accept periodic assessment
  - Be knowledgeable about the
    - Risk periods: infection, surgery
    - Main clinical signs indicating the need for emergency consultation
  - Be prepared: always have the following on hand
    - A summary of the risks related to the disease
    - Personal and pertinent medical data
    - Suggested referral physicians
    - Adequate and necessary equipment
    - Advance directives in case of emergency

Conclusion

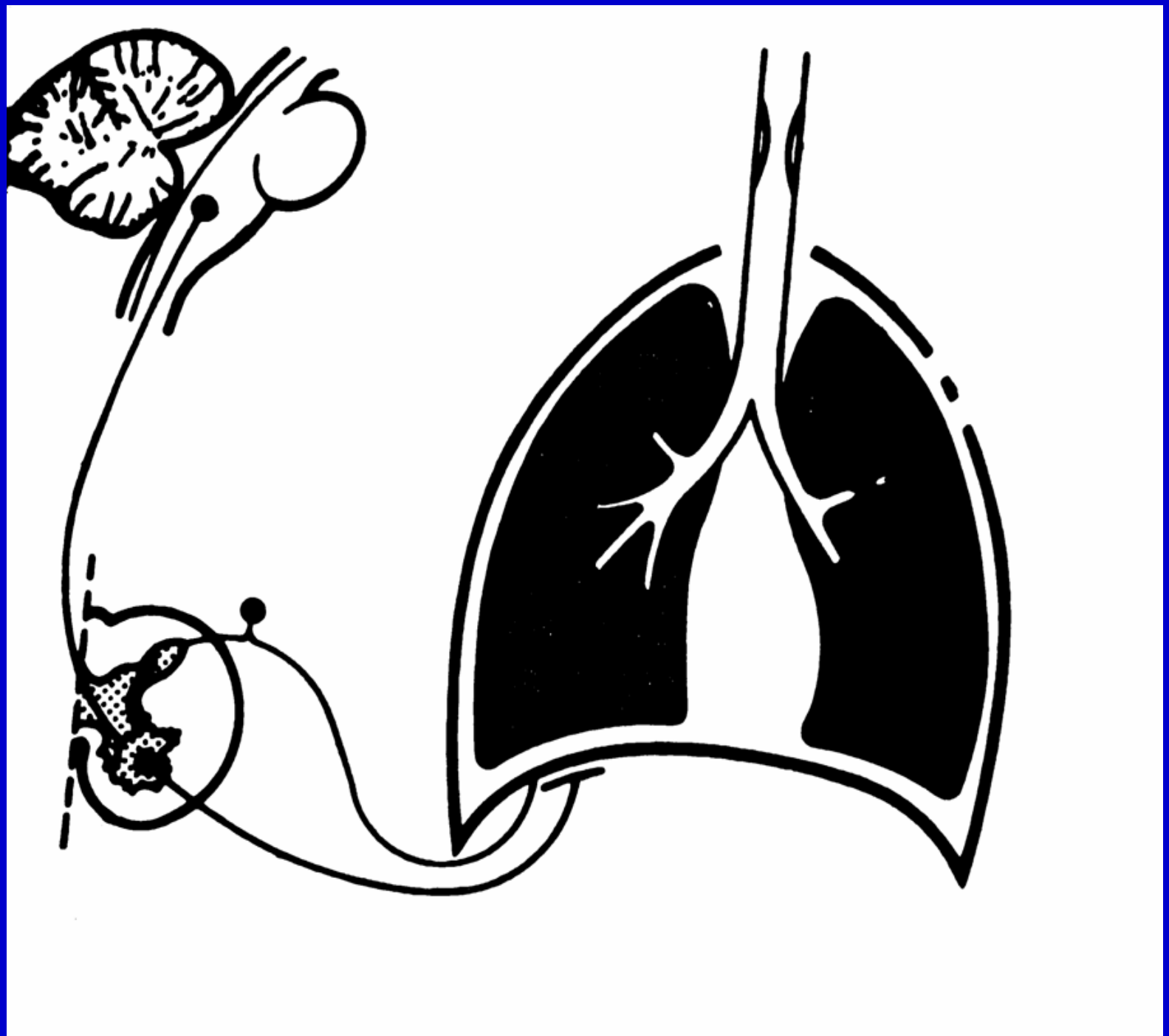
- HMV is feasable and relatively simple but:
  - Requires organization
  - Knowledge
  - Understanding of the medical team and Health Care Authority
  - Commitment

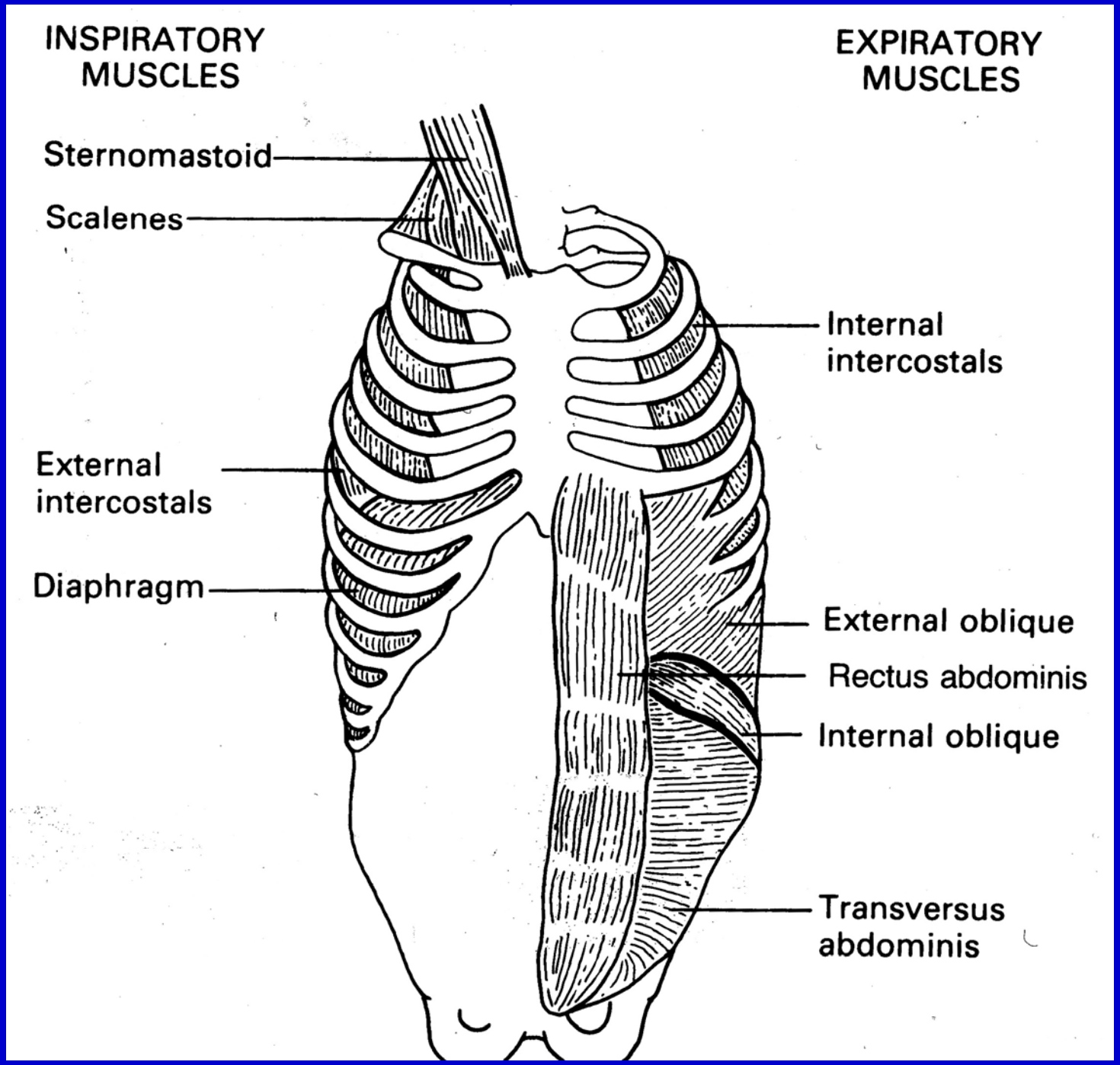
Understanding respiratory involvement in chronic neuropathic or myopathic diseases treated with long term mechanical ventilation

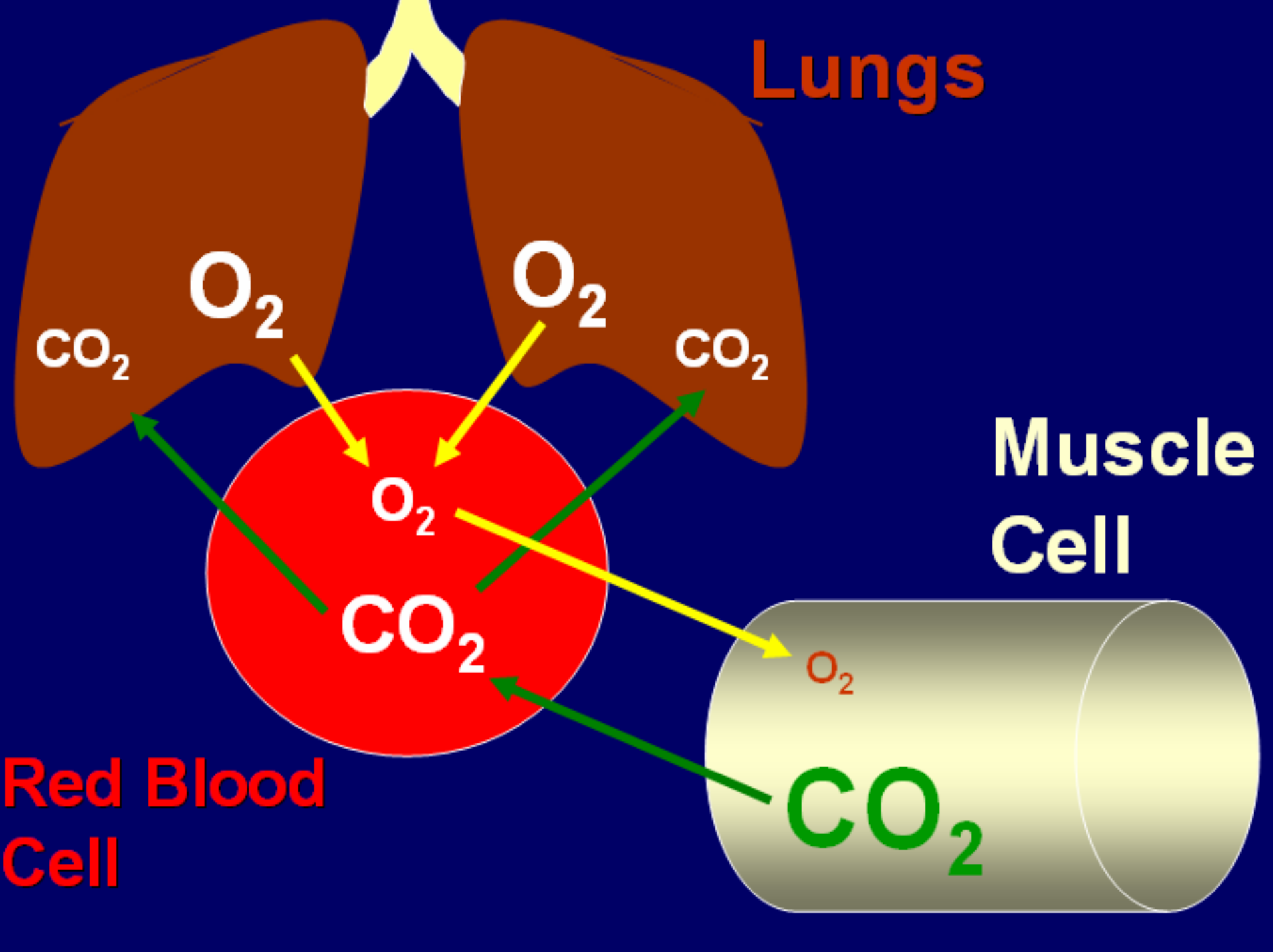
Diseases
Pathophysiology
Methods
Indications
Results

Breathing in normal individuals

- Respiratory control by central respiratory centers (brainstem)
- Conduction through the nerves (phrenic)
- Muscles contraction of the respiratory muscles (diaphragm)
- Expansion of the thoracic cage (thorax and spine)
- Gaz exchanges (lung)




INSPIRATORY MUSCLES
EXPIRATORY MUSCLES
Sternomastoid
Scalenes
Internal intercostals
External intercostals
Diaphragm
External oblique
Rectus abdominis
Internal oblique
Transversus abdominis


Lungs
O_2
O_2
CO_2
CO_2
O_2
CO_2
Muscle Cell
O_2
CO_2
Red Blood Cell

Main diseases severely affecting ventilatory function

- Neuropathic :
  - Spinal injury
  - Motor neuron disease :
    - Polio and post polio
    - Spinal muscular atrophy (type 1, 2, 3)
    - Amyotrophic lateral sclerosis
  - Peripheral neuropathy
- Myopathic :
  - Dystrophin deficit : Duchenne (early) and Becker (late)
  - Myotonic myopathy (Steinert)
  - Congenital muscular dystrophy

Breathing in neuro-muscular diseases

1. Inspiratory muscles weakness :
   - Impaired inspiration + impaired cough
2. Expiratory muscles weakness :
   - Impaired cough
3. Thoraco-vertebral deformities :
   - Volume restriction, ↓ compliance
4. Swallowing difficulties :
   - Aspiration pneumonia, tracheo-bronchial accumulation of secretions
   - Denutrition

Decrease of inspiratory capability

- Inspiratory muscle weakness :
  - In rest muscle condition, the maximal pressure developed by a maximum inspiratory effort is decreased related to normal
- To avoid muscle fatigue or muscle exhaustion the pressure developed at each breath must remain below a certain percentage of the maximum pressure (Ti/Ttot x Pinsp < 15 % of Pimax)
- When insufficient pressure is non sustainable a strategy to insure ventilation is automatically adopted

Strategy to sustain ventilation

1. Change the breathing pattern
   - Decrease the VT
   - Increase the frequency to maintain minute ventilation
   - But this is limited by the cost of the unusefullness in term of gaz exchange of the ventilation of the dead space at each breath
2. Tolerate, until a certain extent, hypoxemia and hypercarbia

Ventilatory failure = alveolar hypoventilation : ABG's : ↑PaCO2, ↓ PaO2

- Usually characterized by a progressive worsening
- First step : nocturnal hypoventilation
  - Related to supine position
  - Related to sleep induced hypoventilation :
    - Mainly during REM sleep particularely when diapragmatic weakness exists
  - Sometimes worsened by obstructive apnea
  - Easily diagnosed by nocturnal pulse oxymetry
- Second step : permanent (night and day)

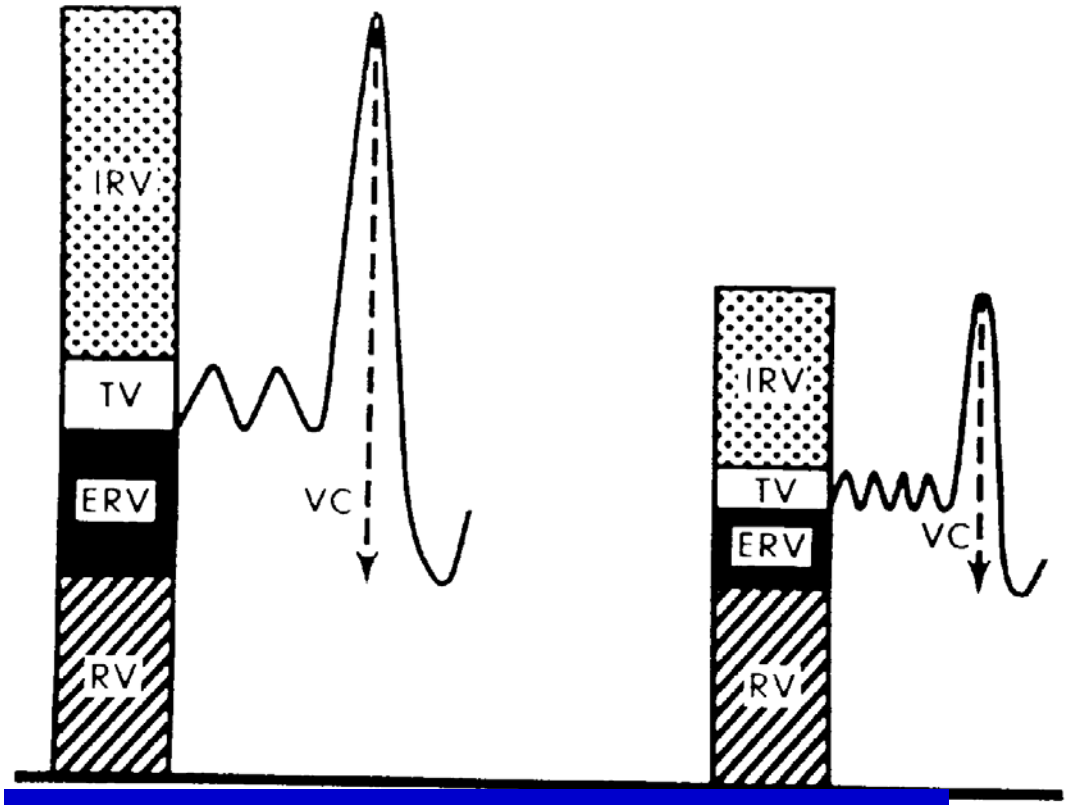
Clinical symptoms

1. Nocturnal
   - Air hunger, chocking, snoring, insomnia, cyanosis (SpO2 fall), headaches
2. Immediately after sleep
   - Headaches, drowsiness, secretion retention, restlessness
3. Daytime
   - Fatigue, depression, impaired cognition, lost of energy
   - Dyspnea, ankle oedema, weight loss

Useful measurements

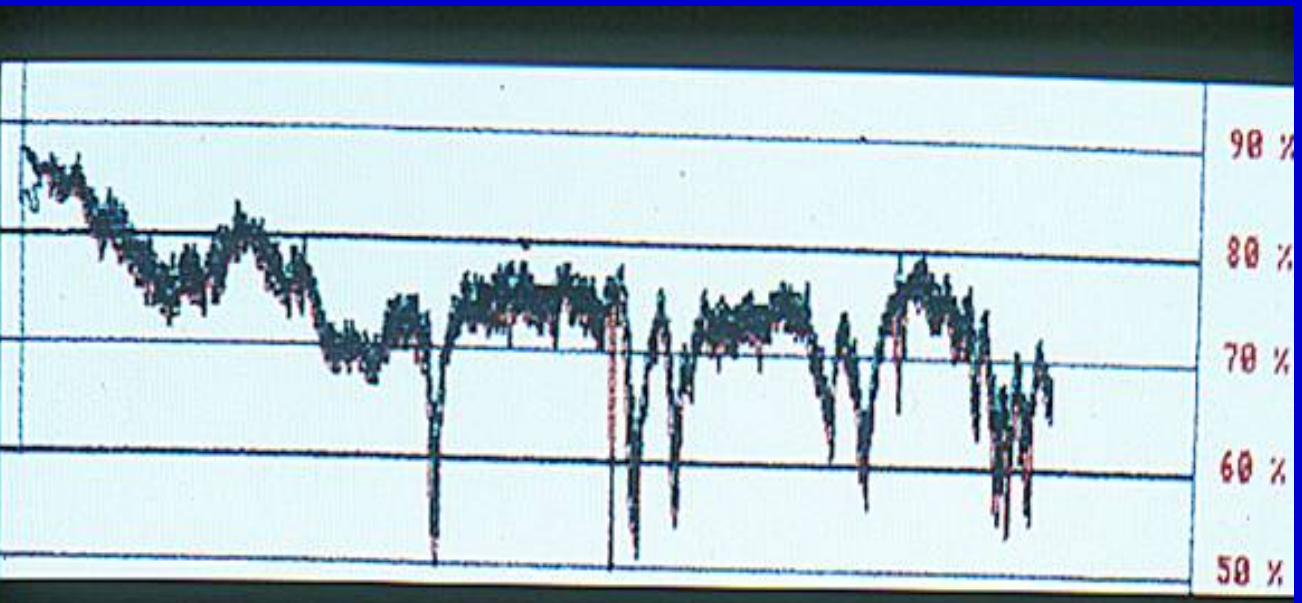
Measurement	Threshold for "alarm"
– Vital Capacity	– < 50% theoritical
– PImax , PEmax	– < 30% theoritical
– ABG's when • VC < 50% • PImax < 30%	– PaCO2 > 45mmHg
– Nocturnal pulse oxymetry	– Episode(s) of sustained desaturation < 88% for more than 5 minutes

normal **restrictive**

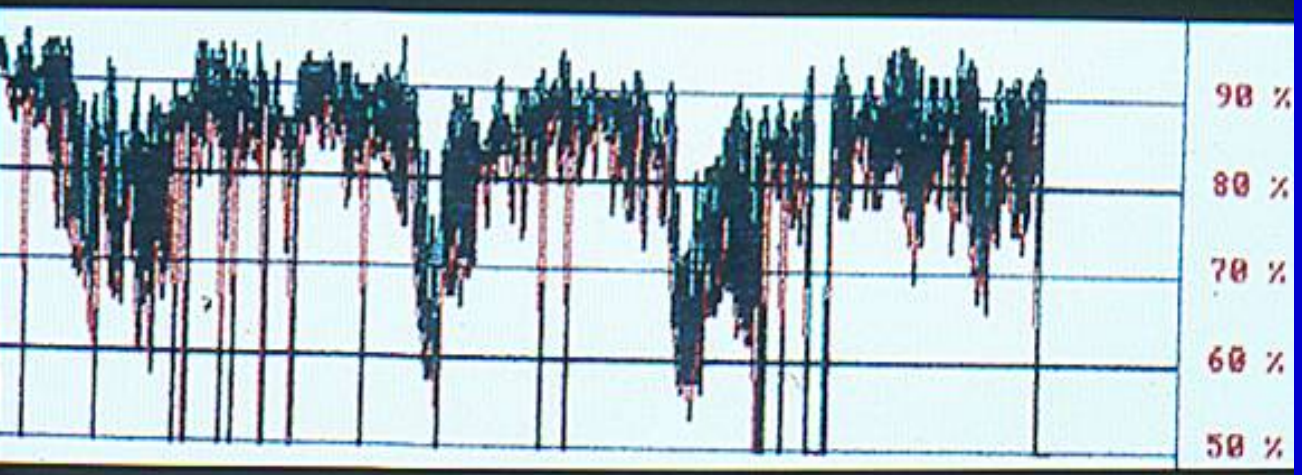


Spirometry

Overnight saturation : NREM & REM desaturation



NREM : << 90% + many minor dips (# 3-5%)
REM : profound desaturation



•NREM : < 90% + many deep dips (# 10%)
•REM : profound
•desaturation

Principles of the treatment

1. Try to prevent thoraco-vertebral deformities
2. Compensate respiratory insufficiency by
    1. Mechanical inspiratory assistance
    2. Cough assistance

Principles of mechanical inspiratory assistance

- Provide totally or partly the inspiratory volume up to "normal value"
- With a machine (ventilator) which does the totality or a part of the work of breathing

In order to insure normal alveolar ventilation

Mechanical ventilator delivering Intermittent Positive Pressure Ventilation

- Interface
  - Non-invasive
  - Tracheostomy
- Mode
  - Volume targeted
  - Pressure targeted

- Note : Negative pressure ventilation used in the past are now obsolete
  - Iron lung, poncho, cuirass
  - Abdominal belt
  - Rocking bed

Principle of cough assistance

- Non Invasive :
  - Increase the maximum expiratory flow
- Invasive : tracheal suctioning
    - Transglottis catheter
    - Tracheostomy catheter

ORGANIZATION

Non Invasive cough assistance aims to increase maximum expiratory flow

1. Use the elastic recoil pressure of the respiratory system by mechanically filling the lung close to its maximum volume by using hand balloon or ventilator
   - Increased VT
   - Accumulated VT (stacking)
2. Apply manual external positive pressure during expiration
3. Apply high positive pressure (forced inspiration) followed by low negative pressure (forced expiration) : in-exsuflator machine

"Tin- Can IN- Exsuffator"
JH EMERSON, 1993

"IN-Exufflator", JH EMERSON, 1996

"Cough Assist", JH EMERSON, 2001

Indications for long term mechanical ventilation : timing

- Once daily hypoventilation exists ($PaCO_2$ > 45 mmHg)
- The natural history of the concerned disease may influence the time for the decision :
  - Earlier in rapidly progressive (ALS)
  - Later in slowly progressive (Becker)

Indications for long term mechanical ventilation : method

- Non-invasive nasal ventilation is undoubtedly the method of choice
- Invasive tracheostomy may remain indicated in case of :
  - Continuous need of mechanical ventilation

    and / or
  - Frequent tracheal aspiration in spite of the suppression of oral feeding (replaced by enteral artificial nutrition)

Results

- Under mechanical ventilation alveolar ventilation is always normalized
- When it remains some ability to spontaneously breathe (depending on the residual muscle ability) daily time without mechanical assistance is possible allowing to reach a good long term stability with mainly nocturnal ventilation more or less completed with additional hours during the day

Results : extension of survival is the main results

- Reaching normal expectancy in stable deficit without other organ disorder : sequels of polio
- Substancially extended but shortened compared to normal according to associated organ or function disorders :
  - Cardiac in Duchenne or myotonic myopathy
  - Swallowing in ALS
  - Neurovegatative in spinal injury

Associated results

- Regression of right heart failure related to chronic alveolar hypoventilation
- Decrease of hospitalization down to # 0
- Improve of vital energy
- Increase of the quality of life

Important Reminder

- Neuro-muscular individuals are always paralyzed and consequently require continuous or quasi continuous personal attendance to provide daily needs for life besides mechanical ventilation
- This may heavily burden the load on family members or mandate continuous hospitalization when available
- And even sometimes become difficult or impossible when paid attendants must be continuously present

Conclusion

1. Long term mechanical ventilation is extremely efficient
2. Non invasive option is now preferred
3. Tracheostomy may be indicated in late evolution
4. Associated organ dysfunctions greatly influence the long term prognosis
5. Personal attendancies are necessary and may limit the use of long term mechanical ventilation

EAMDA - meeting
ZAGREB 23 september 2005

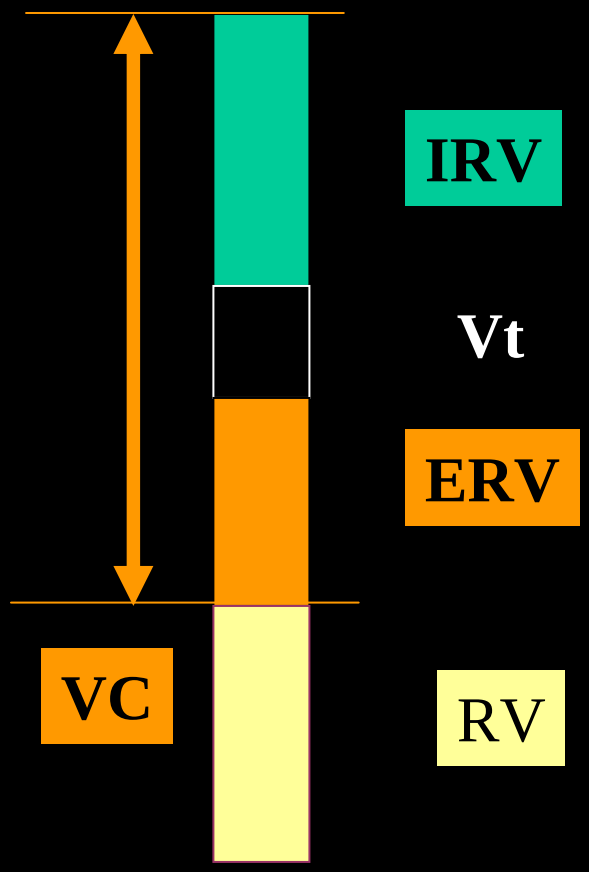
philippe . soudon @ inkendaal.be

How to live without respiratory capacity and without tracheostomy

How to live with a neuromuscular disease without respiratory capacity and without tracheostomy

ZH Inkendaal – Vlezenbeek - Brussels

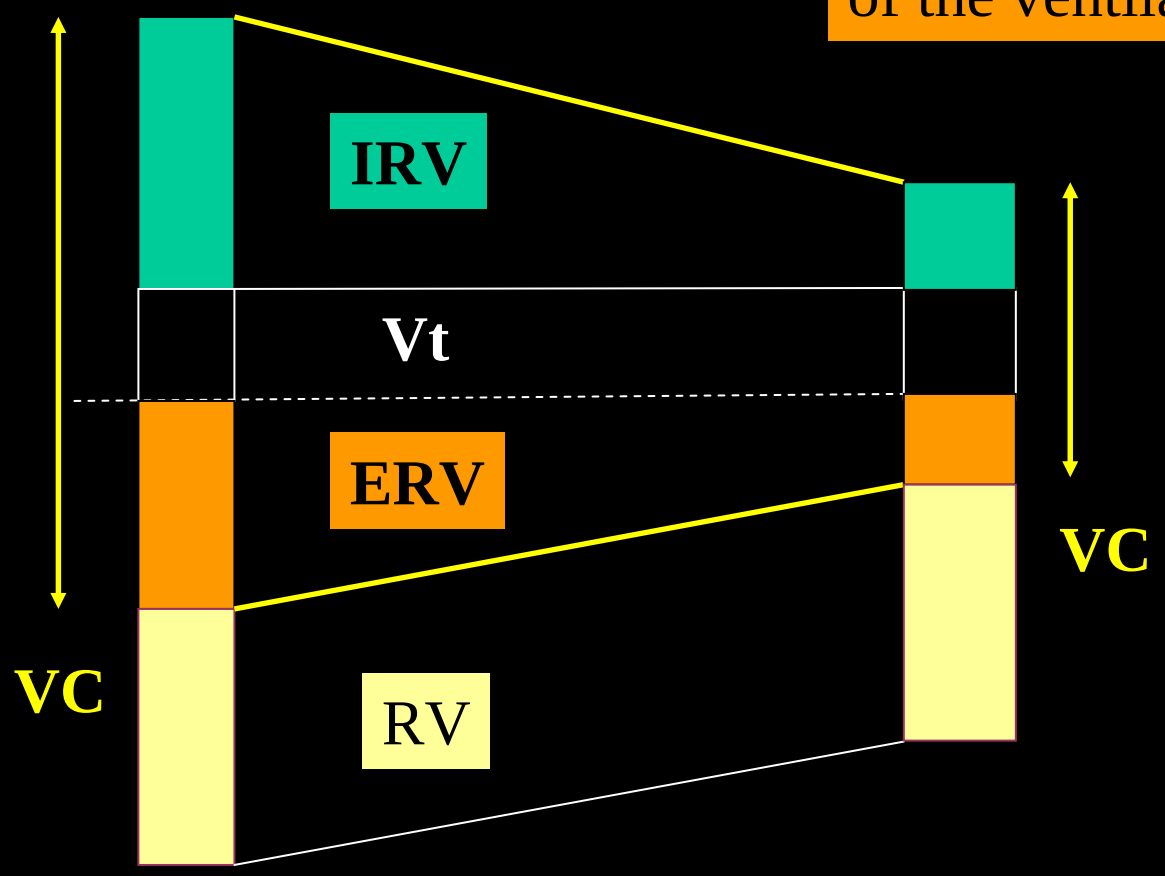
philippe . soudon @ inkendaal . be



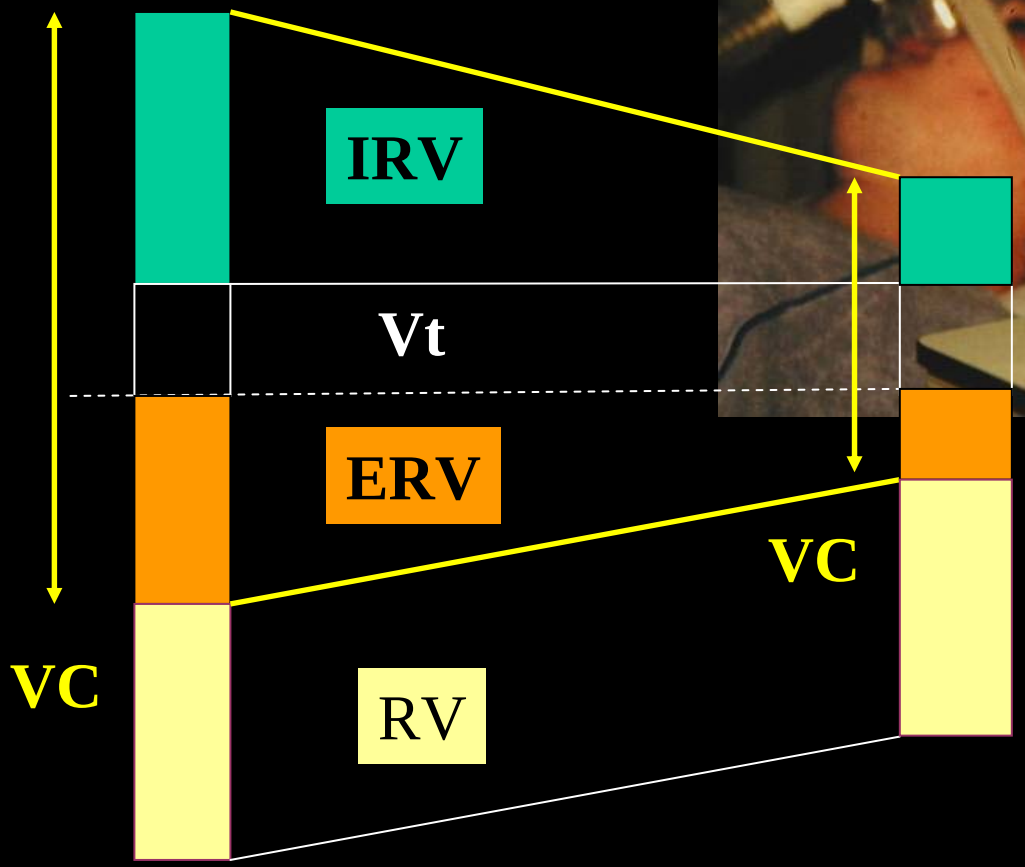
Ventilation is a consequence of the muscular activity of diaphragm and chestwall

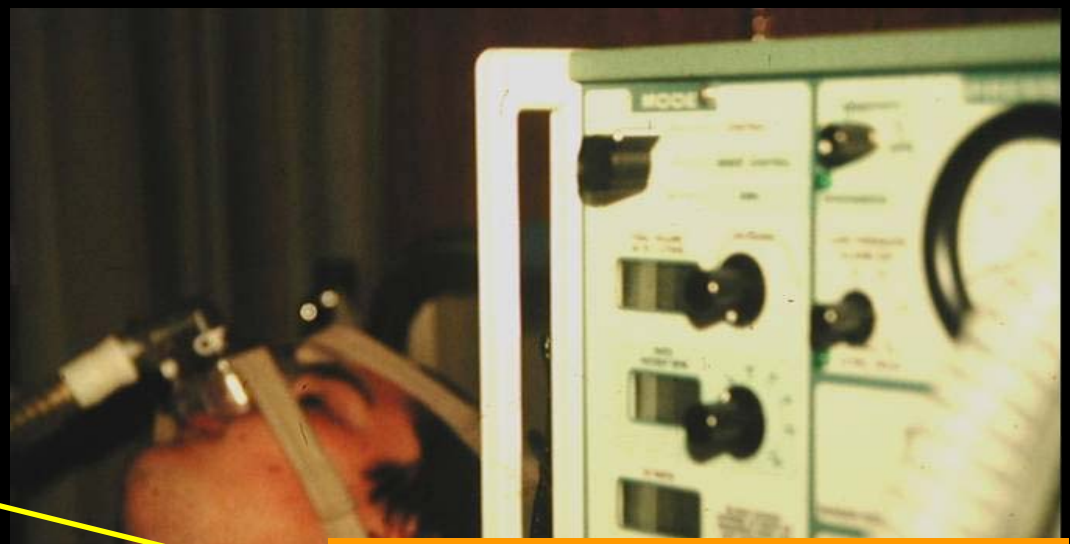
muscular dystrophies

with time,
specially in muscular dystrophies,
there is a spontaneous evolution
to a progressive restriction
of the ventilated volumes



muscular dystrophies



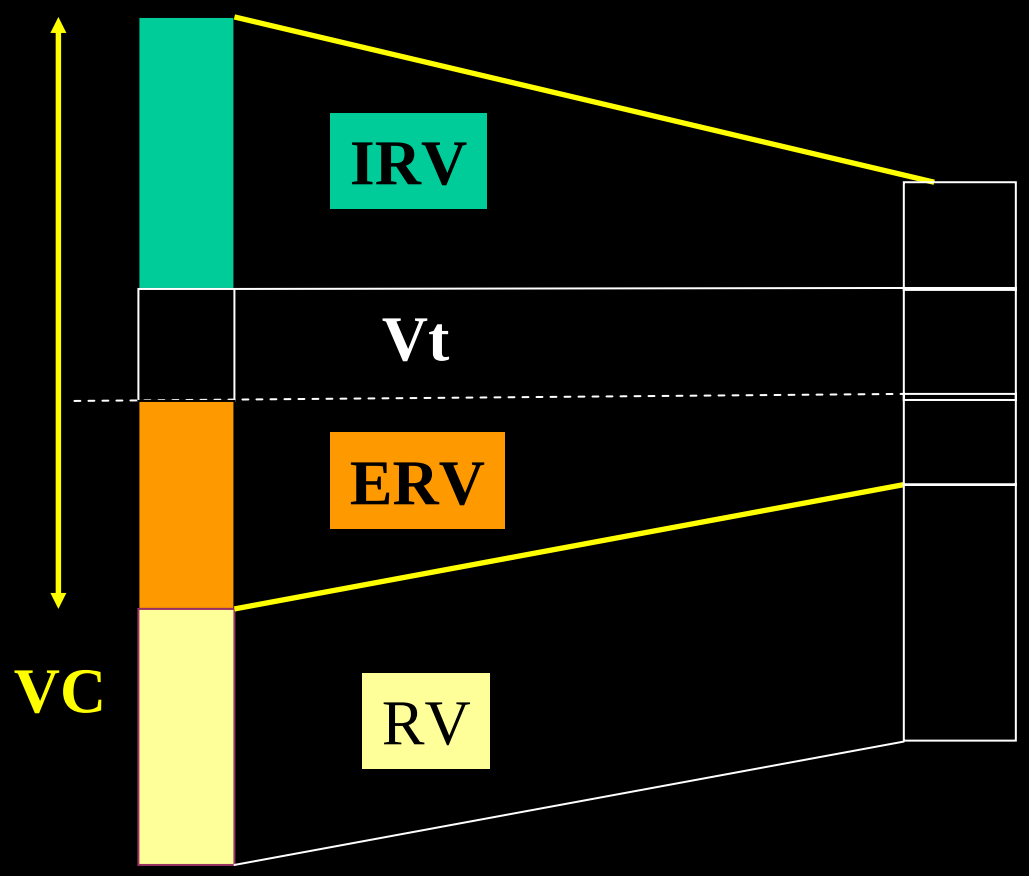


to hold the same ventilation,
reduced VC is then compensated
by a higher frequency;
but the price to pay
is **hypercapnia**,
and this leads to the need
for <u>assisted</u> ventilation
(first) during the night

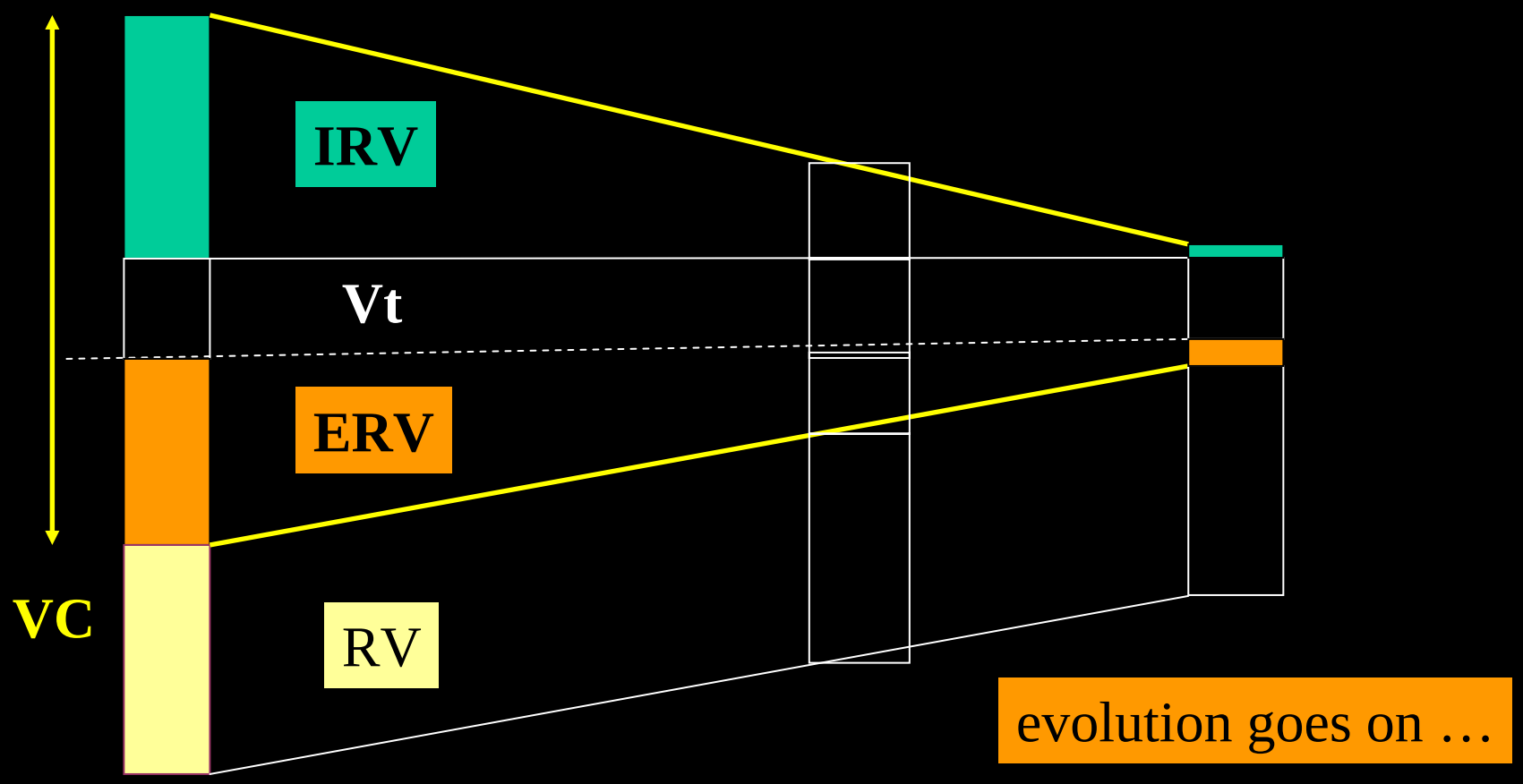
NOCTURNAL VENTILATION
NASAL MASK

- used from 1981 (Yves Rideau, F-Poitiers)
- avoids specific complications of trach.
- sometimes needs to alternate different types of mask (customized)
- fits for nocturnal but also discontinuous (night + afternoon nap) ventilation

muscular dystrophies

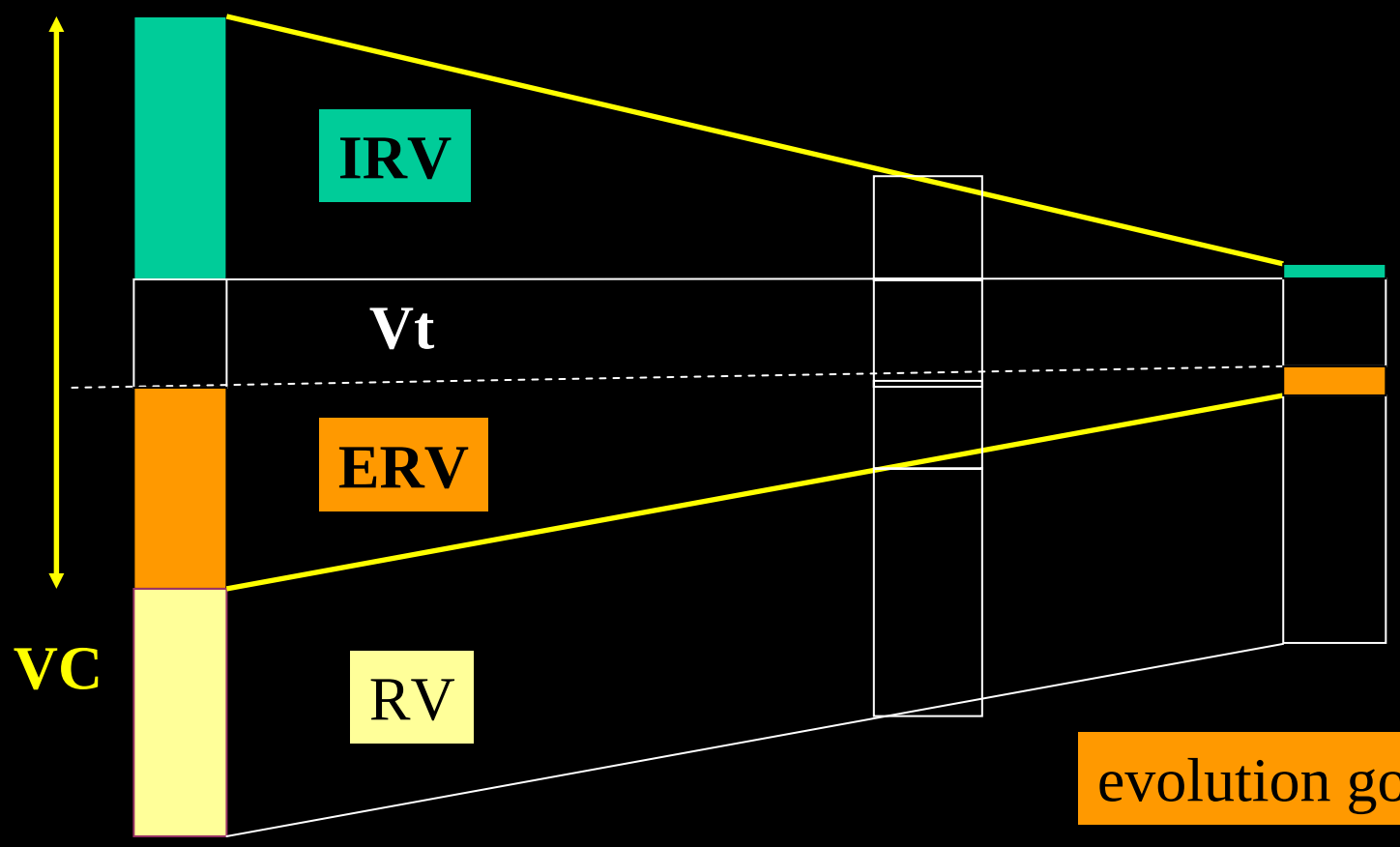


muscular dystrophies



muscular dystrophies

+ swallowing tr / aspirations

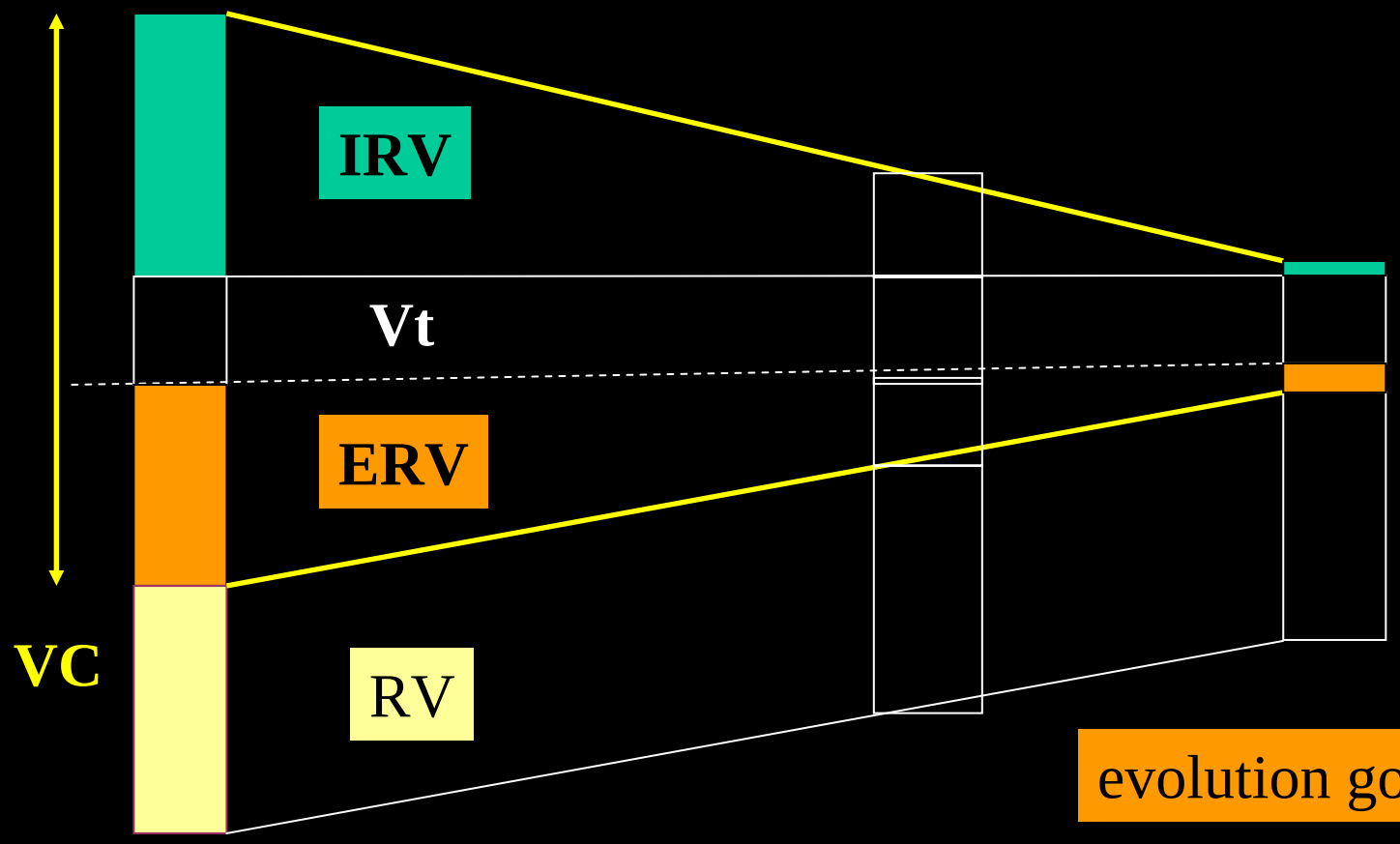


evolution goes on …

muscular dystrophies

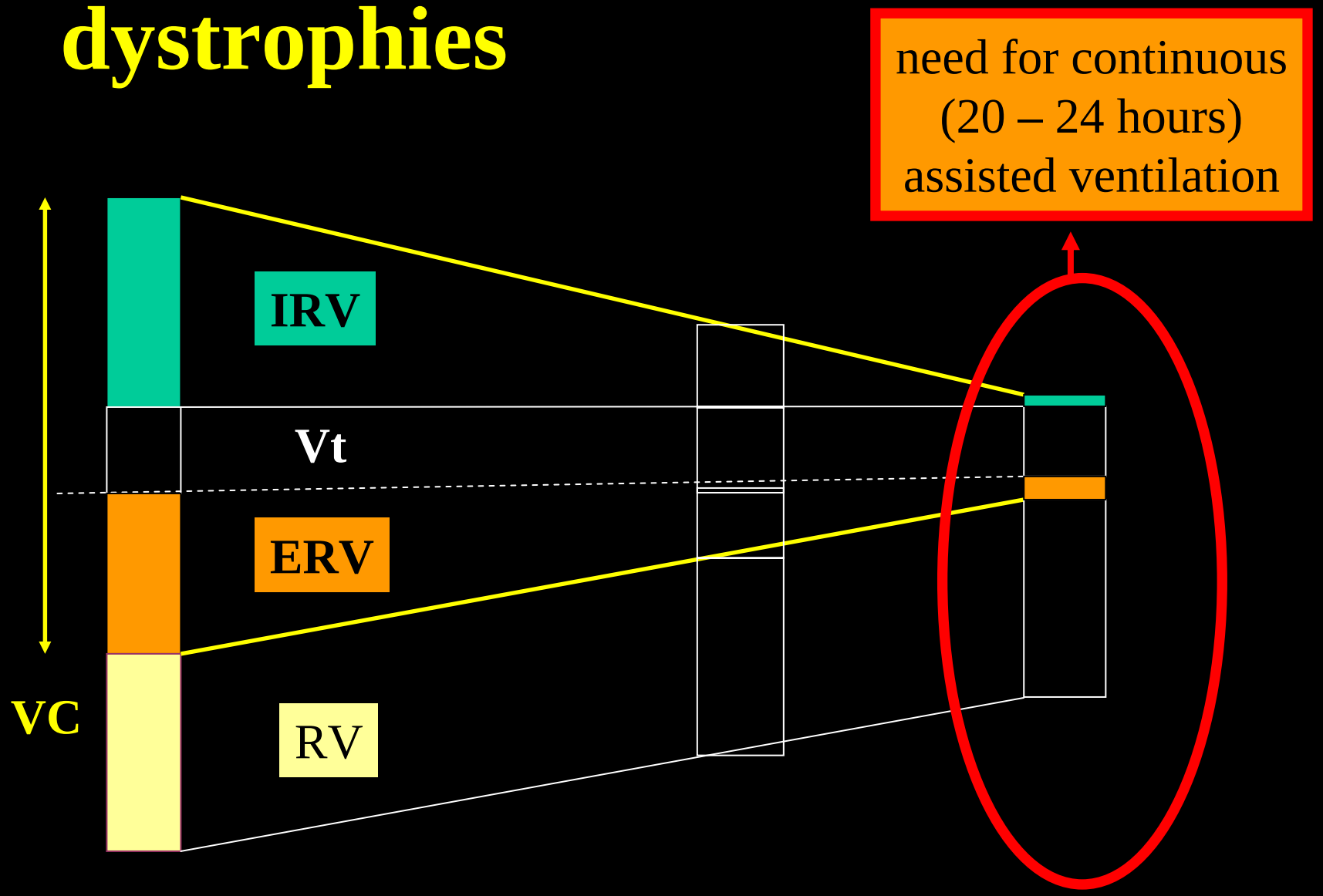
+ swallowing tr / aspirations

+ (very) bad secretion clearing



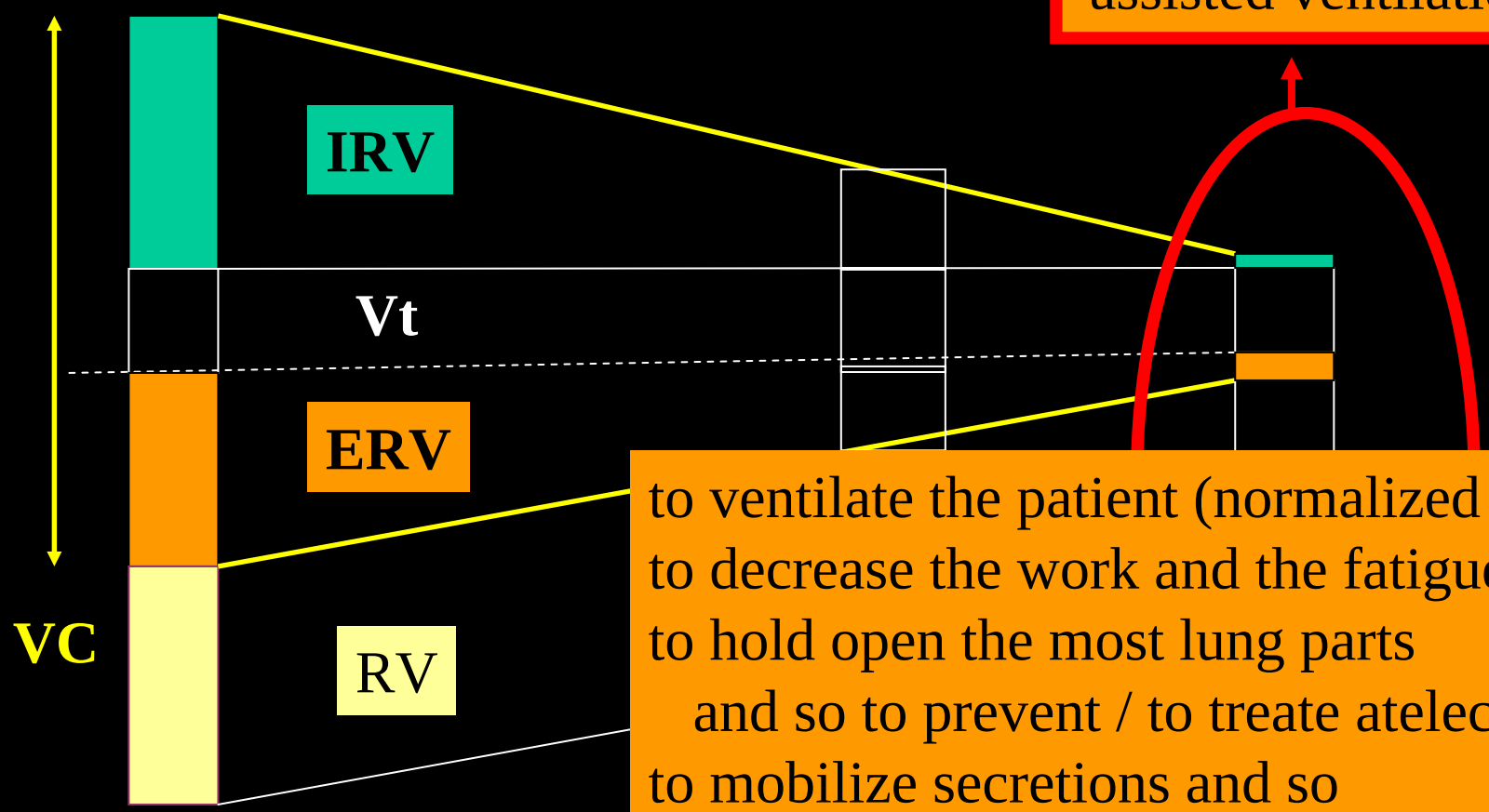
evolution goes on …

muscular dystrophies



muscular dystrophies

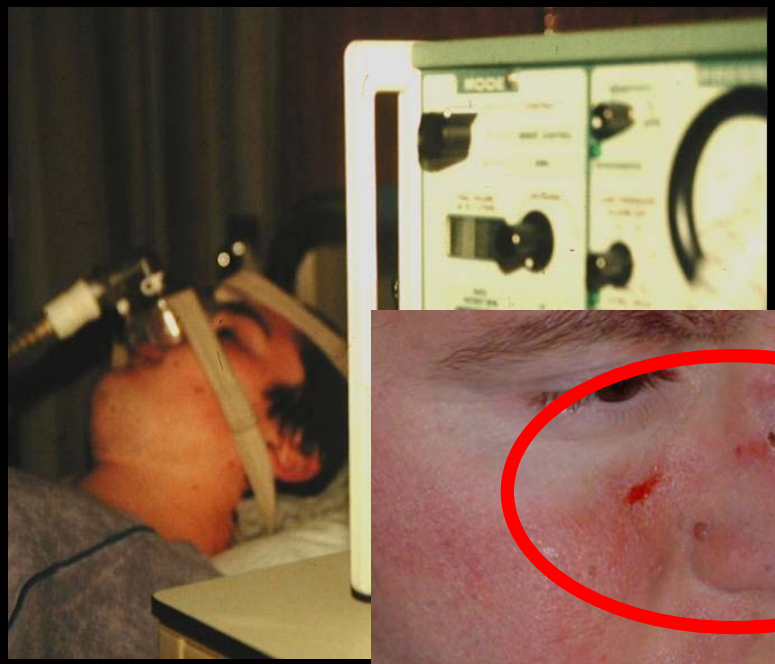
need for continuous
(20 – 24 hours)
assisted ventilation



to ventilate the patient (normalized CO2)
to decrease the work and the fatigue
to hold open the most lung parts
and so to prevent / to treate atelectasiae
to mobilize secretions and so
to prevent hypersecretion / surinfection

VENTILATION in the DAYTIME

noninvasive interfaces



problem:

<u>nasal mask does not fit for 24 hours use:</u>

- no binocular vision
- <u>skin irritation or necrosis</u>
- discomfort, weight on the face
- psychological (appearance)
- difficulty in speaking (risk of desynchronization and so risk of aerogastria).

VENTILATION in the DAYTIME

noninvasive interfaces

tracheostomy

- « first choice » in the world
- cuffless, + speaking valve
- allows (easy) tracheal suction
- but some complications …

some complications of **tracheostomy**

- chronic hypersecretion
- insufficient voice
- chronic infection
- granuloma, bleeding
- tracheal stenosis
- thoracic pain
- **social handicap**
- etc ...

VENTILATION in the DAYTIME
noninvasive interfaces

other possible solution:

VENTILATION in the DAYTIME
noninvasive interfaces

other possible solution:

during the night,
patient keeps using nasal mask

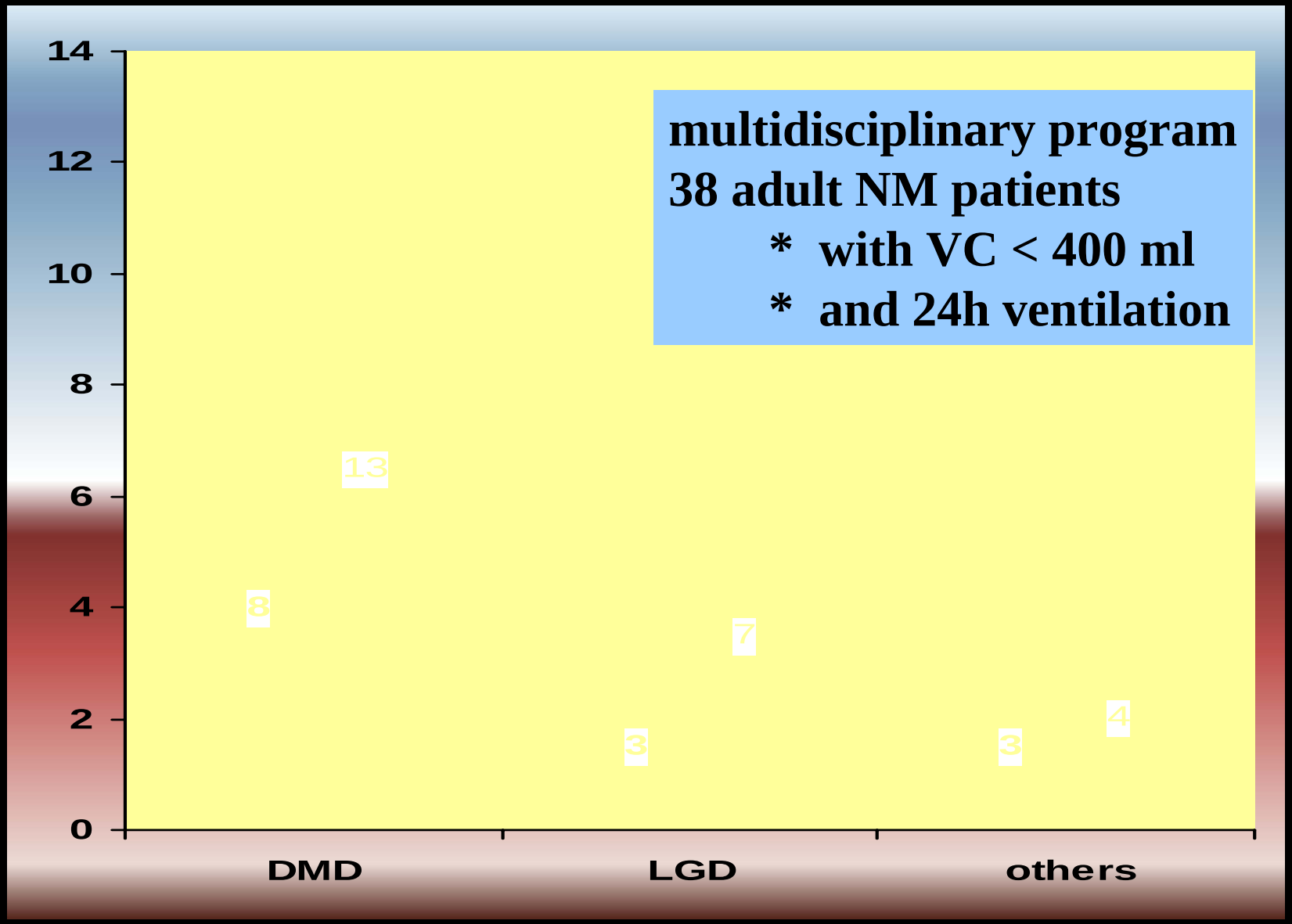
VENTILATION in the DAYTIME
noninvasive interfaces

other possible solution:

during the night,
patient keeps using nasal mask

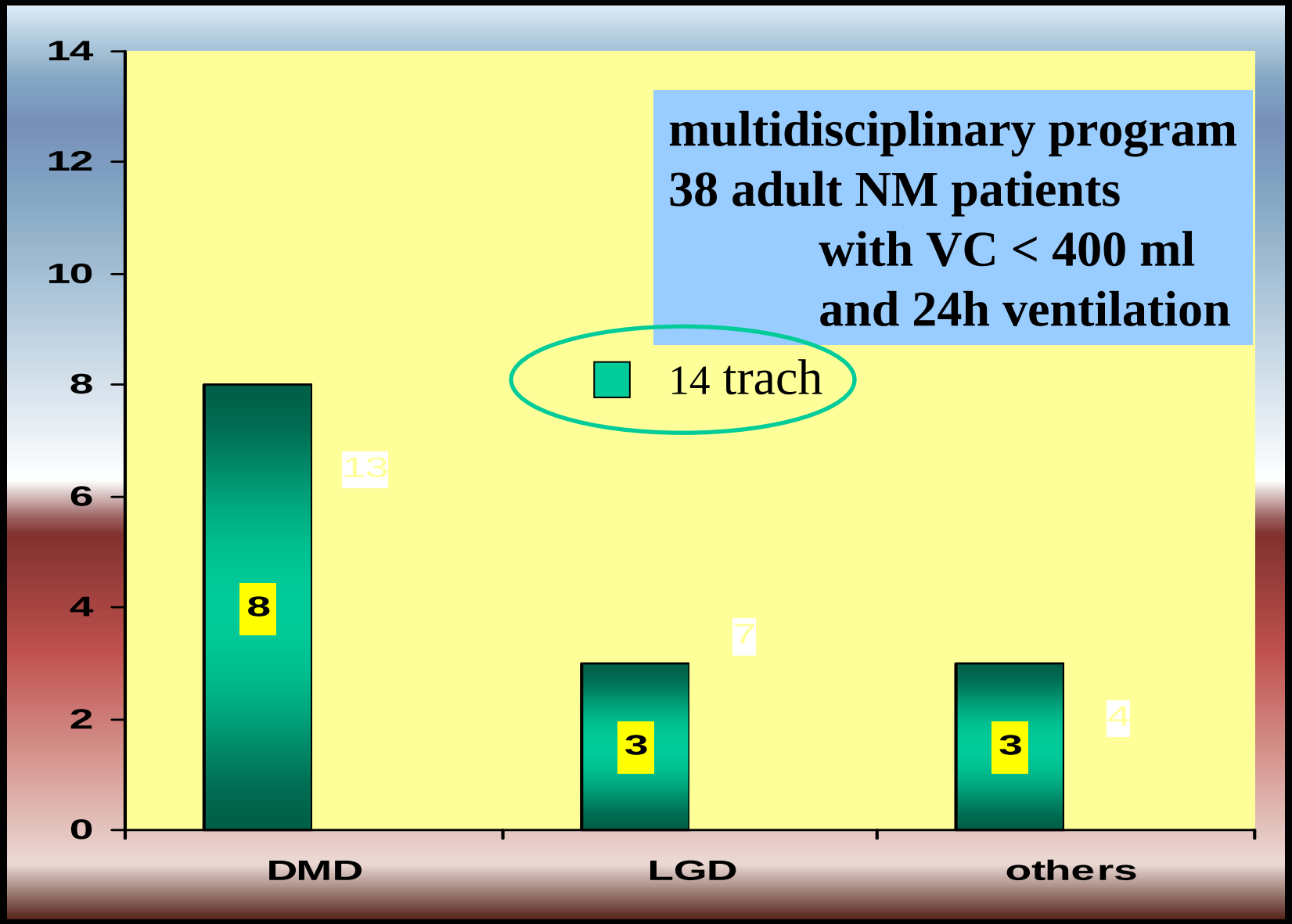
in the daytime,
he’using a mouthpiece

muscular dystrophies with VC < 400 ml

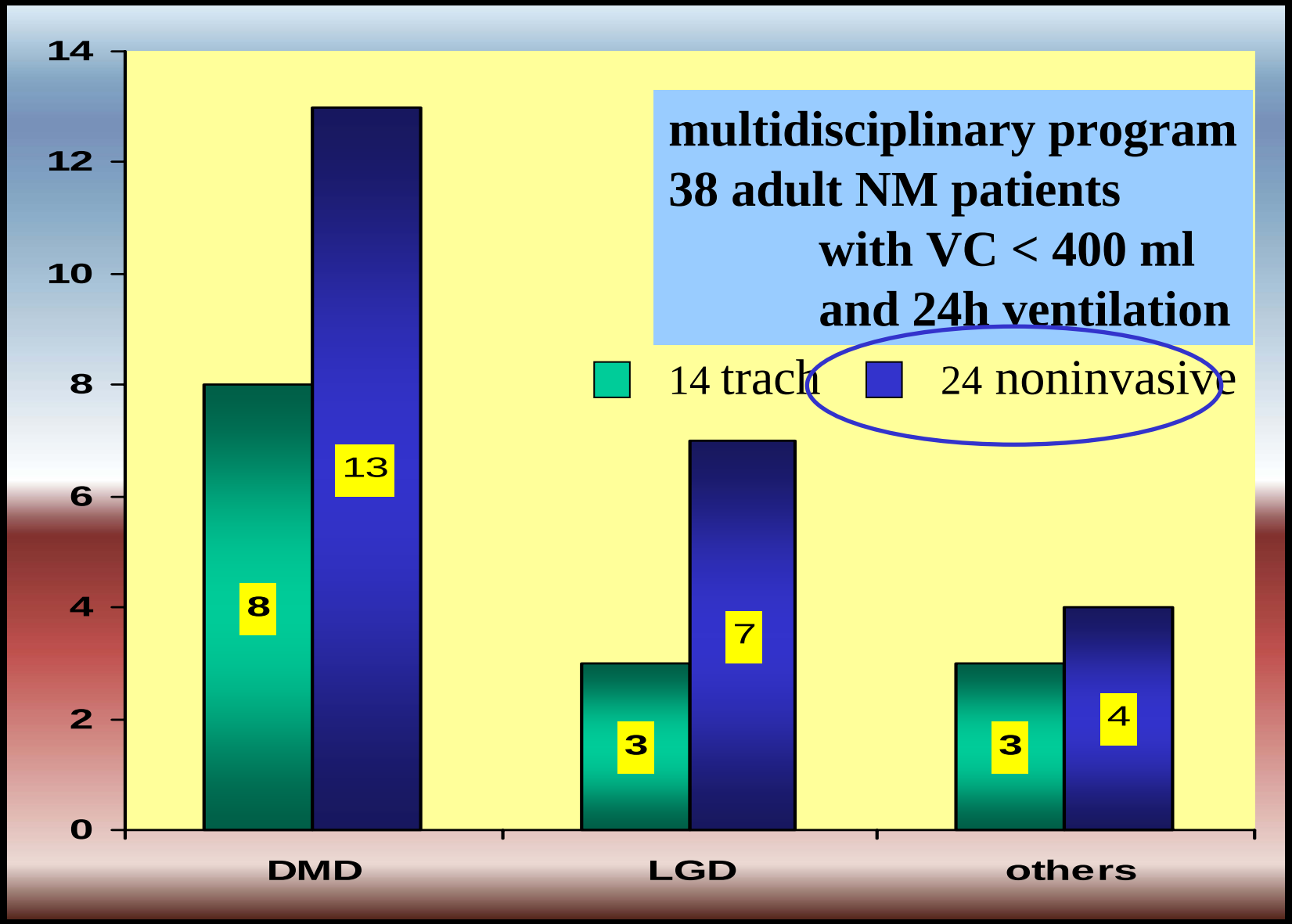


multidisciplinary program
38 adult NM patients
- **with VC < 400 ml**
- **and 24h ventilation**

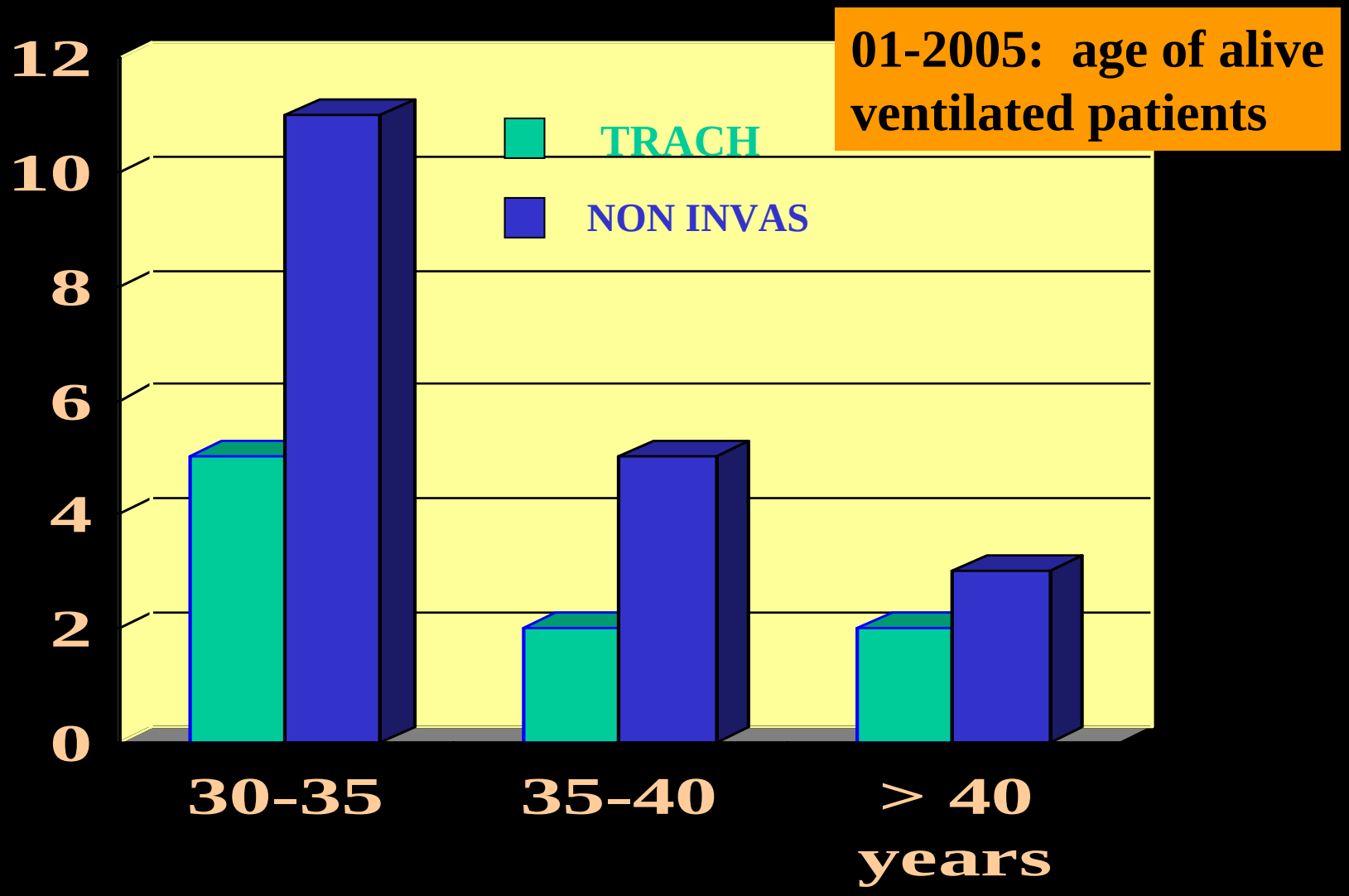
muscular dystrophies with VC < 400 ml



muscular dystrophies with VC < 400 ml

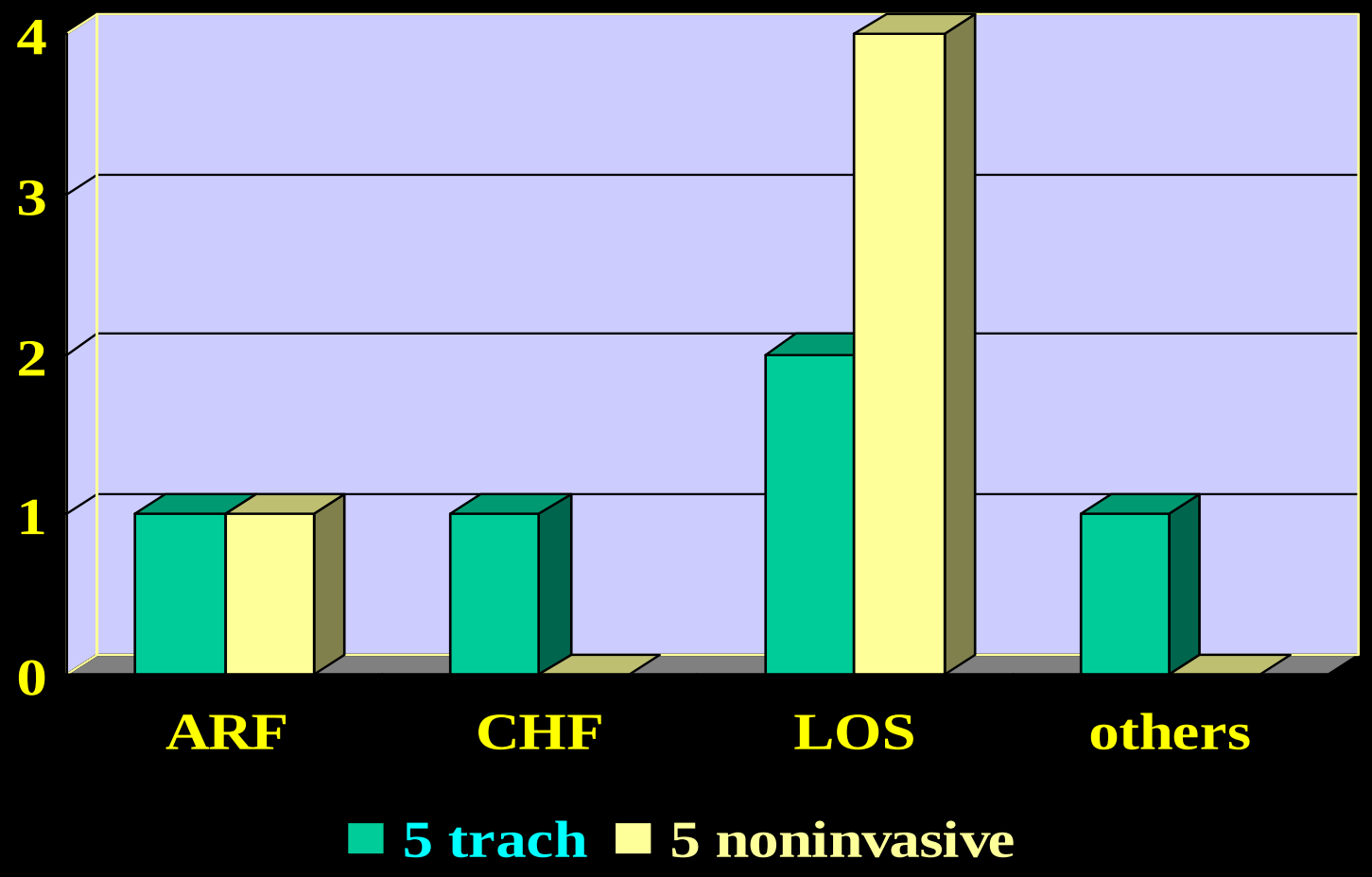


muscular dystrophies with VC < 400 ml

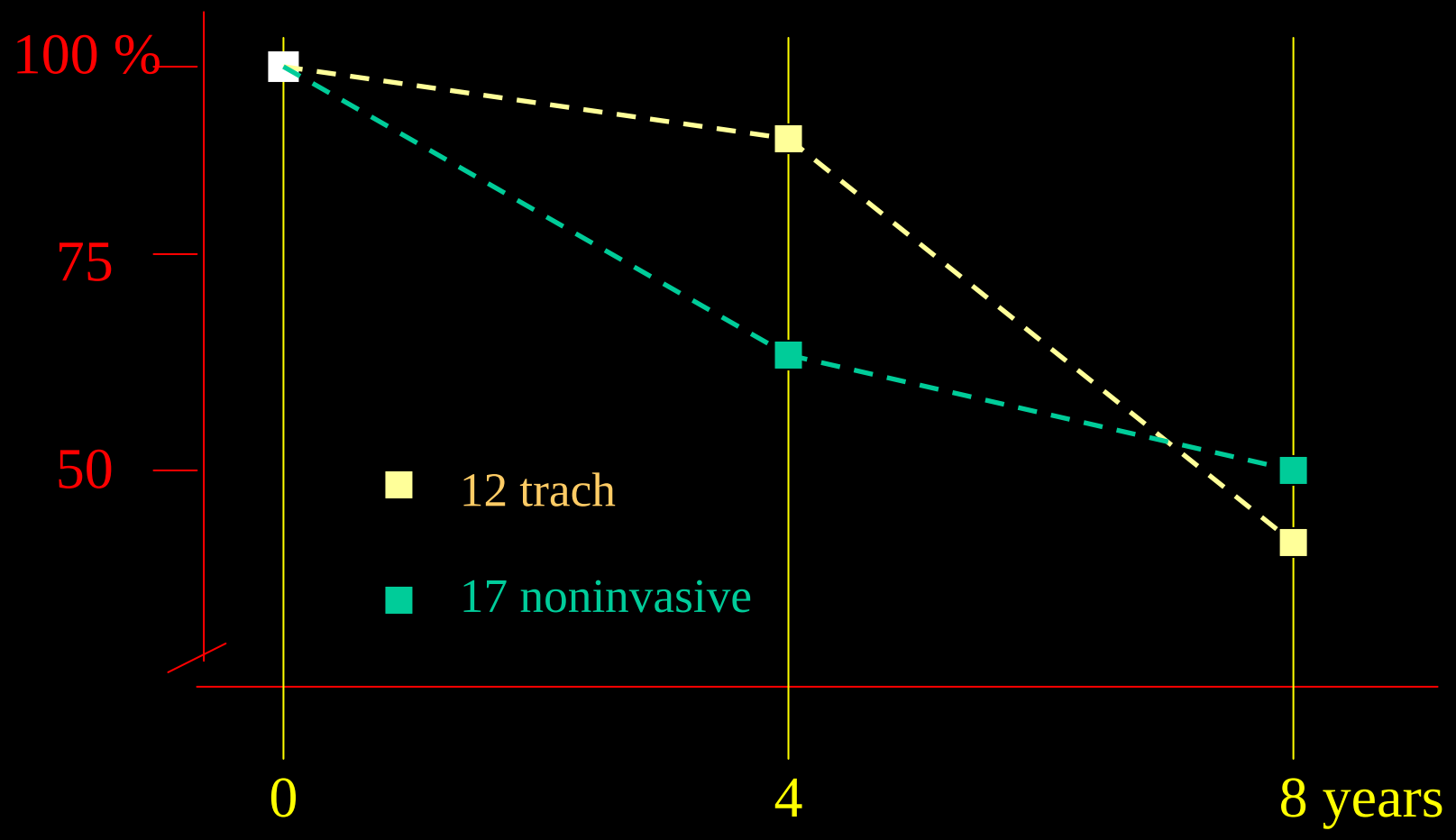


muscular dystrophies with VC < 400 ml

Causes of mortality: acute respiratory failure (ARF), congestive heart failure (CHF), low output syndrome (LOS)



muscular dystrophies with VC < 400 ml



survival <u>trach vs non invasive</u> over 8 years (non randomized comparaison)

How to live without respiratory capacity and without tracheostomy

continuous ventilation requires:

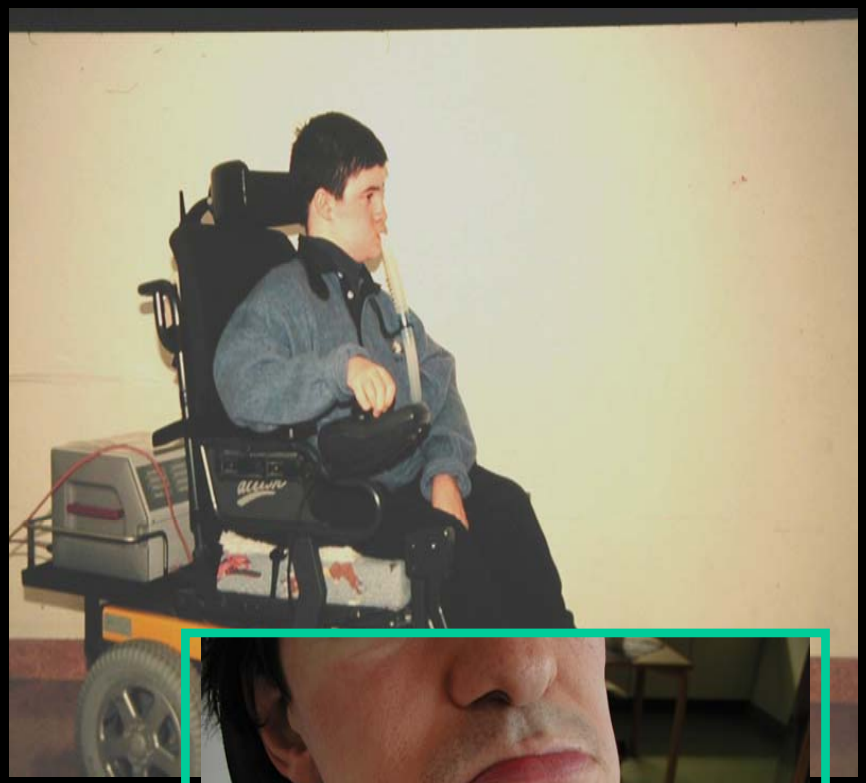
some technical details …

How to live without respiratory capacity and without tracheostomy

continuous ventilation requires:

*** adaptation of ventilatory mode**

How to live without respiratory capacity and without tracheostomy



continuous ventilation requires:

*** adaptation of ventilatory mode**

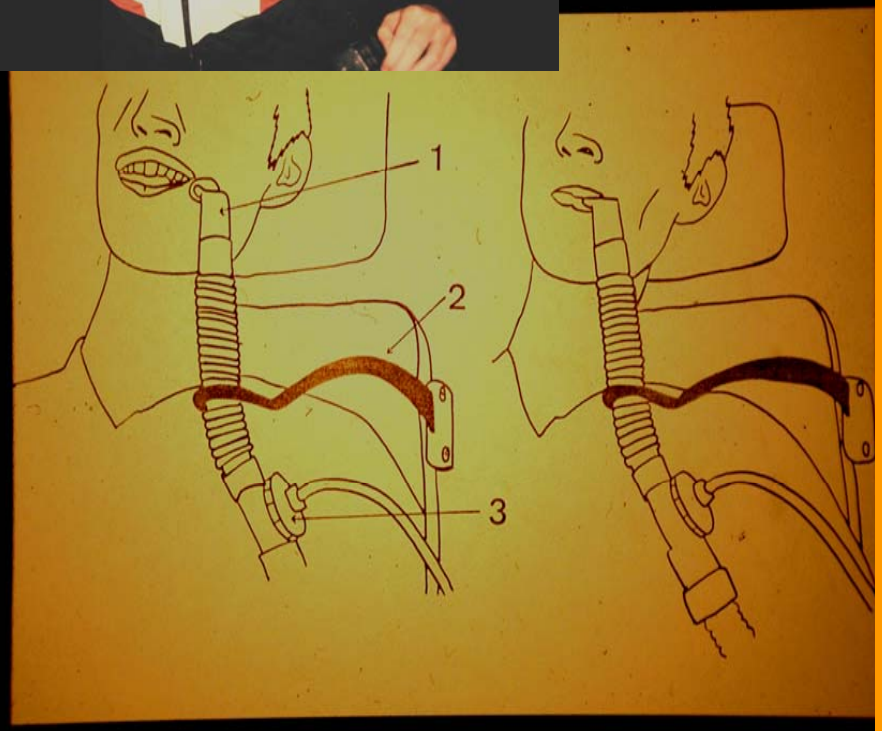
nasal mask doesn't fit for the day:

MOUTHPIECE used in the daytime

- avoids complications of trach
- is no definitive mutilation
- costs less than trach
- allows a better social integration

VENTILATION DURING DAYTIME

MOUTH PIECE



mouth piece

- has to be removed for
  - Speaking
  - Swallowing
- may not fall down
- so it has to be fixed:
  - on the patient (shoulders)
  - on the wheelchair
  - on the bed

How to live without respiratory capacity and without tracheostomy

continuous ventilation requires:

* adaptation of ventilatory mode
* **adjustment of wheelchair**

How to live without respiratory capacity and without tracheostomy

continuous ventilation requires:

* adaptation of ventilatory mode
* **adjustment of wheelchair**

* ventilator installed
  in the back of the wheelchair

How to live without respiratory capacity and without tracheostomy

continuous ventilation requires:

* adaptation of ventilatory mode
* **adjustment of wheelchair**

* ventilator installed in the back of the wheelchair
* installation of an additional electric battery

How to live without respiratory capacity and without tracheostomy

<u>continuous ventilation requires</u>:

* adaptation of ventilatory mode
* **adjustment of wheelchair**

* ventilator installed in the back of the wheelchair
* installation of an additional electric battery
* using an adapted driving system, ex: without resistance (Vic®)

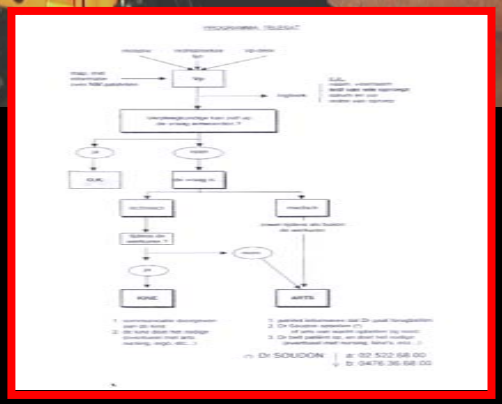
iron piece moving in a magnetic field

How to live without respiratory capacity and without tracheostomy

continuous ventilation requires:

* adaptation of ventilatory mode
* adjustment of wheelchair
* **safety procedures**

How to live without respiratory capacity and without tracheostomy



continuous ventilation requires:

* adaptation of ventilatory mode
* adjustment of wheelchair
* **safety procedures**

* training of all potential helpers, alarms, technical and assistance procedures (ex: fire brigade)

How to live without respiratory capacity and without tracheostomy

continuous ventilation requires:

* adaptation of ventilatory mode
* adjustment of wheelchair
* **safety procedures**

* training of all potential helpers, alarms, technical and assistance procedures (ex: fire brigade)
* 2d ventilator, IP percussion IP and Ambu

How to live without respiratory capacity and without tracheostomy

ALLEGATO TECNICO SISTEMA

ALS con-tel V128

Il servizio di tele-monitoraggio

Dimensioni

ALS con-tel V128:

- Grandezza 230 mm x 246 mm x 146 mm;
- Peso 1,4 Kg (Esclusi alimentatore, modem e moduli aggiuntivi, massimo quattro, in funzione degli apparati medicali da collegare)

Aspetto del sistema

Caratteristiche del sistema

ALS con-tel V128 (configurazione base) è un registratore digitale multifunzione che permette di acquisire in modo accurato e non invasivo la saturazione d'ossigeno in percentuale nell'emoglobina arteriosa, la frequenza del polso tramite l'utilizzo di un apparecchio di misurazione esterno collegato con lo strumento e dotato a seconda della configurazione, è possibile memorizzare circa 60.000 eventi di saturazione e polso per ogni sessione di registrazione, per una durata complessiva superiore alle 16 ore.

ALS con-tel V128 è stato concepito con una serie di funzionalità grazie alle quali è in grado di:

- Ricevere e memorizzare i dati ed il setup trasmessi via seriale ed in forma analogica da apparecchiature esterne a fasce orarie indipendenti con le modalità scelte dal medico;
- Proporre al paziente messaggi personalizzati e questionari che permettano la raccolta di informazioni sintomatologiche;
- Trasferire questi dati via modem ad un centro ricevente ad intervalli di tempo prestabiliti o al bisogno. Il sistema è in grado di chiamare più numeri telefonici (tre per un massimo di tre tentativi ciascuno) così da assicurare la trasmissione dei dati memorizzati. Viene inoltre tenuta traccia tramite log interno dei tentativi di trasmissione falliti così da consentire una rapida diagnosi di eventuali problemi tecnici.

ALS con-tel V128 è un prodotto realizzato per Air Liquide *Sanità* S.p.A. da QUBIsoft S.r.l.

AIR LIQUIDE *Sanità*

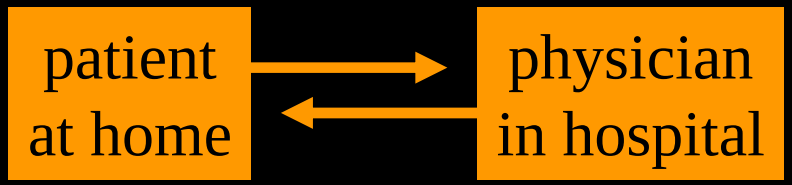
continuous ventilation requires:

* adaptation of ventilatory mode
* adjustment of wheelchair
* **safety procedures**

* training of all potential helpers, alarms, technical and assistance procedures (ex: fire brigade)
* 2d ventilator, IP percussion IP and Ambu
* Telemedicine (survey program)

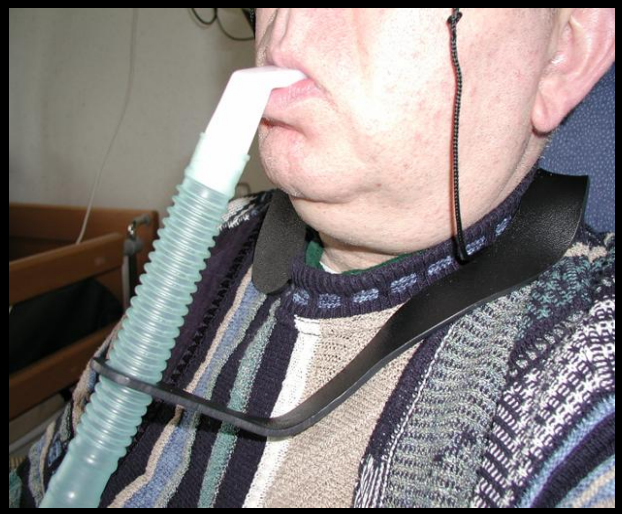
How to live without respiratory capacity and without tracheostomy

TELEMEDICINE can …



1. avoid any physical moving of the patient
2. allow monitoring SpO2 / EtCO2 in vivo conditions
3. transmit the data in (quasi) real time
4. allow the hospital to immediatly react to problems
5. provide more informations on homeventilation

How to live without respiratory capacity and without tracheostomy



1.

40 patients living at home

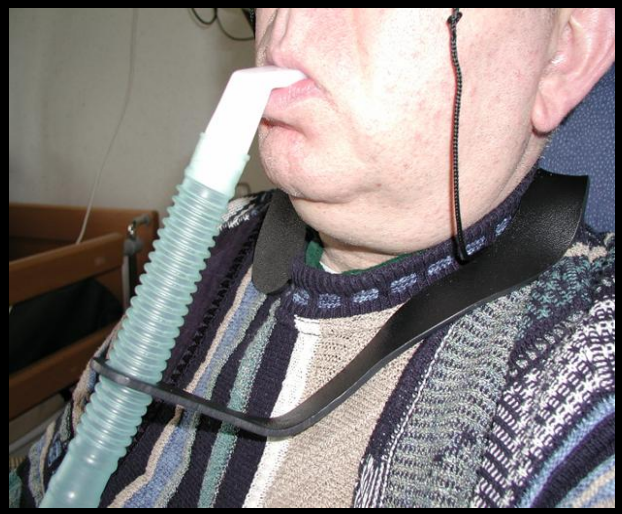
with continuous assisted ventilation

vital capacity < 400 ml

with needed equipment for

using telemedicine

How to live without respiratory capacity and without tracheostomy

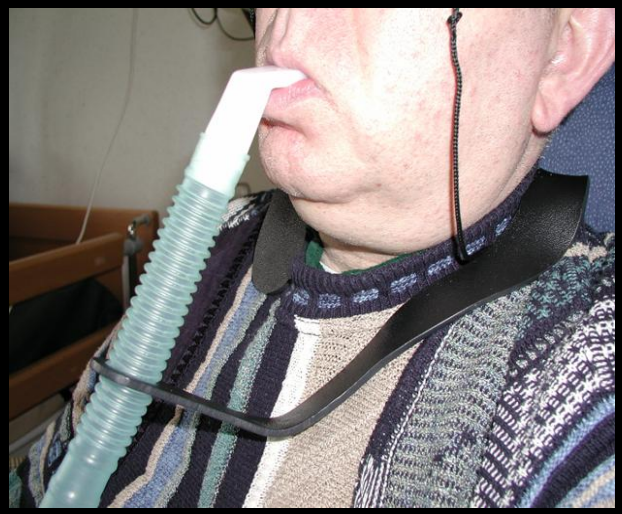


2.

monitoring and transmission:

saturometer / capnometer
Contel ®
modem
phoneline

How to live without respiratory capacity and without tracheostomy



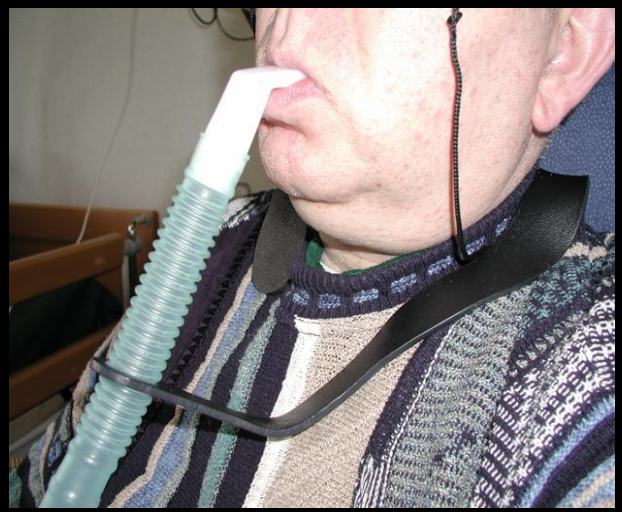
3.

reception of the data

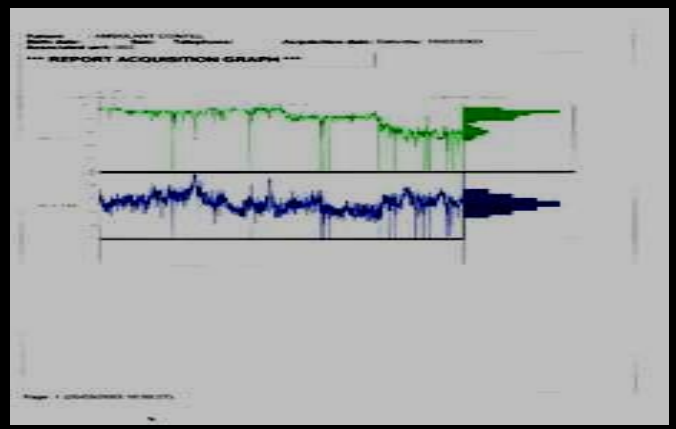
on the personal file of the patient

(PC hospital)

How to live without respiratory capacity and without tracheostomy



4.
analysing
(and reacting)



How to live without respiratory capacity and without tracheostomy

continuous ventilation requires:

* adaptation of ventilatory mode
* adjustment of wheelchair
* safety procedures
* **secretion clearing management**

How to live without respiratory capacity and without tracheostomy

continuous ventilation requires:

* adaptation of ventilatory mode
* adjustment of wheelchair
* safety procedures
* **secretion clearing management**

* chronic secretions
  require **specific** physiotherapy
  and external tracheal suction
  device (ex: cough assist ®)

How to live without respiratory capacity and without tracheostomy

continuous ventilation requires:

* adaptation of ventilatory mode
* adjustment of wheelchair
* safety procedures
* **secretion clearing management**

* chronic hypersecretion
* respiratory surinfections
  antibiotics, corticoids,
  oxygen, etc ...

How to live without respiratory capacity and without tracheostomy

noninvasive ventilation 24h/24
nasal mask (sleep)
mouthpiece (awake)

some questions …

How to live without respiratory capacity and without tracheostomy

noninvasive ventilation 24h/24
nasal mask (sleep)
mouthpiece (awake)

indications for trach ?

How to live without respiratory capacity and without tracheostomy

noninvasive ventilation 24h/24
nasal mask (sleep)
mouthpiece (awake)

indications for trach ?

limits of use ?

24hours noninvasive ventilation: **indication for secondary trach ?**

<u>a priori</u> indications for trach in patient already 24h non invasive ventilated =

24hours noninvasive ventilation: **indication for secondary trach ?**

a priori indications for trach in patient already 24h non invasive ventilated =

* **ventilatory insufficiency**
not encountered

if efficient at the installation, noninvasive ventilation remains always even efficient

24hours noninvasive ventilation: **indication for secondary trach ?**

<u>a priori</u> indications for trach in patient already 24h non invasive ventilated =

* ventilatory insufficiency not encountered
* **swallowing disturbances improved with trach**

tracheo-cannula is an additional weight (resistance) for the larynx

24hours noninvasive ventilation: **indication for secondary trach ?**

<u>a priori</u> indications for trach in patient already 24h non invasive ventilated =

* ventilatory insufficiency not encountered
* swallowing disturbances improved with trach
* **chronic hypersecretion not encountered**

more less secretions with noninvasive ventilation

24hours noninvasive ventilation: **indication for secondary trach ?**

<u>a priori</u> indications for trach in patient already 24h non invasive ventilated =

* ventilatory insufficiency
  not encountered
* swallowing disturbances
  improved with trach
* chronic hypersecretion
  not encountered
* **acute respiratory surinfection**
  death if trach

linked to
the severity ?

24hours noninvasive ventilation: **limits of the method ?**

limits of the method ?

24hours noninvasive ventilation: **limits of the method ?**

limits of the method ?

respiratory evolution goes on, and patient becomes more and more dependent on the ventilator.

24hours noninvasive ventilation: **limits of the method ?**

limits of the method ?

respiratory evolution goes on, and patient becomes more and more dependent on the ventilator.

it is impossible to breathe and to have food in mouth <u>together</u>

24hours noninvasive ventilation: **limits of the method ?**

limits of the method ?

respiratory evolution goes on, and patient becomes more and more dependent on the ventilator.

it is impossible to breathe and to have food in mouth <u>together</u>

with time, patient must choice between breathing and eating …

24hours noninvasive ventilation: **limits of the method ?**

limits of the method ?

respiratory evolution goes on, and patient becomes more and more dependent on the ventilator.

it is impossible to breathe and to have food in mouth <u>together</u>

with time, patient must choice between breathing and eating …

So, he eats more less, and starts losing weight …

24hours noninvasive ventilation: **limits of the method ?**

therapeutic answer = probably

<u>not</u> to place a trach
(more risks of swallowing troubles)

but **to place a gastrostomy:**

* **<u>P</u>**er **<u>E</u>**ndoscopic **<u>G</u>**astrostomy (**<u>PEG</u>**)
* under local anaesthesia
* no severe complications
* allows to feed the patient during sleep
* keeps mouth feeding for pleasure only

How to live without respiratory capacity and without tracheostomy

some conclusions …

How to live without respiratory capacity and without tracheostomy

some conclusions …

24hours **noninvasive** ventilation is technically **possible**

How to live without respiratory capacity and without tracheostomy

some conclusions …

24hours **noninvasive** ventilation is technically **possible**

mouth piece ventilation is **even** efficient as tracheostomy

How to live without respiratory capacity and without tracheostomy

some conclusions …

- 24hours **noninvasive** ventilation is technically **possible**
- mouth piece ventilation is **even** efficient as tracheostomy
- mouth piece ventilation costs **much less** than tracheostomy

How to live without respiratory capacity and without tracheostomy

some conclusions …

- 24hours **noninvasive** ventilation is technically **possible**
- mouth piece ventilation is **even** efficient as tracheostomy
- mouth piece ventilation costs **much less** than tracheostomy
- mouth piece ventilation allows a **better social integration**

How to live without respiratory capacity and without tracheostomy

some conclusions …

- 24hours **noninvasive** ventilation is technically **possible**
- mouth piece ventilation is **even** efficient as tracheostomy
- mouth piece ventilation costs **much less** than tracheostomy
- mouth piece ventilation allows a **better social integration**
- **no demand for euthanasia** from these patients (Belgium)

many thanks
for your attention …

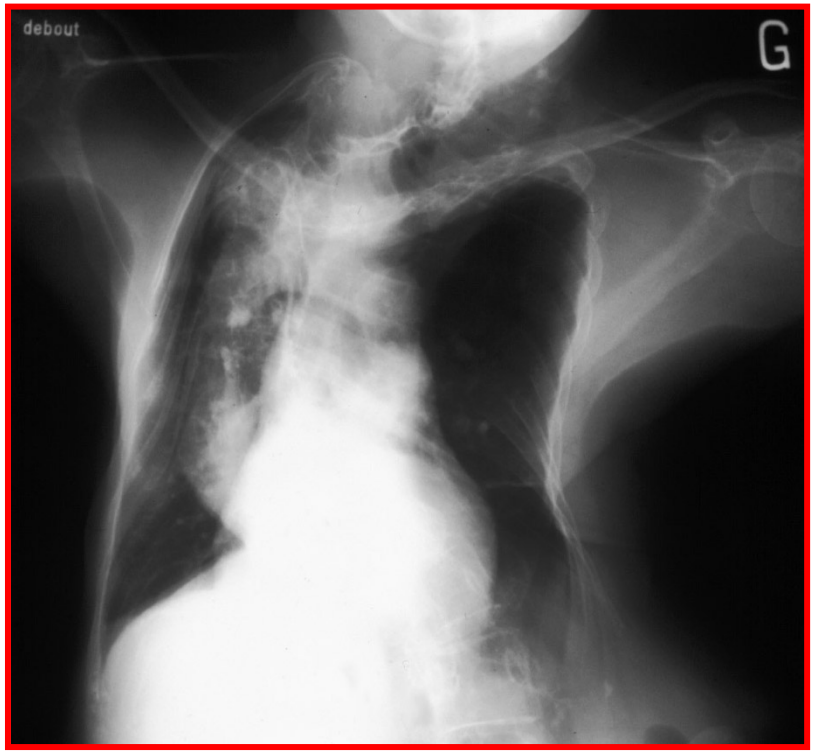
philippe . soudon
@ inkendaal . be

Monitoring long term home mechanical ventilation

Dr JP Janssens, Division of Pulmonary Diseases,
Geneva University Hospital, Switzerland

Home mechanical ventilation: for whom?

- Restrictive disorders:
  - Kyphoscoliosis
  - Neuro-muscular disorders
  - Post-tuberculosis
  - Severe obesity
- Obstructive disorders:
  - COPD
  - Bronchiectasis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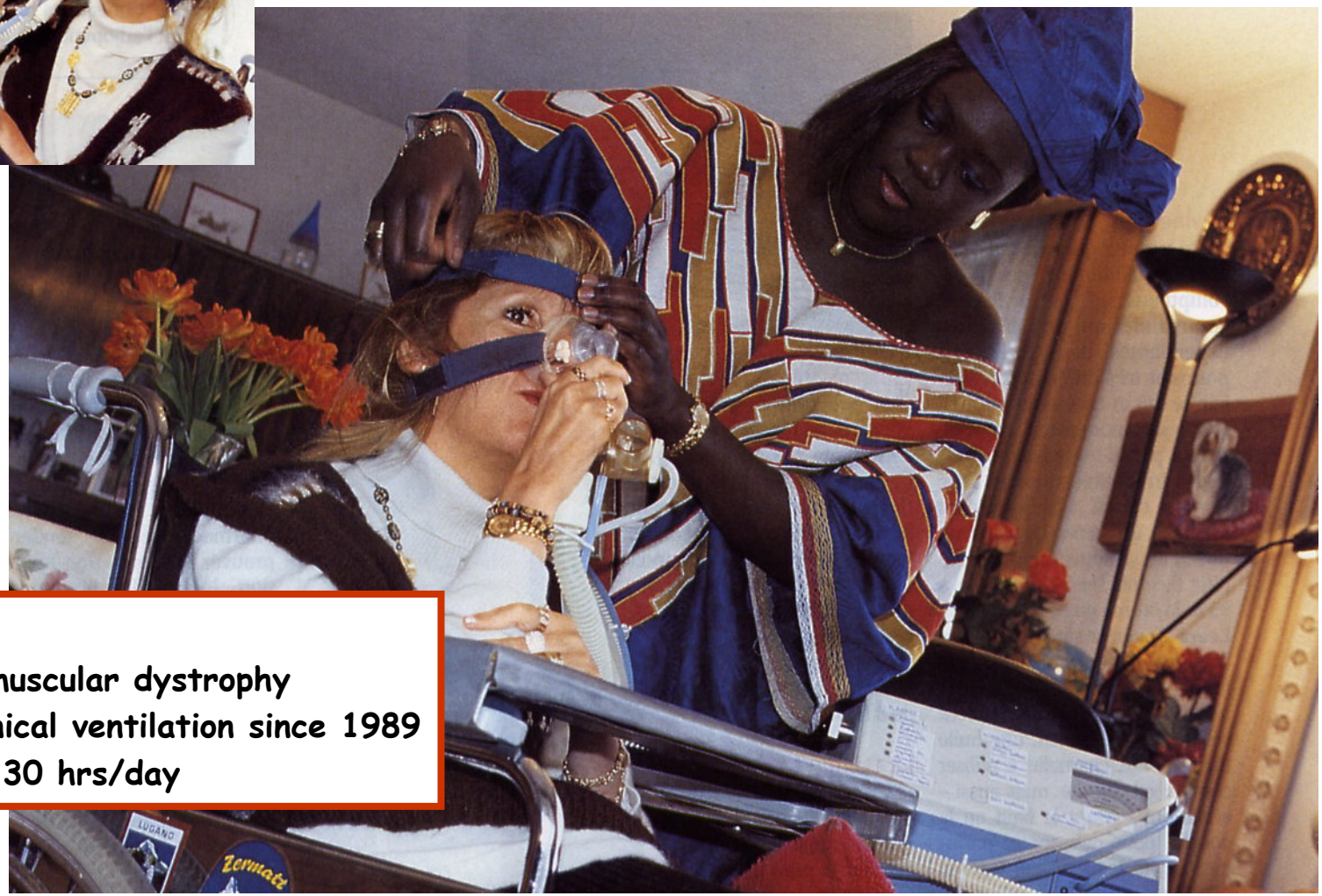


- 44 years old
- Limb-girdle muscular dystrophy
- Home mechanical ventilation since 1989
- Presently 23:30 hrs/day

Consequences of chronic hypercapnic respiratory failure

Respiratory pump failure

Alveolar hypoventilation

Hypoxia
Hypercapnia
Respiratory acidosis

Pulmonary hypertension
Cor pulmonale
Fluid retention

Symptoms of hypercapnia:
Fatigue, obnubilation, morning headaches
profuse sweating, dyspnea...

Positive pressure ventilators: pressure-cycled or volume-cycled

Measuring the quality of long term home mechanical ventilation (HMV): goals

- Assessment of correction of hypoventilation and related symptoms
- Assessment of nocturnal SaO_2
- Assessment and correction of side-effects of HMV
- Quality of ventilation: leaks? Patient-ventilator synchronization?
- Quality of sleep
- Adherence to treatment
- Quality of life

Necessary Logistics

Necessary logistics for monitoring home mechanical ventilation

- « Expert » referral center (hospital with medical team experienced in HMV) with a clearly identified medical supervisor
- Outpatient structure with specially trained personnel for checking technical equipment, and performing basic medical evaluation at home
- Regular home visits
- Collaboration with family general practitioner
- Financial coverage (medical insurance, Social Security) of equipment, necessary accessories and domiciliary interventions

Frequency of monitoring for patients with home mechanical ventilation

- **Home visits:** frequent after implementing HMV (after discharge from hospital, after 1 month), then according to medical condition, and acceptance of treatment (once every 2 – 6 months)
- **Short (24 hour) hospital evaluations:** within 2 months of hospital discharge, then at least on a yearly basis, unless problems identified by team performing home visits.

Home visits

- Performed by experienced personnel
- In our area, nurses with a training in intensive care
  - Standardized questionnaire for medical history
  - Simple physical examination: Weight, Heart rate, BP, Respiratory rate, detection and quantification of peripheral oedema
  - Recording of basic parameters: FEV_1, FVC, SaO_2 w and w/o ventilator
  - Optional: possibility of downloading data from ventilator and organizing pulsoxymetry and/or $TcPCO_2$ recording at home

Hospital routine « check-ups »

- Usually less than 24 hours
- Short medical history and physical examination
- Electrocardiogram; Chest X-ray
- Pulmonary function tests including respiratory muscle testing
- Arterial blood gases with and without ventilator
- Nutritional assessment: bio-impedance measurements
- Night recording of SaO_2, $TcPCO_2$, and (optional) polygraphy
- Downloading of software of ventilator, ventilator counter
- Verification of equipment
- Necessary adjustments and recommendations

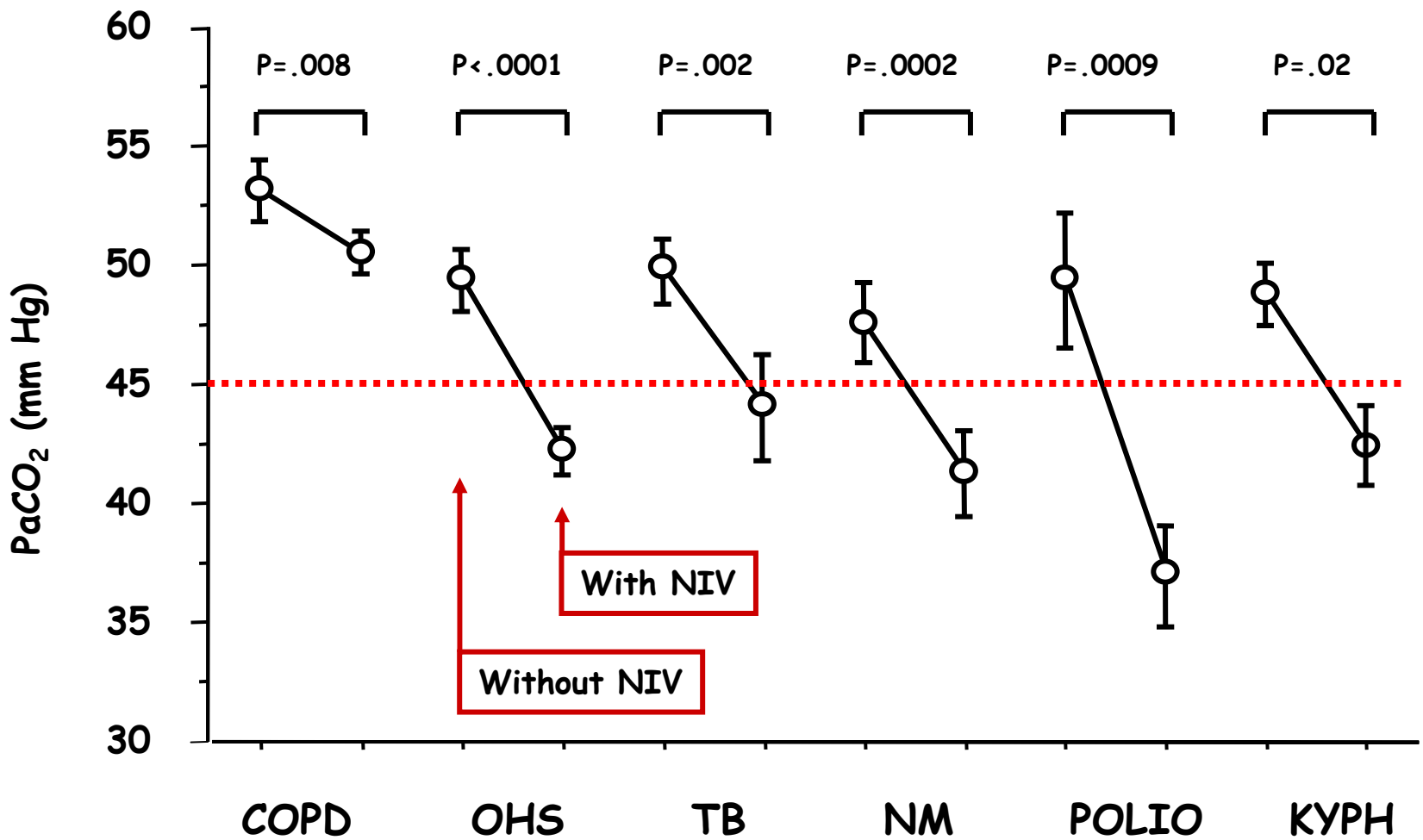
Tools for monitoring home ventilation

Monitoring quality of long term home mechanical ventilation (HMV): tools for studies vs. clinical practice

Clinical practice	Study purposes
• Daytime arterial blood gases • Pulsoximetry • Nocturnal $TcPCO_2$ (or $ETCO_2$) • Built-in ventilator software • Polygraphy • Check-list for side-effects • Bioimpedance • Epworth Sleepiness Scale	• Polysomnography • MSLT, MWT* • Generic or specific QoL questionnaires

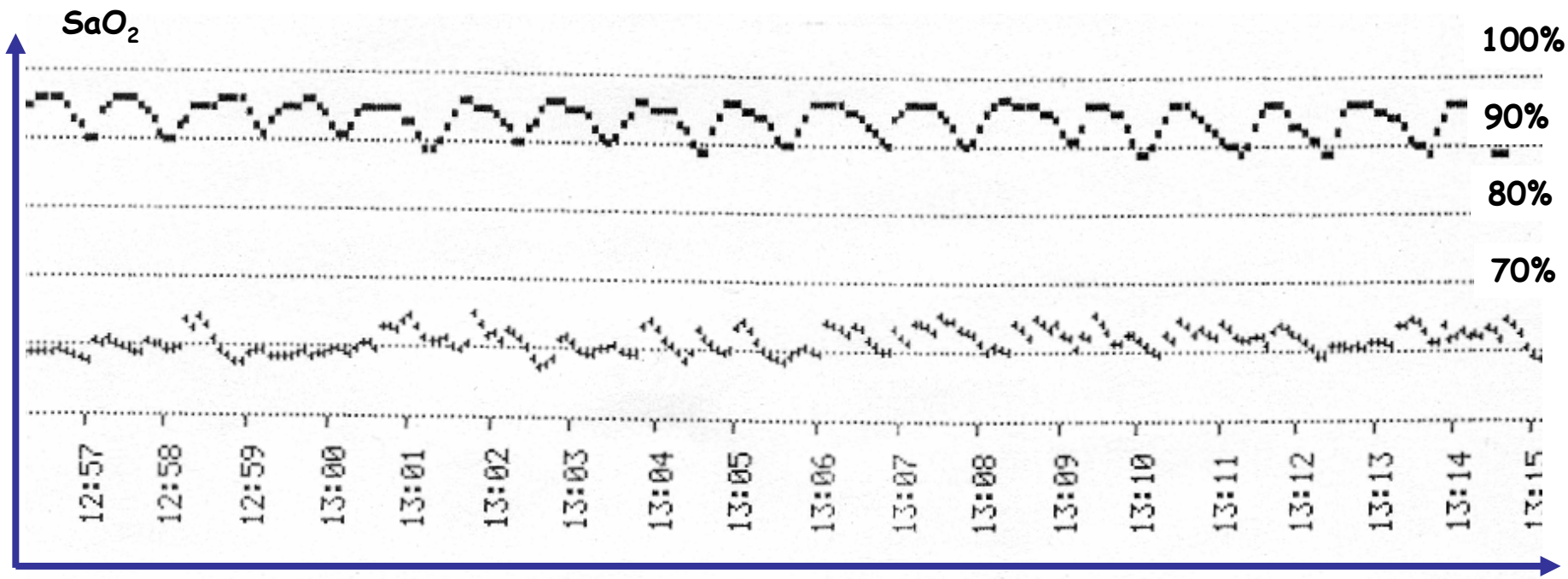
*Multiple Sleep Latency Test; Wakefullness Maintenance Test

Daytime arterial blood gases with and without ventilator (n=211)

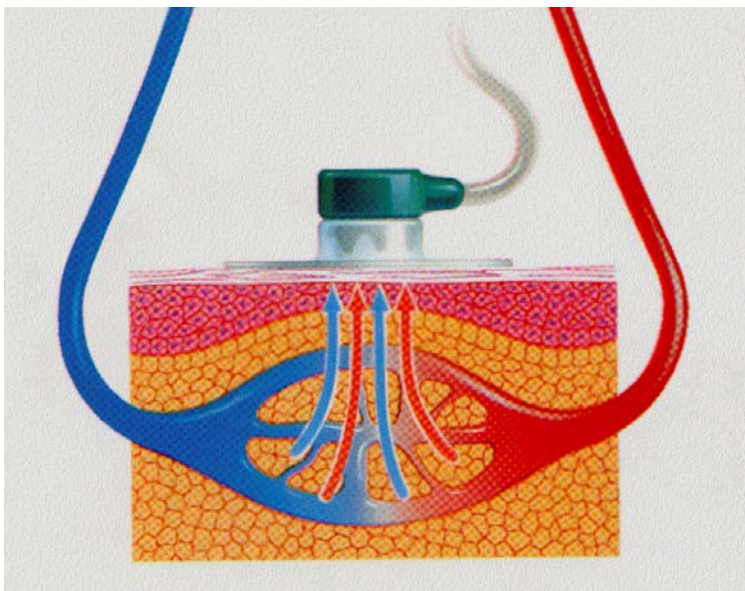


Pulsoximetry

- Reasonable goal: Mean SaO_2 > 90% and less than 20% of total recording with a SaO_2 < 90%
- No specificity of desaturations
- Not very sensitive in patients with supplemental oxygen



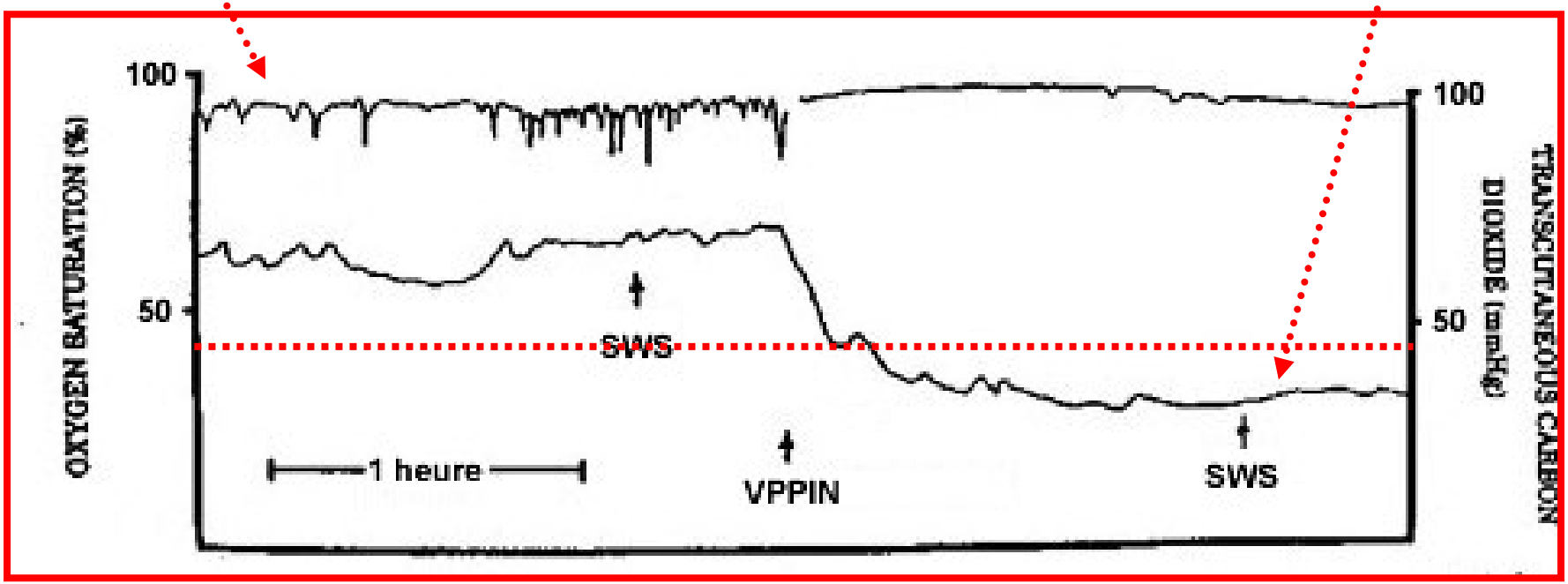
Transcutaneous CO_2 ($PtcCO_2$) for monitoring long-term home mechanical ventilation



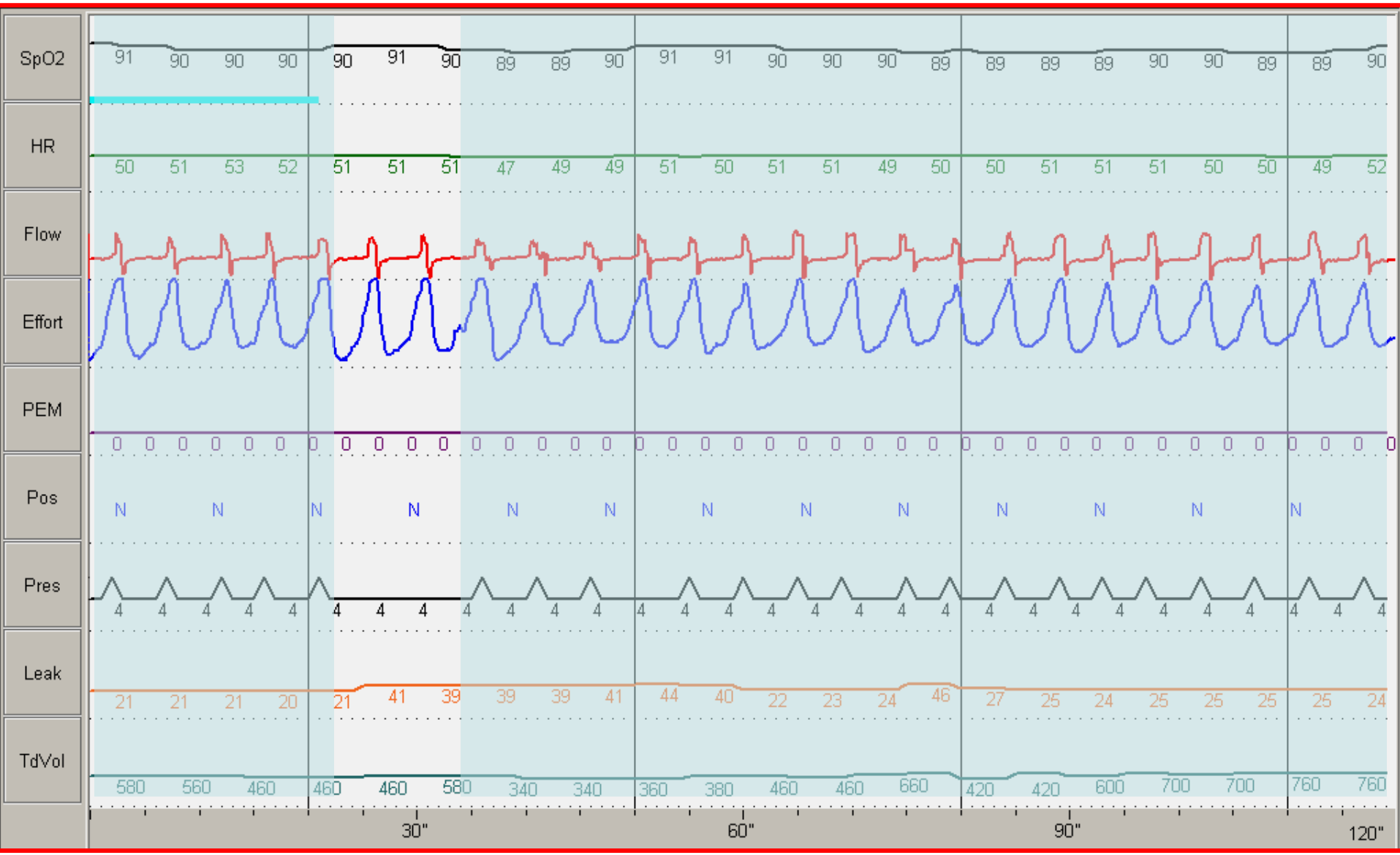
SaO_2

Introduction of nasal PPV

$PtcCO_2$



Built-in computer software with polygraphy



Synchrony® ventilator (Respironics) & Stardust® polygraph

Profil d'utilisation | Statistiques | Détails patient | Commentaires patient | Données détaillées

Afficher 30 minutes | Produit VPAP III ST A | No. de série 000000020040461182

Date 08/06/2005 | Heure de début 06:49:38 | Heure de fin 07:19:38

mercredi, 8 juin 2005

0:00 am 06:55:00 am 07:00:00 am 07:05:00 am 07:10:00 am 07:15:00 am 07:20

Fuite l/sec — 0.70, 0.40, 0.00

Ventilation minute l/min — 20.00, 14.00, 7.00, 0.00

Pouls bpm — 90, 60, 30, 0

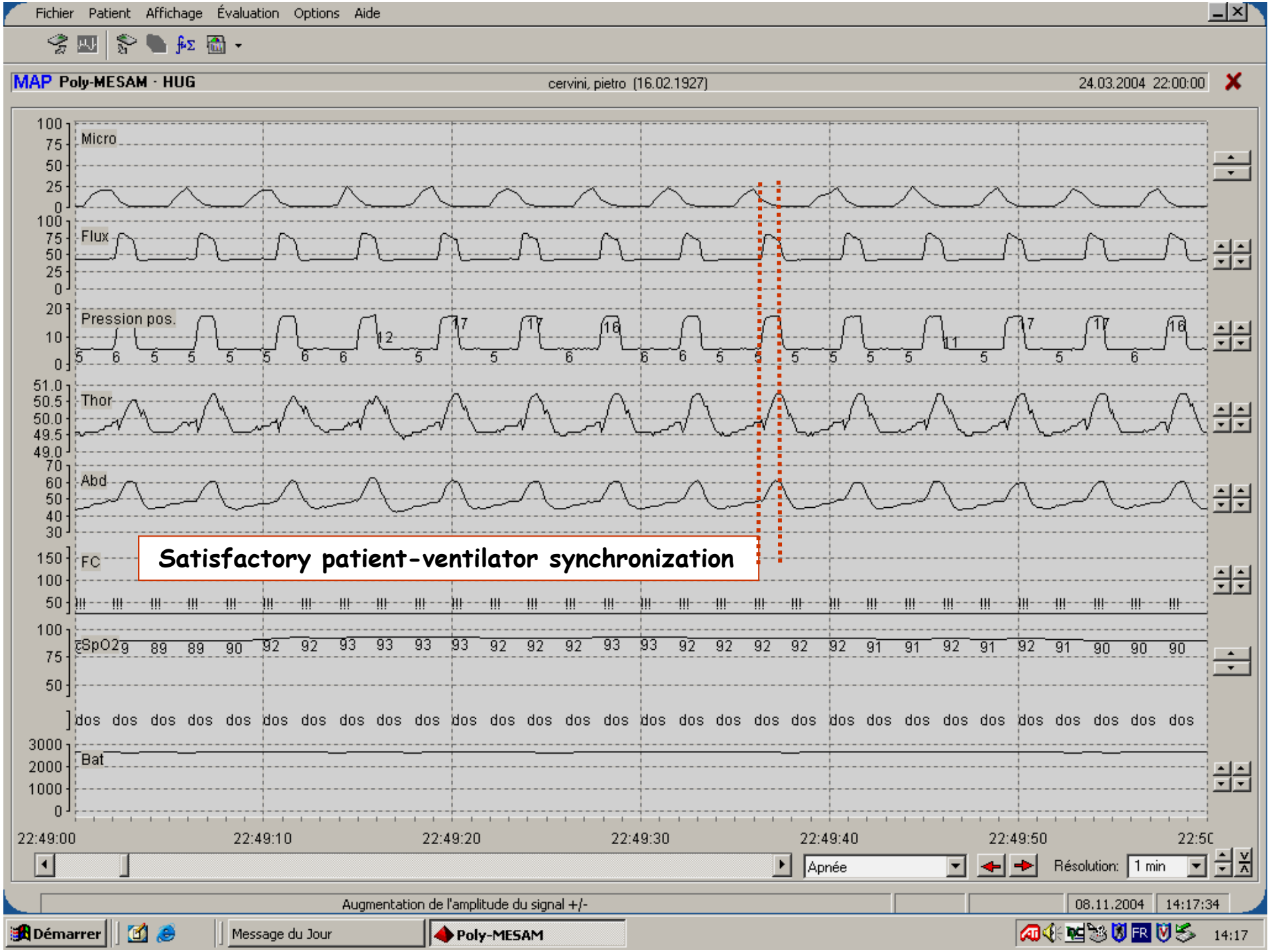
SpO_2 % — 95, 80, 65, 50

Evénements sec. — 100, 70, 40, 0

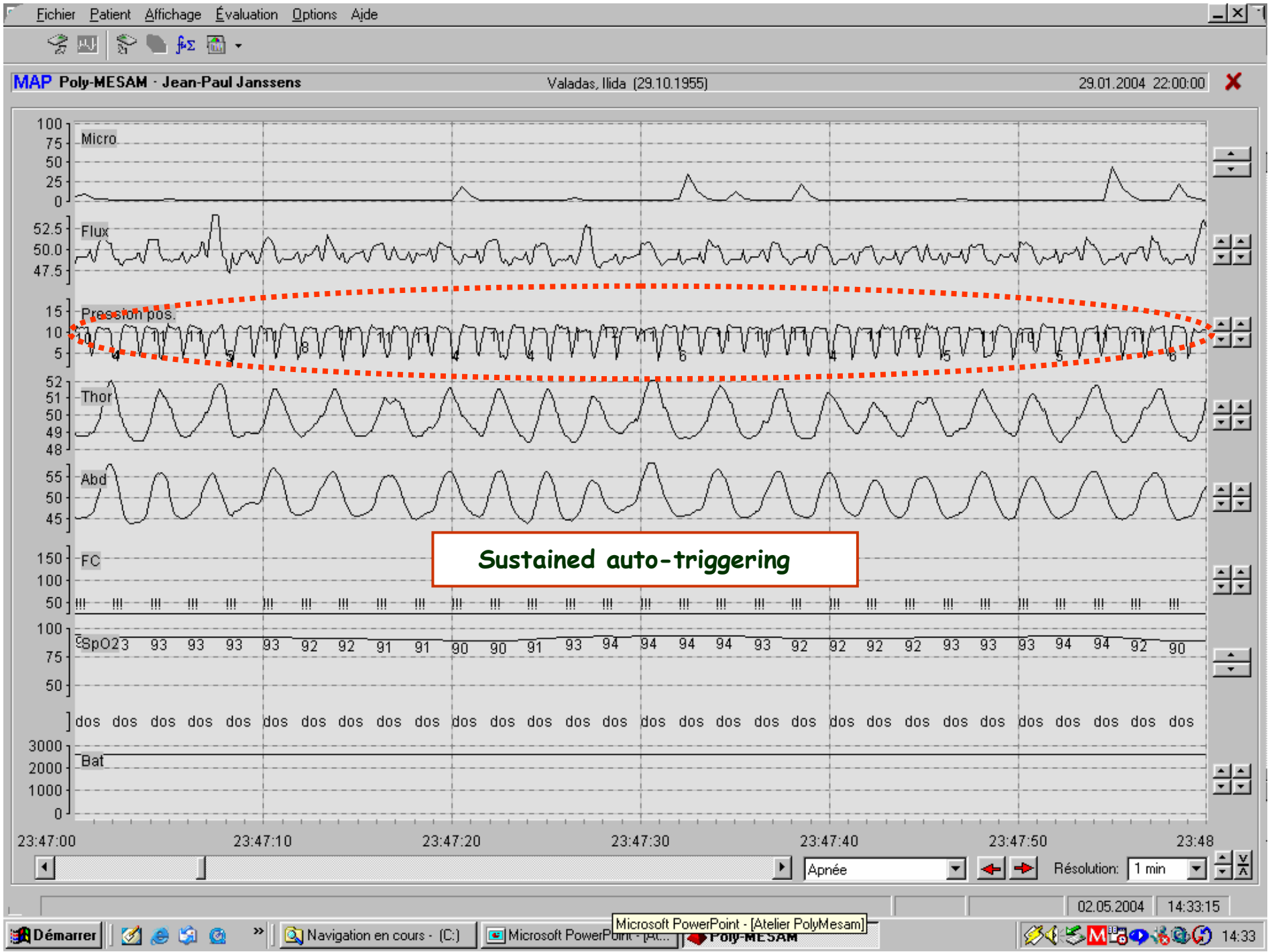
IAH événements/h — 30, 20, 10, 0

Fréq resp bpm (respirations par minute) — 30.00, 20.00, 10.00, 0.00

Analysis of Patient-Ventilator Synchronisation


Fichier Patient Affichage Évaluation Options Aide
MAP Poly-MESAM - HUG
cervini, pietro (16.02.1927)
24.03.2004 22:00:00
Micro
Flux
Pression pos.
Thor
Abd
FC
cSpO2
Bat
Satisfactory patient-ventilator synchronization
22:49:00
22:49:10
22:49:20
22:49:30
22:49:40
22:49:50
Apnée
Résolution: 1 min
Augmentation de l'amplitude du signal +/-
08.11.2004
14:17:34
Démarrer
Message du Jour
Poly-MESAM
14:17

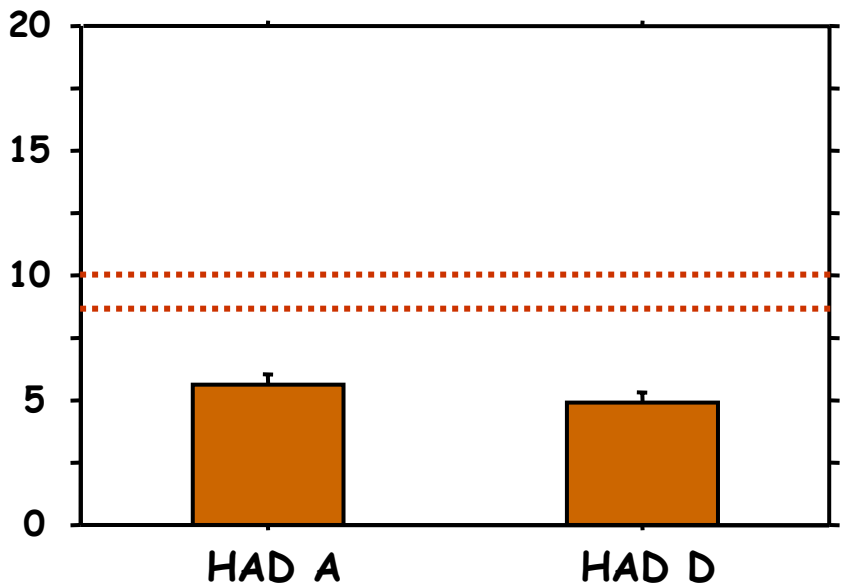
Unrewarded inspiratory efforts
—only every second inspiratory effort was supported by the ventilator


Fichier Patient Affichage Évaluation Options Aide
MAP Poly-MESAM · Jean-Paul Janssens
Valadas, Ilida (29.10.1955)
29.01.2004 22:00:00
Micro
Flux
Pression pos.
Thor
Abd
FC
SpO2
Bat
Sustained auto-triggering
23:47:00
23:47:10
23:47:20
23:47:30
23:47:40
23:47:50
23:48
Apnée
Résolution: 1 min
02.05.2004
14:33:15
Démarrer
Navigation en cours - [C:]
Microsoft PowerPoint - [Atelier PolyMesam]
Poly-MESAM
14:33

Monitoring quality of life

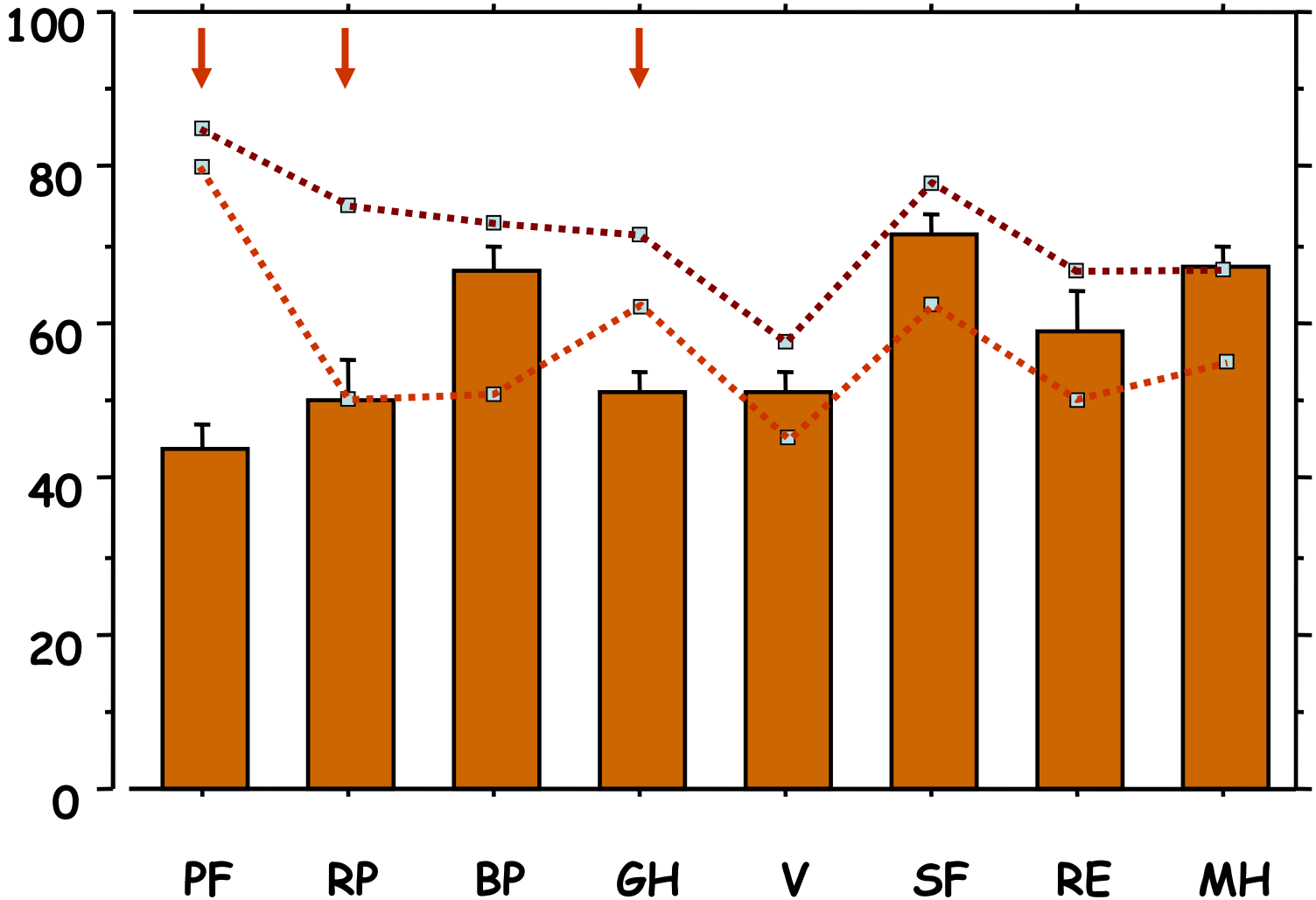
Health-related quality of life in home mechanical ventilation: a multicentric Swiss study

JP Janssens[1]; A Héritier-Praz[2]; L Burdet[3]; JW Fitting[3]; C Uldry[4]; JM Tschopp[2]; T Rochat[1]
1: HUG, Genève; 2: CVP, Crans-Montana; 3: CHUV, Lausanne; 4: Hôpital de Rolle, VD



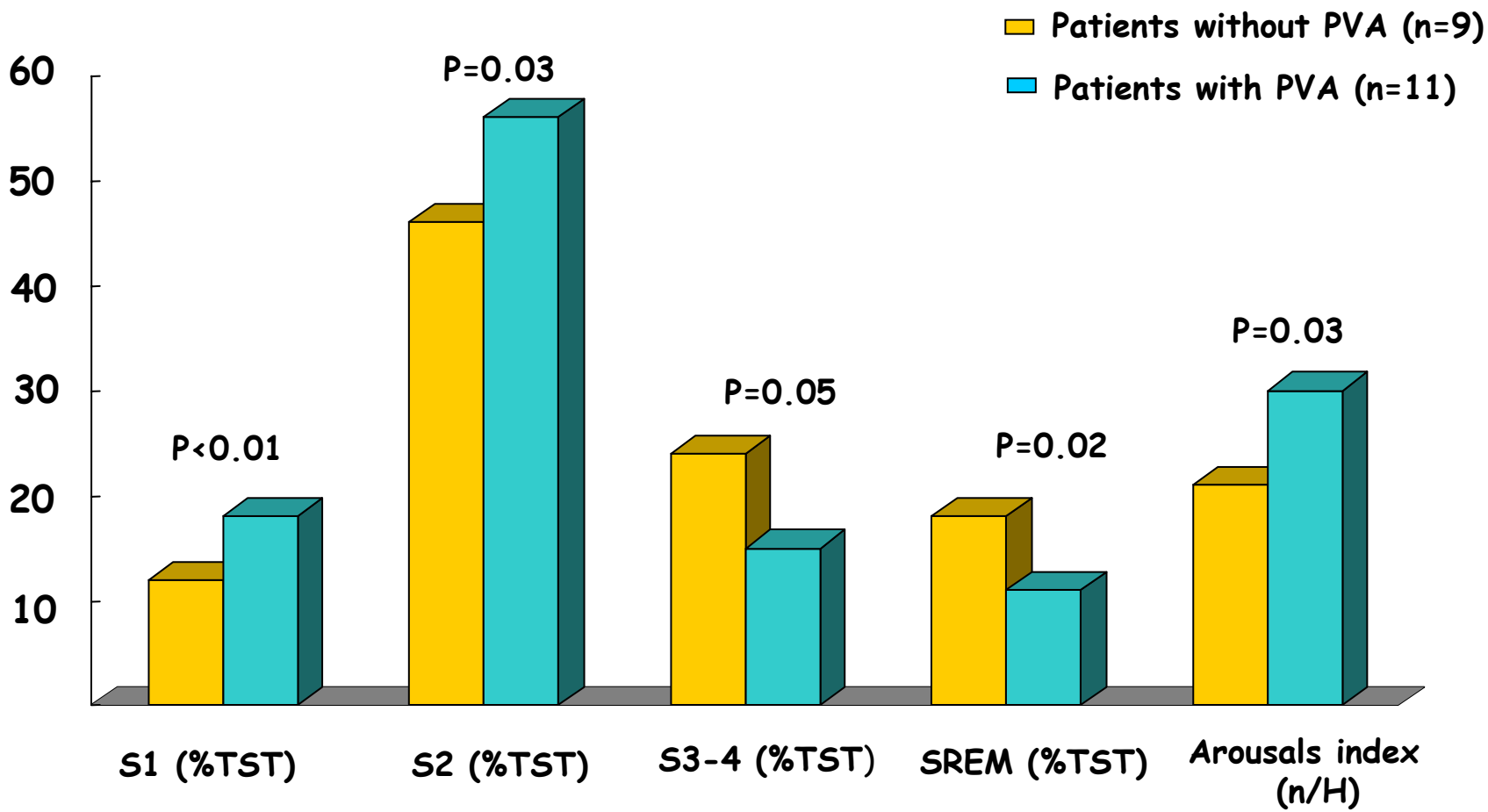
HAD scores for anxiety & depression

Presented at the European Respiratory Society meeting 2002; Stockholm



SF-36 in patients under NIV. Upper broken line indicates mean values for a Swiss normal reference population. Lower line indicates 25thpercentile for the same population

Patient-Ventilator Asynchrony (PVA) is associated with a decrease in sleep quality and an increase in arousals



Y Guo, E Sforza, JP Janssens; European Respiratory Society 2005, Copenhagen

Concluding remarks

- Home ventilation is only possible if the patient can rely on optimal co-operation between an expert center (hospital) and an outpatient team with a 24-hour home support
- Financial support must allow access to up-to-date equipment and monitoring
- Newer ventilators allow a better follow-up of adherence to treatment and efficacy of ventilation
- Regular monitoring is mandatory to detect changes in efficacy of NIV or in clinical status
- Sleep studies, although not mandatory, may prove helpful for optimal adjustment of patient-ventilator synchronisation

Thank you for your attention…

Swallowing problems- indication and benefit of PEG application in patients with neuromuscular disorders

Assist. Prof. Željko Krznarić
Zagreb, Croatia

Swallowing is an important part of our daily lives.

Swallowing transports water and food from our mouth to our stomach to provide nutrition and hydration.

- Normal swallowing can be divided into the following phases:
  - Oral phase, which involves oral preparatory phase and oral transit phase
  - Pharyngeal phase
  - Esophageal phase

- Any disease which alters the nerves or muscles which are involved with swallowing produces swallowing difficulty.

Swallowing disorders, or dysphagia, is a general term used to describe the inability to move food from the mouth to the stomach.

Significant swallowing problems were reported in:

- 51% of individuals with polymyositis,
- 44% with myasthenia gravis,
- 33% of those with myotonic muscular dystrophy,
- 32% with spinal muscular atrophy,
- 20% with Duchenne muscular dystrophy,
- 6% of those with facioscapulohumeral dystrophy.

- The percentage of swallowing disorders in amyotrophic lateral sclerosis approaches 100% as the disorder progresses (dysphagia may also be the first symptom of the disease).

- Symptoms and signs:
  - Difficulty in initiating swallowing
  - A feeling of obstruction as if food has become stuck in the throat
  - Difficulty with chewing or weakness of muscles of mastication
  - Pocketing of food in the mouth
  - Globus sensation or pain in the hypopharynx
  - Coughing after eating
  - Nasal regurgitation
  - Inappropriate breathing or speaking while swallowing
  - Weight loss
  - Recurrent pneumonia

- Many of those swallowing problems in neuromuscular disease are mild and can be dealt with by care with the type and consistency of food.

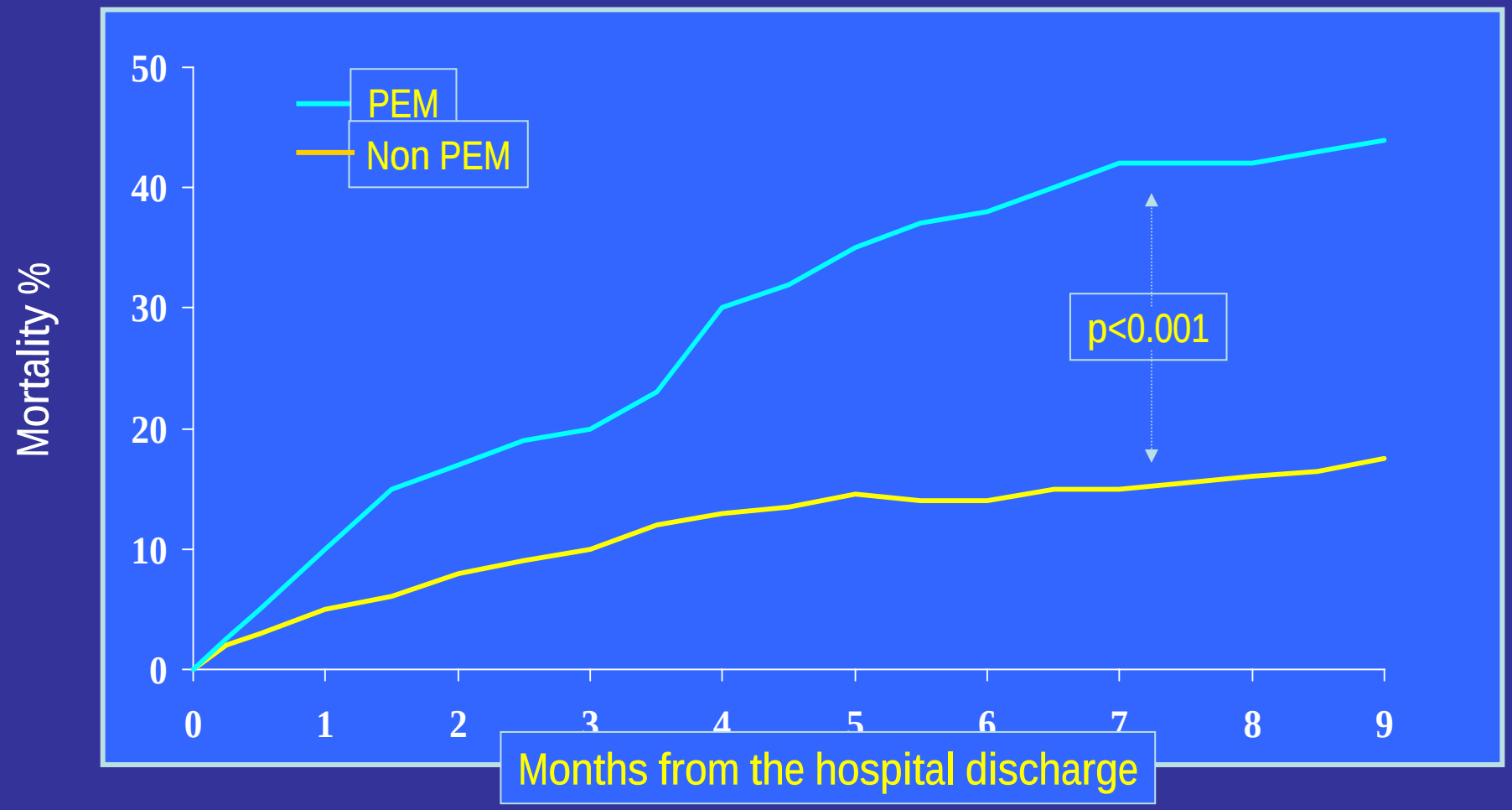
- The reasons to obtain a formal work-up are:
  - Frequent coughing or choking after taking food.
  - Pneumonia after an episode with coughing after food.
  - Two or more episodes of pneumonia in one year.
  - More than five kg loss when you are not trying to lose weight.
  - The time it takes to eat is more prolonged when compared to one year ago.

Important issue !

- all families of the patients with neuromuscular disorders should be trained in the Heimlich maneuver in order to provide resuscitation if a family member chokes on something that blocks the airway.

- Many people who have ALS will develop dysphagia (difficulty swallowing) at some point. Swallowing can go from adequate to significantly impaired in months. This can lead to **malnutrition** and **weight loss** from inadequate protein and calorie intake. When this occurs, the body uses muscle as a source of energy and protein. This accelerates the progression of weakness.
- A feeding can restore adequate protein and calorie intake.

EFFECTS OF PROTEIN-ENERGY MALNUTRITION ON CUMULATIVE MORTALITY



Cederholm T, Am J Med, 1995

Main goals of nutritional support

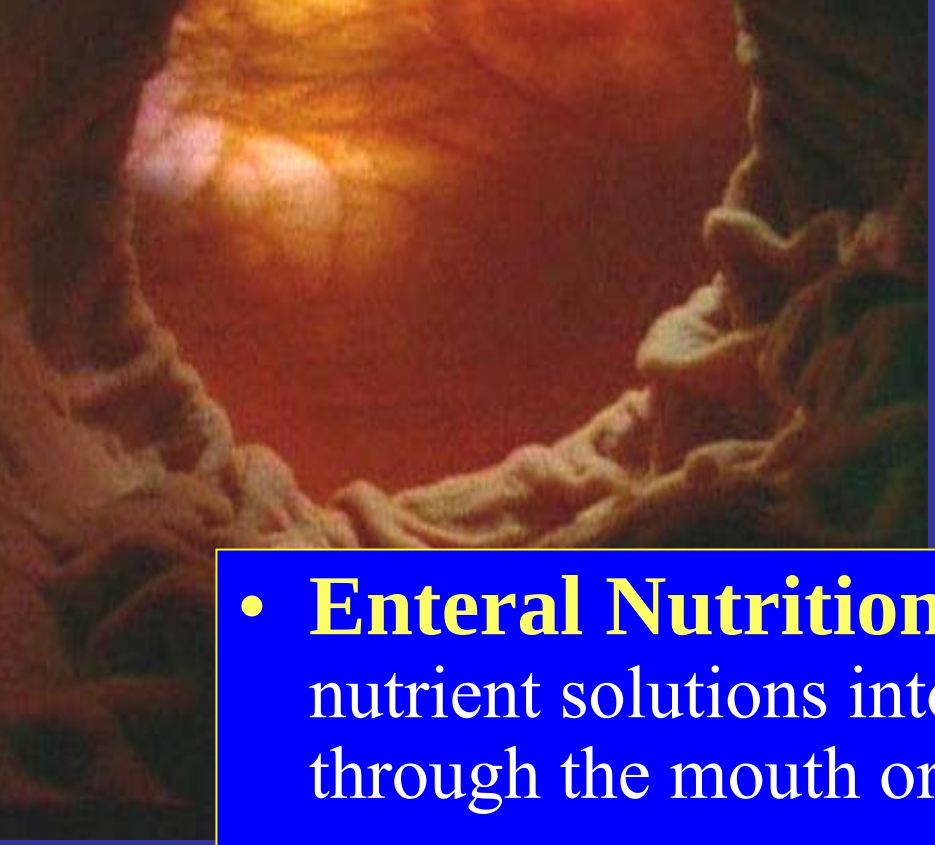
- prolong life and not prolong dying
- improve comfort (eg. discharge from the hospital)
- prevent nutritional decline and its consequences (eg pressure sores and infections)

Nutritional support

PARENTERAL

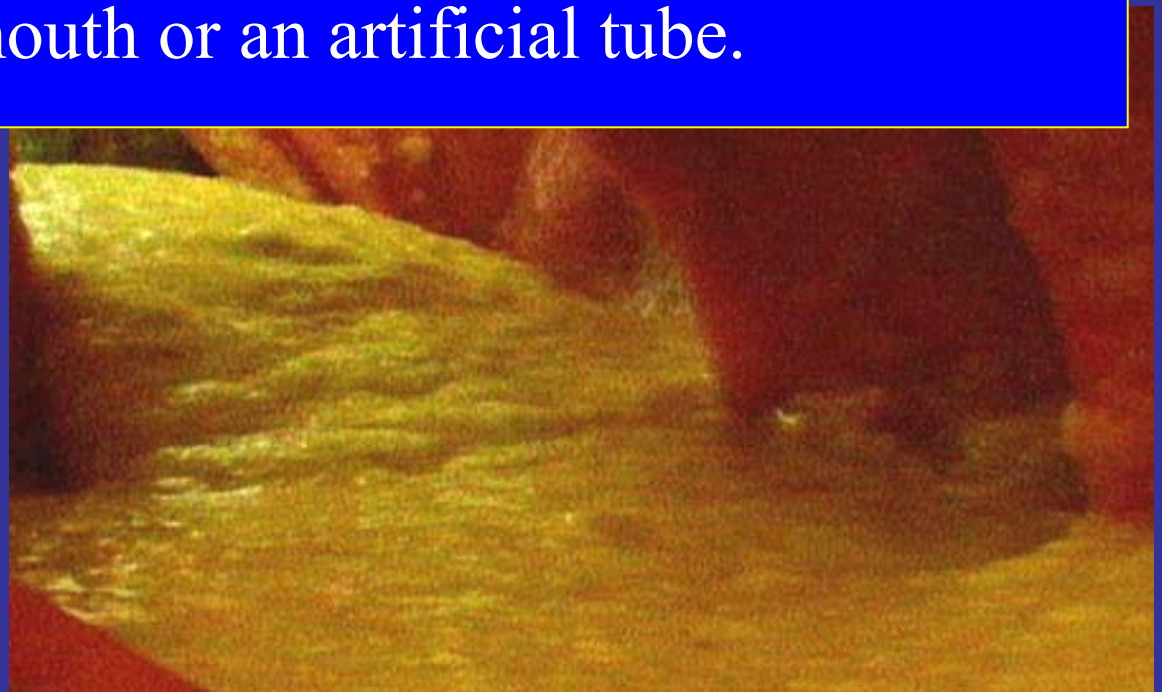
ENTERAL

Parenteral alimentation and IV fluid replacement should be prescribed to meet caloric and fluid requirements. The necessary caloric/fluid needs must be calculated to meet the patient's daily needs.



Definition

- **Enteral Nutrition:** method of providing nutrient solutions into the gastrointestinal tract through the mouth or an artificial tube.



Enteral nutrition
Tube Feeding

Prior to the insertion of an enteral feeding tube, the expected benefit of enteral feeding via a tube must be considered in the individual case, taking into account the clinical progress of the patient concerned, the prognosis, the ethical aspects and the expected quality of life of the patient with respect to retention of autonomy

Löser Chr, Müller MW. Z Gastroenterol 1998
Rabeneck L, McCullough LB, Wray NP. Lancet 1997

Proper device selection

The choice of access to the intestinal tract for enteral nutrition is directed by several factors:

- clinical prognosis,
- anticipated duration of feeding,
- patency and motility of the gut,
- risk of aspirating gastric contents

Nasogastric (NG) tubes are convenient for the short term, but their use is limited by complications including regurgitation, irritation, bleeding, and discomfort.

Q & A

- ***Q:*** What is a feeding tube by PEG?

- ***A:*** A feeding tube is a small, flexible tube, used to allow liquid to enter the stomach directly, bypassing the mouth, throat and esophagus.

PEG

- Means:
- percutaneous (through the skin)
- endoscopic (into the GI tract)
- gastrostomy (hole in the stomach).

- It's also simply called a gastrostomy tube, or g tube

Q & A

- ***Q:*** What's the right time to put in a feeding tube?
- ***A:*** Because of the nutritional deficiencies and the risk of aspiration, prefer placing a PEG relatively soon after dysphagia symptoms begin.
- Another reason for placing the PEG early is that progression of respiratory weakness often parallels the dysphagia. When respiratory function is better, the procedure can be done with less difficulty and less risk of respiratory problems immediately following tube placement.

Q & A

- ***Q:*** Can you still eat and drink if you have a PEG tube?
- ***A:*** The gastrostomy tube creates an alternate route of entry into the stomach but doesn't affect oral intake.
- In the early stages of dysphagia, it can be used to supplement oral intake of food and fluids. Later on, when it's dangerous or impossible to consume any food or drink orally, it can be used as the sole method of nutrition.

PEG and enteral nutrition

- Many readily available liquid pre- parations designed to provide a complete and balanced diet can be used as the sole source of nutrition- ENTERAL NUTRITION.
- Feeding through the tubes placed in the stomach and intestine is useful for providing nutritional support in patients who cannot or will not eat,
- But, who have a functional GI tract !

Refeeding Syndrome

Keys, Henschel & Taylor, Am J Physiol 1947

"The heart is closer to failure during recovery than in starvation"

Plasma Insulin	**postabsorptive**	**1-5 µU/ml**
	absorptive	**30-50 µU/ml**
	parenteral nutrition	**80-100 µU/ml**

Refeeding

K^+, Mg^{++}, phosphat into cell

Fluid imbalance

Sodium imbalance

PEG

- To optimize the outcome, physicians must pay attention to the folowing key issues:
- proper patients selection
- proper device selection
- proper placement technique
- proper follow-up

Proper placement technique
P E G

Since its first introduction in 1980 (Gauderer & Ponsky), PEG has been increasingly adopted as a safe and cost-effective method for enteral nutrition that is well tolerated by patients.

Proper placement technique PEG

- PEG is technically easier than surgical gastrostomy, performed more rapidly, usually within 15-30 min,
- PEG is performed with the use of only local (xylocain) and intravenous sedation (midazolam)
- PEG is less expensive than surgical gastrostomy

Proper placement technique PEG

- The “pull” technique was the one originally described, and probably remains the most popular
- The “push” technique (Seldinger technique) - simpler method involves pushing the feeding tube through abdominal wall
- The “introducer” technique

PREPARATION OF THE PATIENT

- Operative consent from the patient
- Nothing by mouth for 8-12 hr
- iv. line to administer meperidin & midazolam
- Remove dentures

PREPARATION OF THE PATIENT

- There is no consensus as to the general benefit of prophylactic use of antibiotics, so that most centres provide antibiotic prophylaxis only in patients with a particularly high-risk profile (cephalosporin)

Löser Chr, Keymling M. Antibiotic prophylaxis during placement of percutaneous endoscopic gastrostomy tubes (PEG tubes). Z Gastroenterol 2000; 38: 271-273

Contraindications

- Absolute

  - Negative diaphanoscopy
  - Coagulopathy and thrombocytopenia
  - Peritonitis
  - Obstructing ileus

Success rate

- Most large statistical analyses report a success rate of 99%, with a method-related mortality rate close to 0%, assuming that the contraindications are observed

- In patients who have undergone prior gastric surgery (B I or B II resection, total gastrectomy etc.) the success rate is reduced to approximately 80%

Foutch PG,et al. Am J Gastroenterol 1988; McCarter TL, et al. Am J Gastroenterol 1998; Löser Chr., et al. Nutrition 2003

POSTPROCEDURE

- Nothing by mouth 1-2 hr.
- Flush the tube with tap water every 24 hr.
- Change the gastrostomy site dressing and clean the wound with hydrogen peroxide for 3 days. Remove sutures after 7 days
- Inform the patient and his family about further managament of the tube

---

patients can ingest nutrients orally immediately after removal of a PEG system and that the puncture canal heals rapidly, without complications, when covered externally with a sterile compress

PEG COMPLICATIONS

PEG tube placement is associated with:

- Mortality of 0,5% - 1.0%. The 30-day mortality after PEG is 10-30% (progression of underlying disease)
- Peristomal leakage with peritonitis, necrotizing fascitis and gastric hemorrhage in 1% and
- Minor wound infections, stomal leaks, tube extrusion or migration, aspiration, ileus, gastrocolic fistula, fever in 8% of patients

NG vs. PEG in pts with neurological disorders

N=30

	PEG N=16	NG N=14
Mortality 6 weeks	2(12%)	8(57%)*
Prescribed food intake	100%	78%**
Albumin	↑	↓
Stop feeding (n pts.)	0	3

* $p<0.05$, ** $p<0.001$

Norton et al, Br Med J, 1996

Follow-up

- Long-term follow-up studies show that tube failure requiring tube replacement occurs in 8 to 33% of patients

Wassef W. Curr Opin Gastroenterol 2003.

Lijec Vjesn 2003; 125(11-12):292-5

Percutaneous endoscopic gastrostomy: 5-year experience at our center (1997-2002)

- Brkic T, Pulanic R, Krznaric Z, Opacic M, Rustemovic N, Vegar V, Vucelic B, Hrstic I, Cavka SC, Premuzic M, Ostojic R, Kalauz

- PEG was introduced as a routine method in the Division of Gastroenterology, University Hospital Rebro, Zagreb, in 1995.
- We inserted PEG in 86 patients from January 1, 1997 until January 31, 2002.
- The most frequent indication for PEG insertion was a neurological condition (60/86).
- There were no deaths directly related to the procedure.
- Our experience confirms that PEG is a relatively safe and well tolerated procedure.

Conclusions

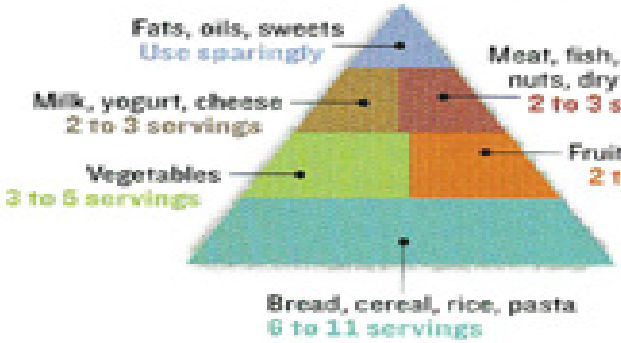
1. PEG is an economical, highly effective and easy to use technique, associated with a low rate of complications, which allows the maintenance of adequate enteral nutrition of patients who are unable to ingest sufficient nutrients orally

2. Clinical studies have demonstrated the high level of acceptance by patients and the marked improvement in nutritional status and general well being of such patients

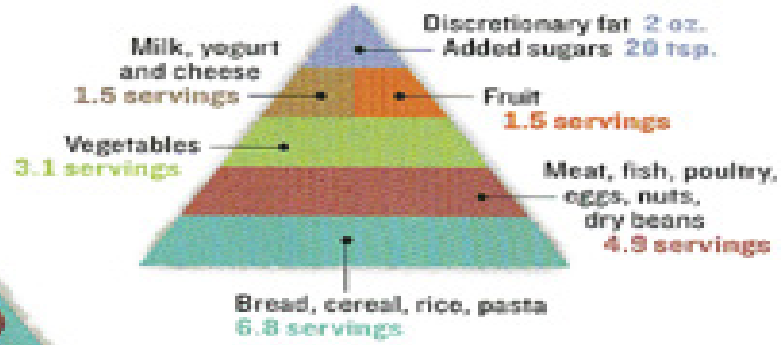
3. In order to prevent deterioration of nutritional status and to improve their overall quality of life, the decision whether or not to use an enteral tube system for feeding should be taken at a much earlier stage

Redesigning the Food Pyramid

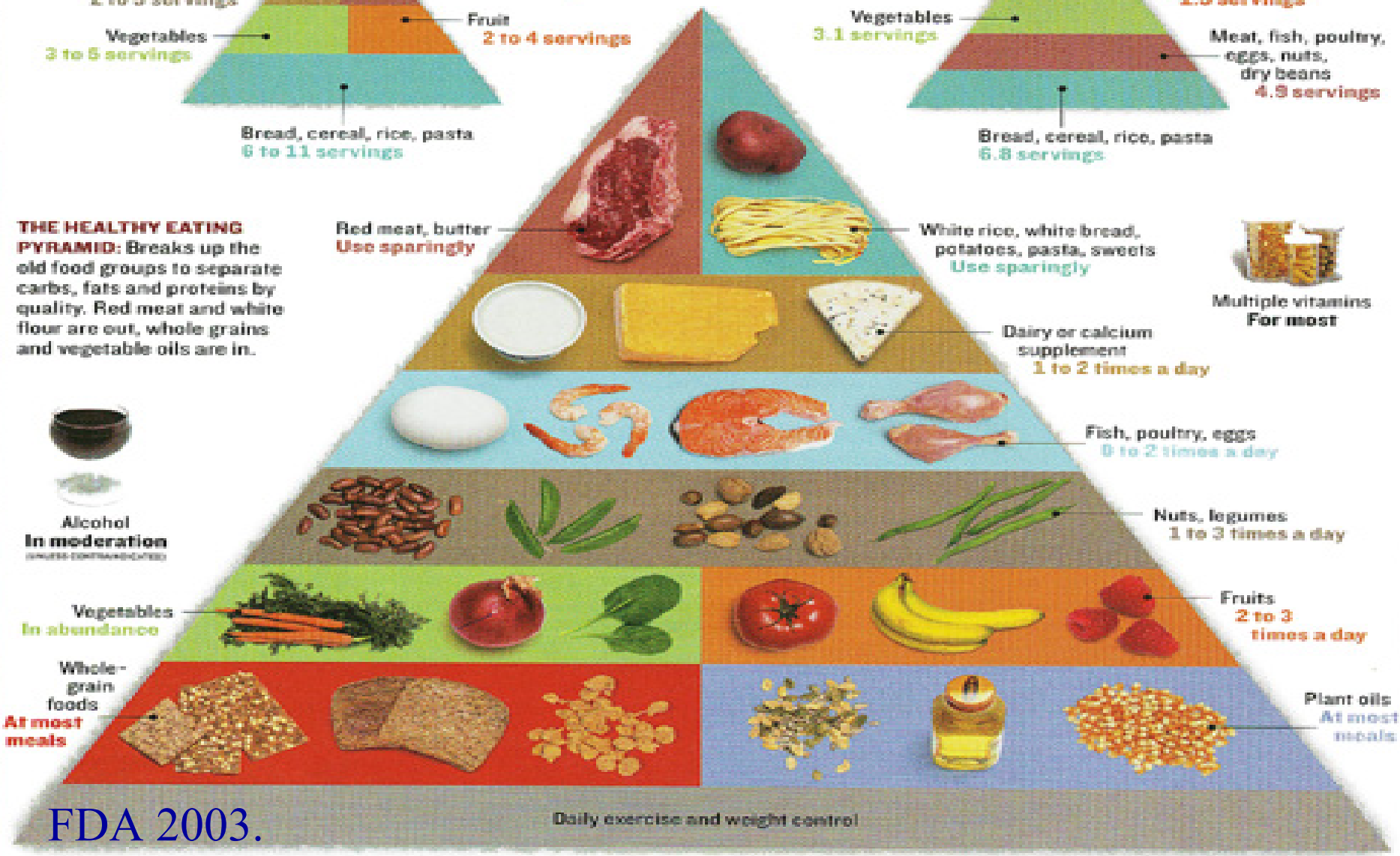
THE FOOD GUIDE PYRAMID: The government's 1992 effort to wean us from fat conveyed a sense that all carbs are harmless.



THE AVERAGE AMERICAN DIET: With a cherry on top. Sweets and meats are a bigger draw than healthful fruits and vegetables.



THE HEALTHY EATING PYRAMID: Breaks up the old food groups to separate carbs, fats and proteins by quality. Red meat and white flour are out, whole grains and vegetable oils are in.



FDA 2003.

ENDOCRINOLOGICAL ASPECTS OF PATIENTS WITH NEUROMUSCULAR DISORDERS

Mr. sc. Darko Kaštelan
Department of Endocrinology
University Hospital Zagreb

NEUROMUSCULAR DISEASES

- weakness
- fatigue
- reduced mobility
- diminished physical working capacity

NEUROMUSCULAR DISEASES - CONSEQUENCES

- obesity
- sindrome X
- osteoporosis

NEUROMUSCULAR DISEASES - CONSEQUENCES

- obesity

- sindrome X

- osteoporosis

OBESITY - OVERWEIGHT

Obesity is a serious condition characterized by excessive body mass, which causes development of clinical complications, reducing quality of life, fitness for work and life expectancy.

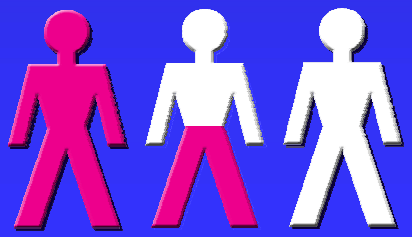
Body mass index = Weight/Height^2

BMI<25 (normal); BMI 25-30 (overweight); BMI> 30 (obesity)

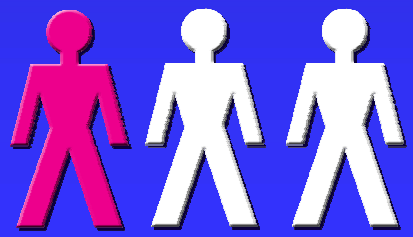
Obesity - epidemia

Over 135 million people from the EU are obese or overweight[1]

More than half of the adult population is overweight[1]



30% of adults are clinically obese[1]



1. International Obesity TaskForce. *Obesity in Europe*. 2002.
2. Presidency of the Council of the European Union. *Diabetes – Information by the Presidency*. May 2004.

OBESITY / DISEASE OF ALL GENERATIONS

- prenatal
- childhood
- adolescence
- adults
- senior age

OBESITY DEVELOPMENT FACTORS IN ADULTS

- ⇓ BASAL METABOLISM -1% per year
- LIFESTYLE CHANGES:

  LACK OF PHYSICAL ACTIVITY

  SITTING AT WORK

- DISEASES
- BAD EATING HABITS

APPETITE REGULATION

PSYCHOLOGICAL FACTORS

AFFERENT NERVES (VAGUS)

PEPTIDES
INTESTINE
CCK

NPY
MCH
AgRP
OREXIN

MSH
CART
GLP-1
SEROTONIN

CULTURAL FACTORS

HORMONS
LEPTYN
INSULIN
CORTIZOL

METABOLITES
GLUCOSE
KETONES

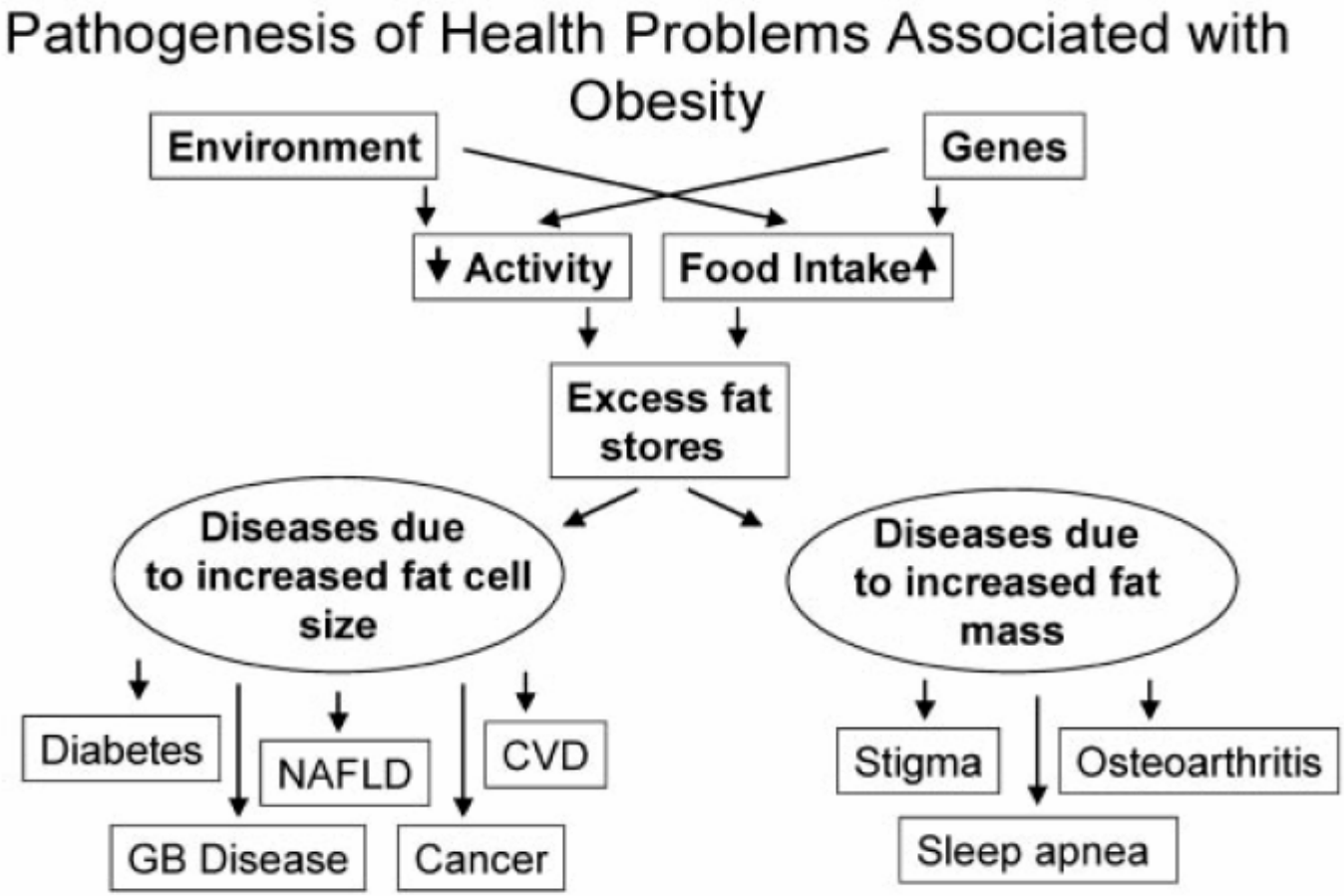
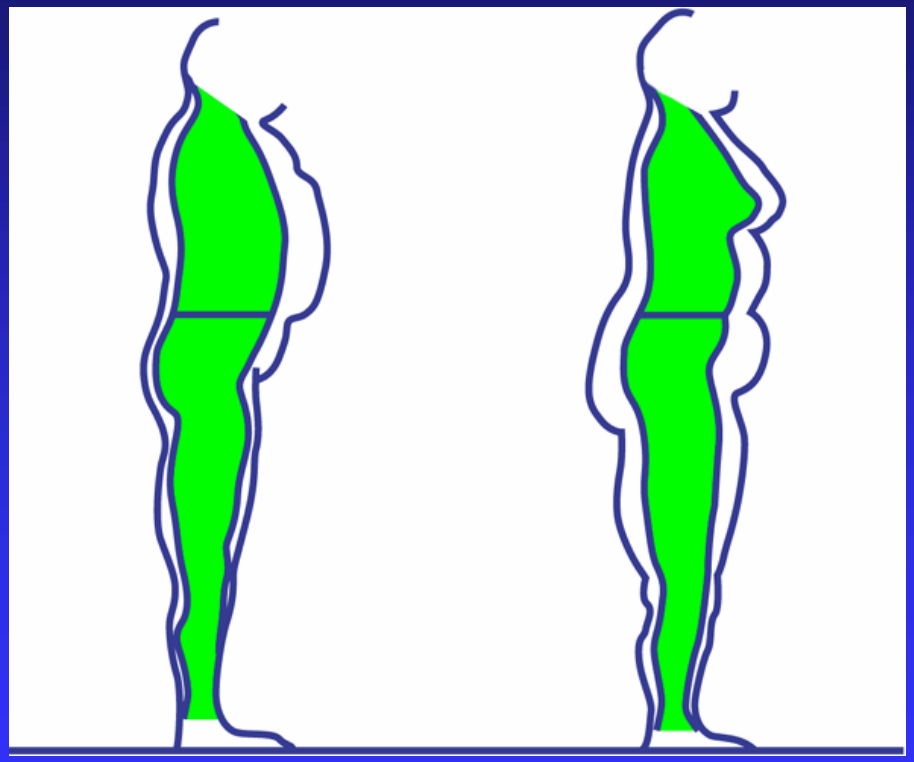


FIG. 1. The pathology of obesity produces the myriad of health related problems. These health-related problems can be attributed to either the increased mass of fat or the increased release of peptides from enlarged fat cells. CVD, Cardiovacular disease; GB, gallbladder.

ANDROGENIC AND GYNOIDAL FAT TISSUE DISTRIBUTION

Central fatness (the apple shape) stemming from visceral fat tissue



Lower body fatness (the pear shape) stemming from subcutaneous fat tissue

Androgenic fat tissue distribution

Gynoidal fat tissue distribution

FIG. 1. Differences in metabolic characteristics in MHO individuals and at risk obese individuals.

NEUROMUSCULAR DISEASES - CONSEQUENCES

- obesity
- sindrome X
- osteoporosis

SYNDROME X

- insulin resistance
- dislipidemia
- obesity
- hypertension

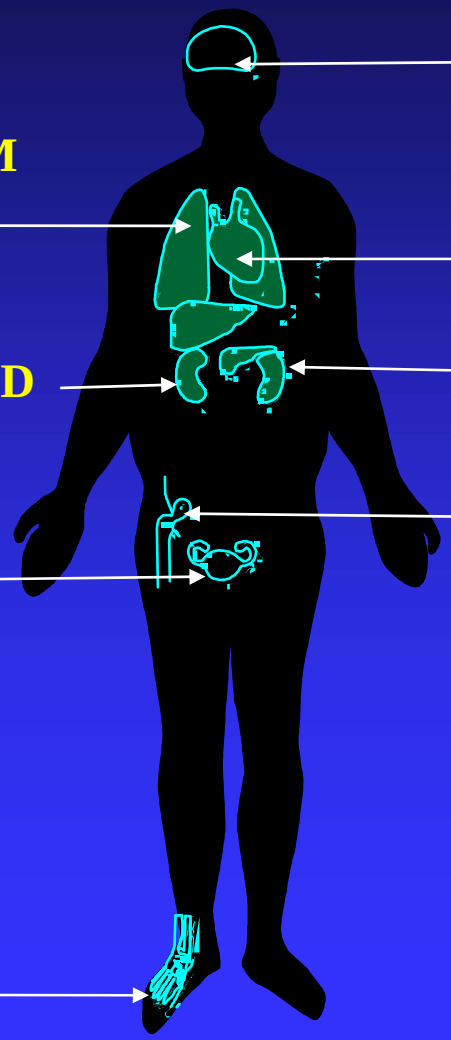
Identifying a patient with insulin resistance

Metabolic syndrome criteria:

- Abdominal obesity:
  > 102 cm men, > 88 cm women
- Fasting plasma glucose > 6.1 mmol/L
- Blood pressure > 130/85 mmHg
- HDL cholesterol:
  < 1.04 mmol/L men,
  < 1.29 mmol/L women
- Triglycerides > 1.69 mmol/L

1. National Institutes of Health. *Third Report of the National Cholesterol Education Program Expert Panel on Detection, Evaluation and Treatment of High Blood Cholesterol in Adults (Adult Treatment Panel III)*. National Institutes for Health, 2001.

CLINICAL CONSEQUENCES OF OBESITY



STROKE

RESPIRATORY SYSTEM DISEASES

CARDIOVASCULAR DISEASES

DIABETES

DISEASES OF LIVER AND GALL DUCT

OSTEOARTHRITIS

HORMONAL DISORDERS

MALIGNANT DISEASES

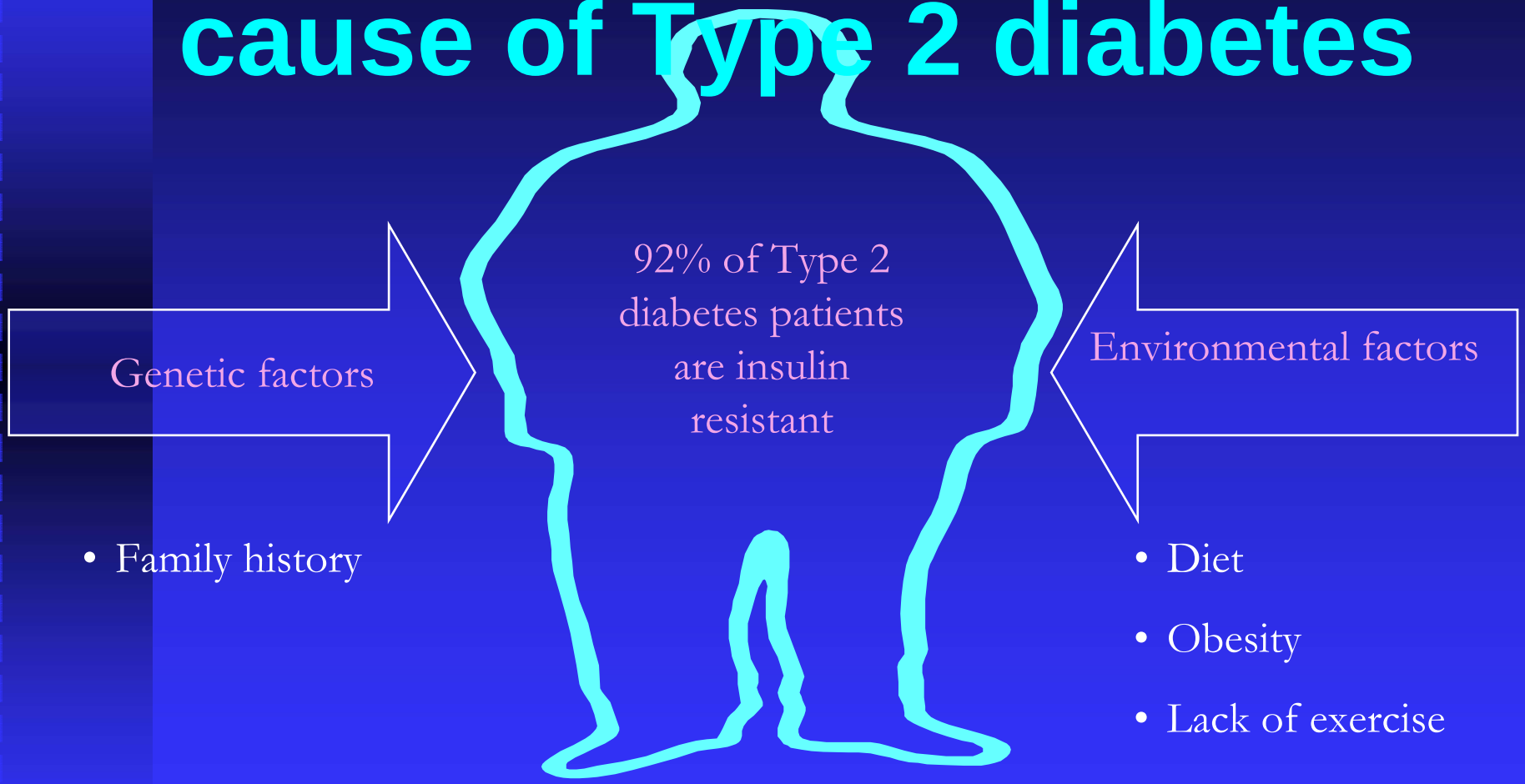
HYPERURICAEMIA

PREVALENCE OF OBESITY – RELATED DISEASES

type 2 diabetes	61%
hypertension	17%
coronary disease	17%
gall bladder disease	30%
osteoarthritis	24%
breast cancer	11%
uterus tumor	34%
colon tumor	11%

Wolf et al. *Obes Res.* 1998;6:97-106.

Insulin resistance is a root cause of Type 2 diabetes

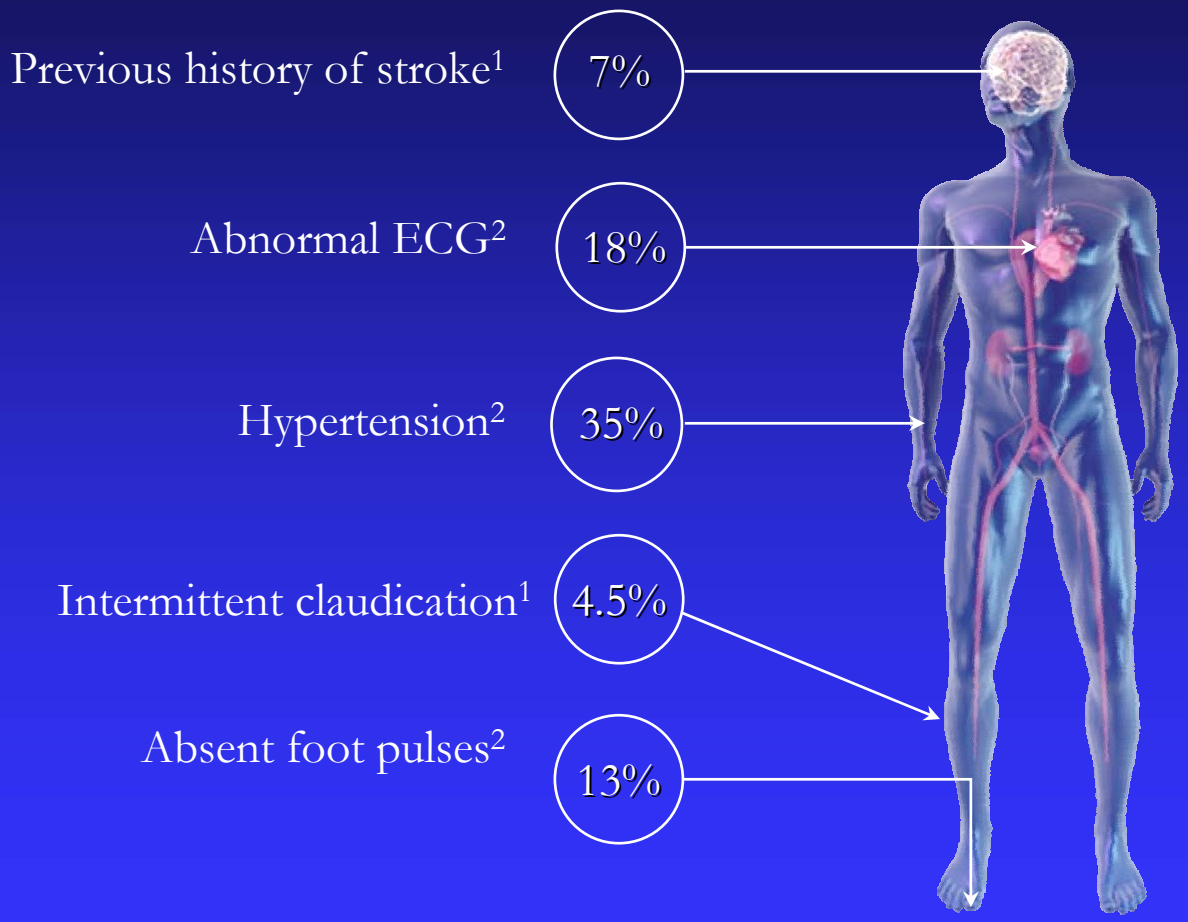


- Family history

- Diet
- Obesity
- Lack of exercise

1. Haffner SM *et al. Diabetes Care* 1999; **22**: 562–568.
2. Bloomgarden ZT. *Clin Ther* 1998; **20**: 216–231.

Type 2 diabetes – the cardiovascular burden at diagnosis



1. Wingard DL *et al. Diabetes Care* 1993; **16**: 1022–5.
2. UKPDS Group. *Diabetes Res* 1990; **13**: 1–11.

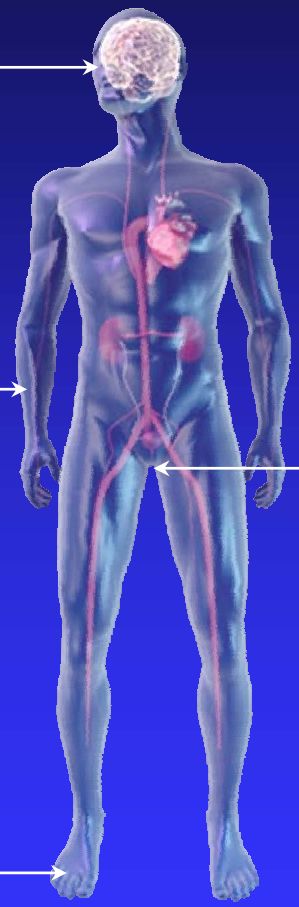
Type 2 diabetes – the microvascular burden at diagnosis

Retinopathy[1] — 21%

Nephropathy[2] — 18%

Erectile dysfunction[1] — 20%

Neuropathy[1] — 12%



1. UKPDS Group. *Diabetes Res* 1990; **13**: 1–11.
2. The Hypertension in Diabetes Study Group. *J Hypertens* 1993; **11**: 309–317.

The impact of Type 2 diabetes

Patients are at risk of:

75% of all deaths in Type 2 diabetes are due to cardiovascular disease[1]

Diabetes is the leading cause of blindness in people of working age[1]

Overt nephropathy occurs in approximately 25% of people with Type 2 diabetes[1]

People with diabetes are 15 times more likely to undergo amputation than the general population[1,2]

1. King's Fund. *Counting the cost: The real impact of non-insulin dependent diabetes*. British Diabetic Association, 1996.
2. Stamler J *et al. Diabetes Care* 1993; **16**: 434–444.

OBESITY AS A DISEASE

- A chronic problem that requires lifelong treatment

OBESITY TREATMENT

PATIENT`S DESIRE:

ideal body mass - unfeasible

CONSEQUENCE:

cyclic weight fluctuations
disappointment

OBESITY TREATMENT

OBJECTIVE

To decrease the health risks stemming from obesity!

ASSUMPTIONS

- Moderate body mass reduction
- Lifelong treatment

RESULT

- increased self - esteem
- improved quality of life
- longer life expectancy

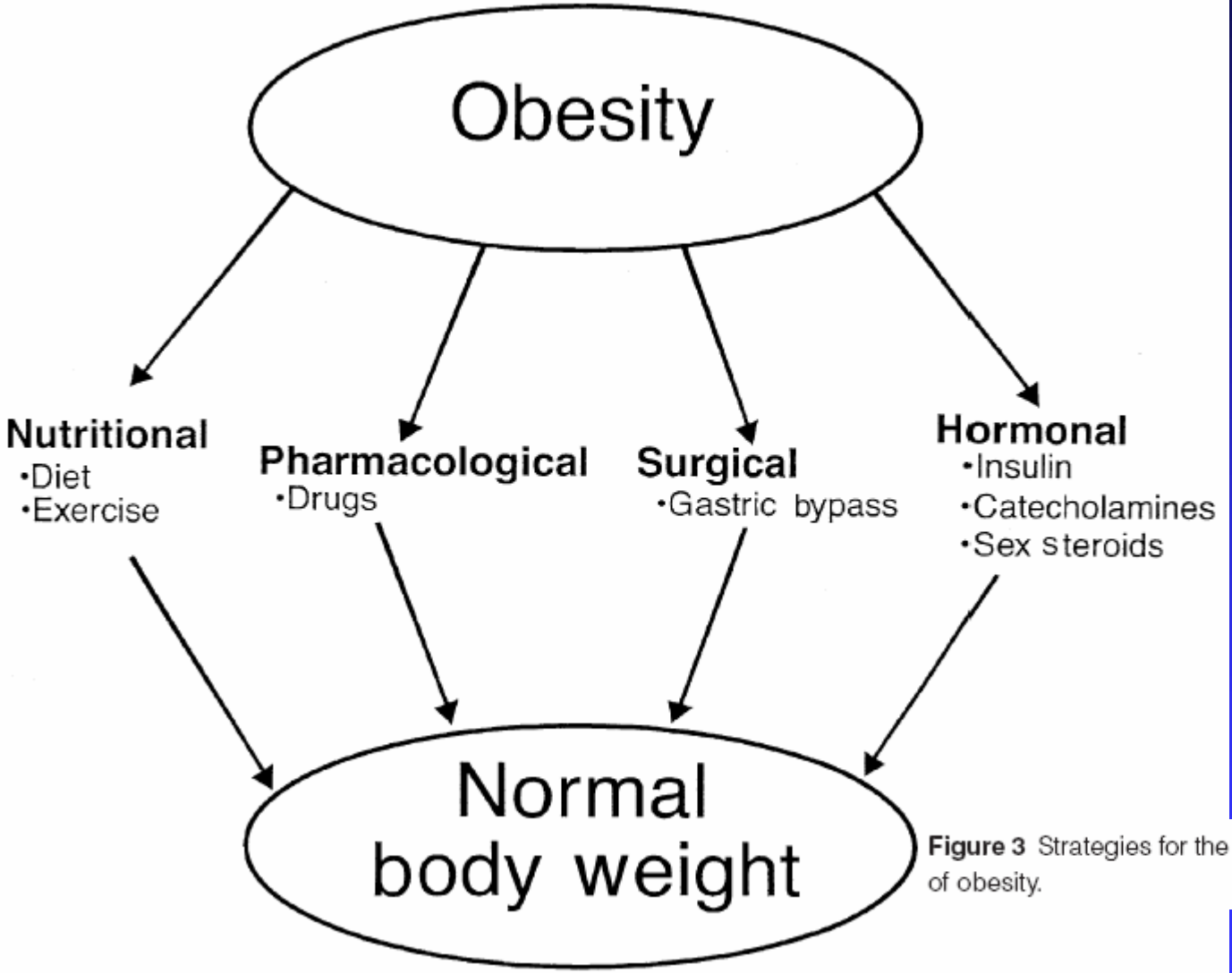


Figure 3 Strategies for the treatment of obesity.

NEUROMUSCULAR DISEASES - CONSEQUENCES

- obesity

- sindrome X

- osteoporosis

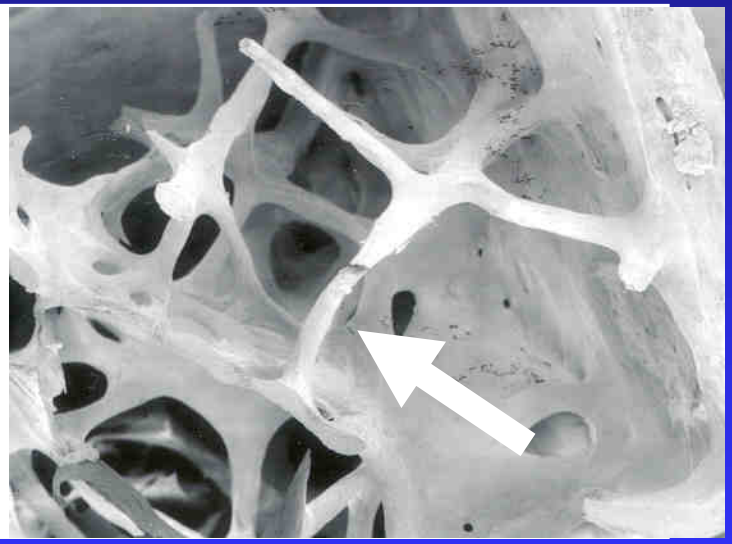
Osteoporosis Defined
World Health Organization (WHO)

- Systemic skeletal disease characterized by:
  - Low bone mass
  - Microarchitectural bone tissue deterioration
  - Decreased bone strength
  - Increased fracture risk

Osteoporosis Defined

Bone strength reflects bone density and quality[1]

Normal Bone



Osteoporotic Bone

WHO Diagnostic Criteria for Osteoporosis

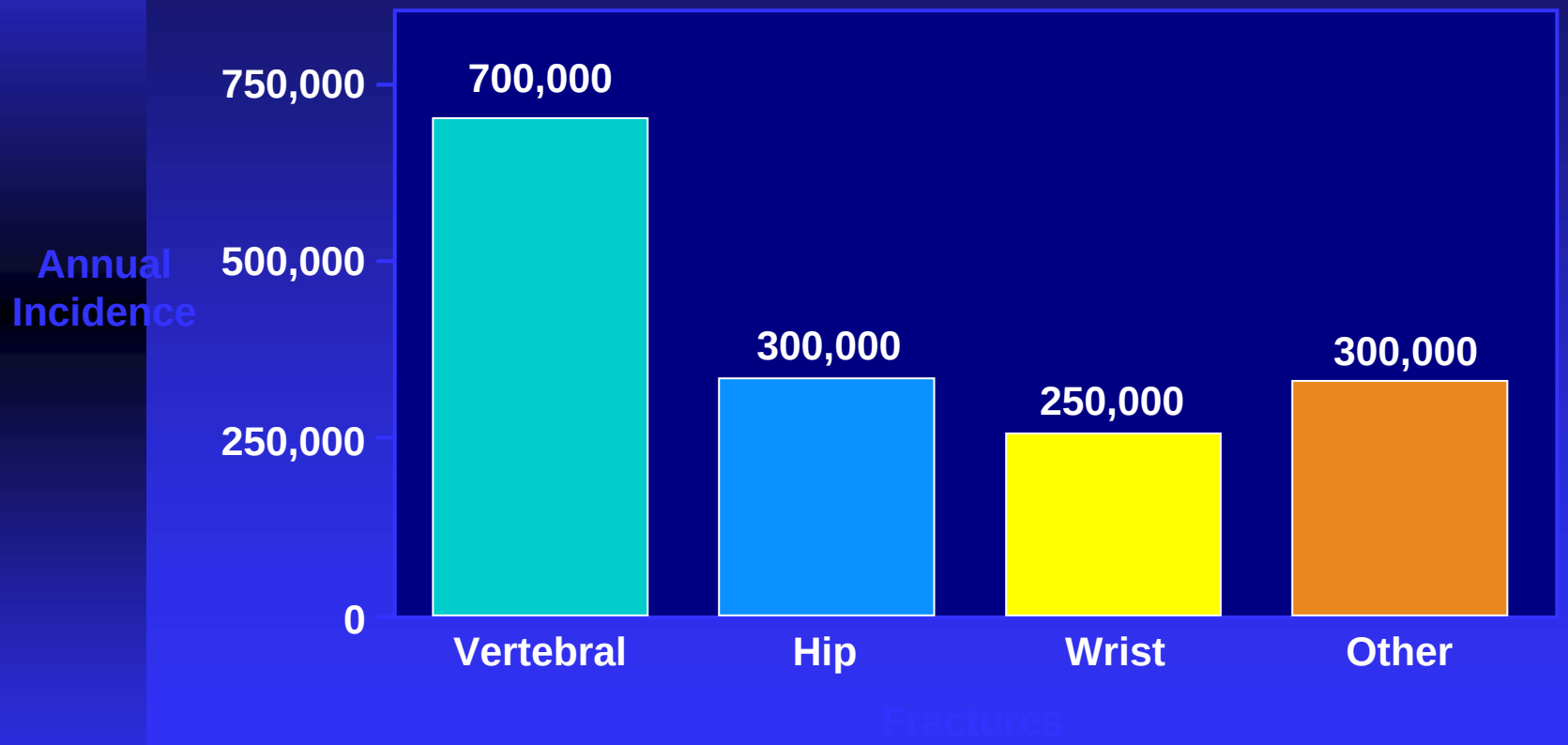
A reduction by 1 SD equals a 10 - 12% decrease in bone density[1]

Diagnosis	BMD: Number of SD Below Healthy Young Mean
Normal	No lower than 1
Osteopenia	Between 1 and 2.5
Osteoporosis	2.5 or more
Severe osteoporosis	2.5 or more with fragility fractures

Risk factors for osteoporosis in patients with neuromuscular disorders

- Sedentary lifestyle
- Housebound (hypovitaminosis D)
- Glucocorticoid therapy

Osteoporotic Fractures

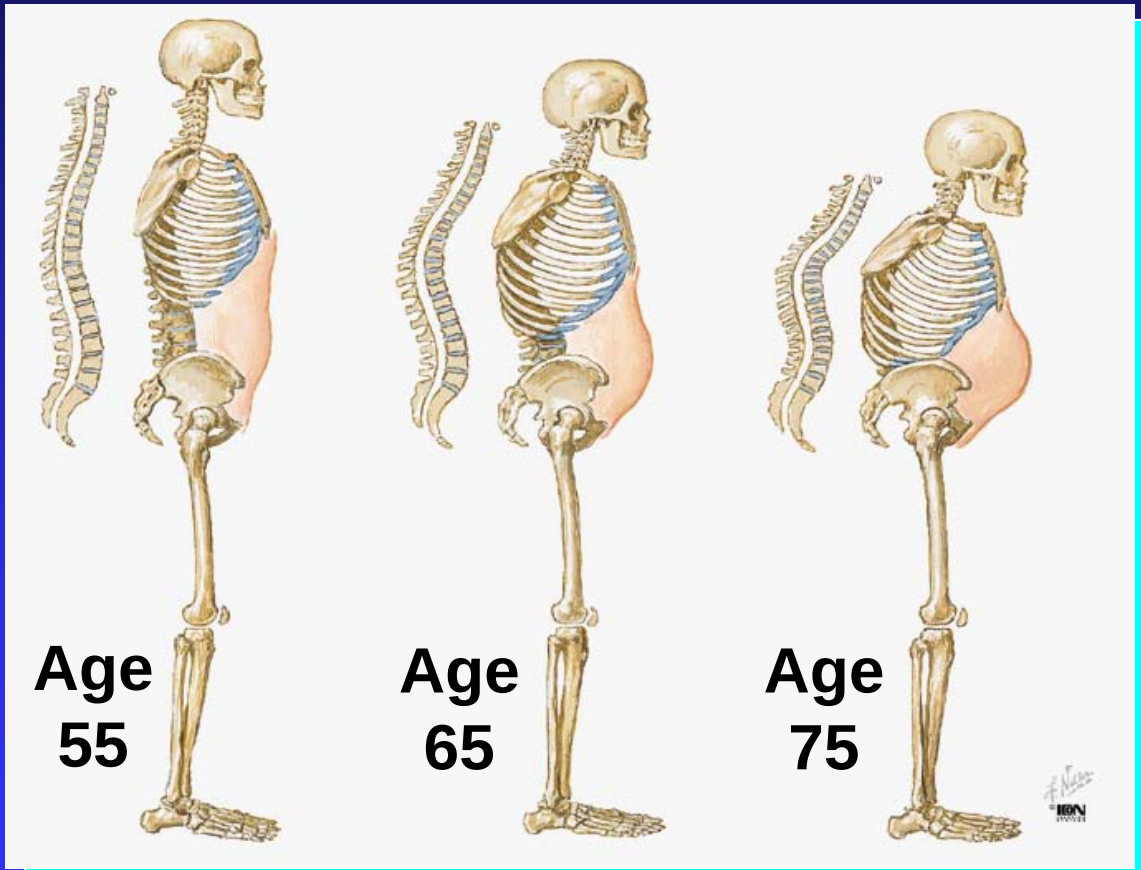


http://www.nof.org/osteoporosis/stats.htm

Consequences of Vertebral Fractures

- Loss of height and kyphosis
- Acute and chronic back pain
- Gastrointestinal and pulmonary dysfunction
- Depression and loss of self-esteem
- Severely limits of daily activities

Progressive Spinal Deformity in Osteoporosis



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Hip Fracture Impact

- Acute
  - Hospitalization and surgery usually required
- Chronic
  - Up to 50% unable to walk independently
  - Approximately 1/3 require nursing home care
- Mortality
  - 20% at 1 year post-fracture

Bone Mineral Density (BMD) Measurement: Full-Body DXA

- Predicts fracture risk[1]
- "Gold standard" for BMD[2]
- High precision, accuracy[1]
- Low radiation exposure[1]
- Measures spine, hip, total body[1]
- Outpatient procedure

1. Genant HK, et al. *J Bone Miner Res*. 1996;11:707-30.

Treatment of osteoporosis

- calcium and vitamin D
- physical activity
- pharmacological drugs

Conclusions

- obesity, osteoporosis and diabetes are important issues in patient with neuromuscular disorders
- significant morbidity and mortality
- early recognition with appropriate intervention can help prevent morbidity and mortality

REHABILITATION AND WEIGHT PROBLEMS IN PATIENTS WITH NEUROMUSCULAR DISORDERS

eamda

35 th Annual General Meeting

22-25 th September 2005, Croatia

Kovac Ida, MD.M.Sc. Zagreb Croatia

The goals of rehabilitation in patients with NMDs are:

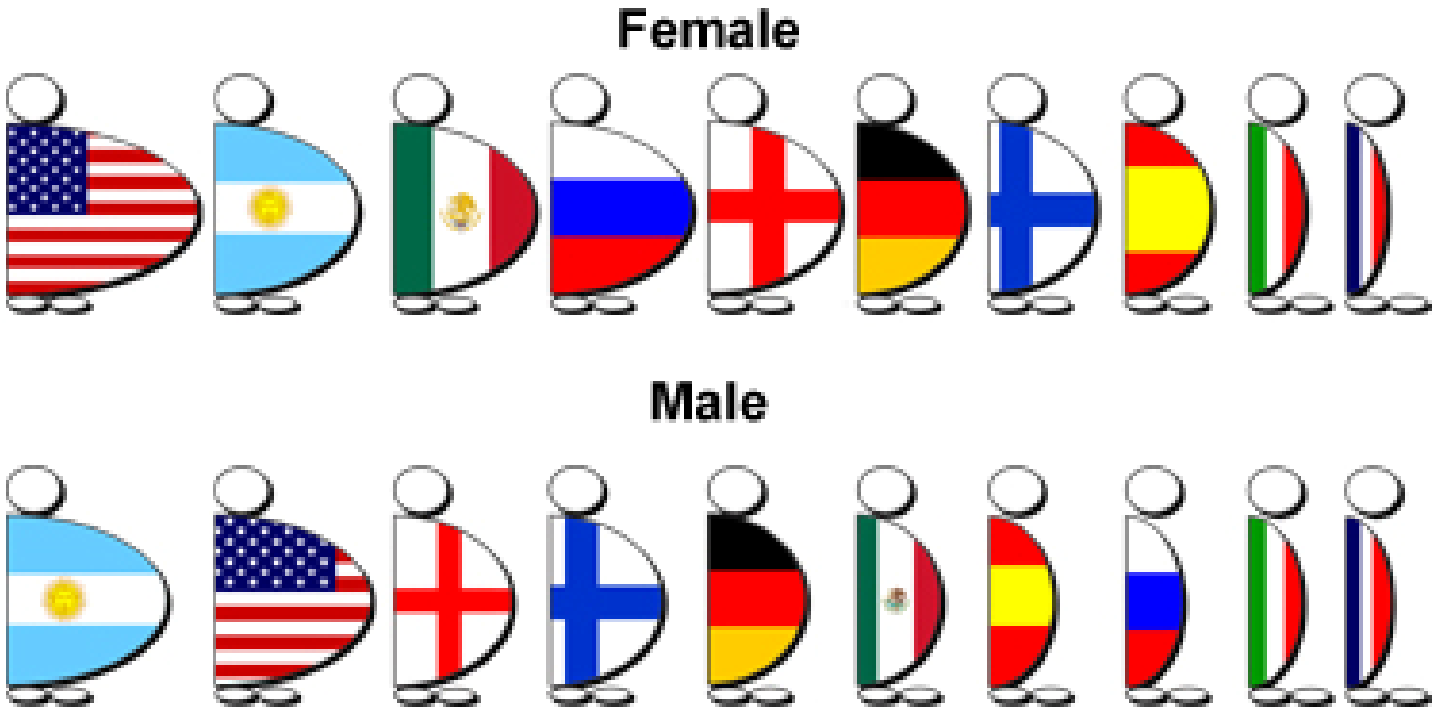
- a) to maximize functional capacities
- b) prolong or maintain independent function and locomotion
- c) inhibit or prevent physical deformity
- d) provide access to full integration into society with a good quality of life

The most frequent problems that negatively impact their quality of life, noted by people with neuromuscular disorders are:

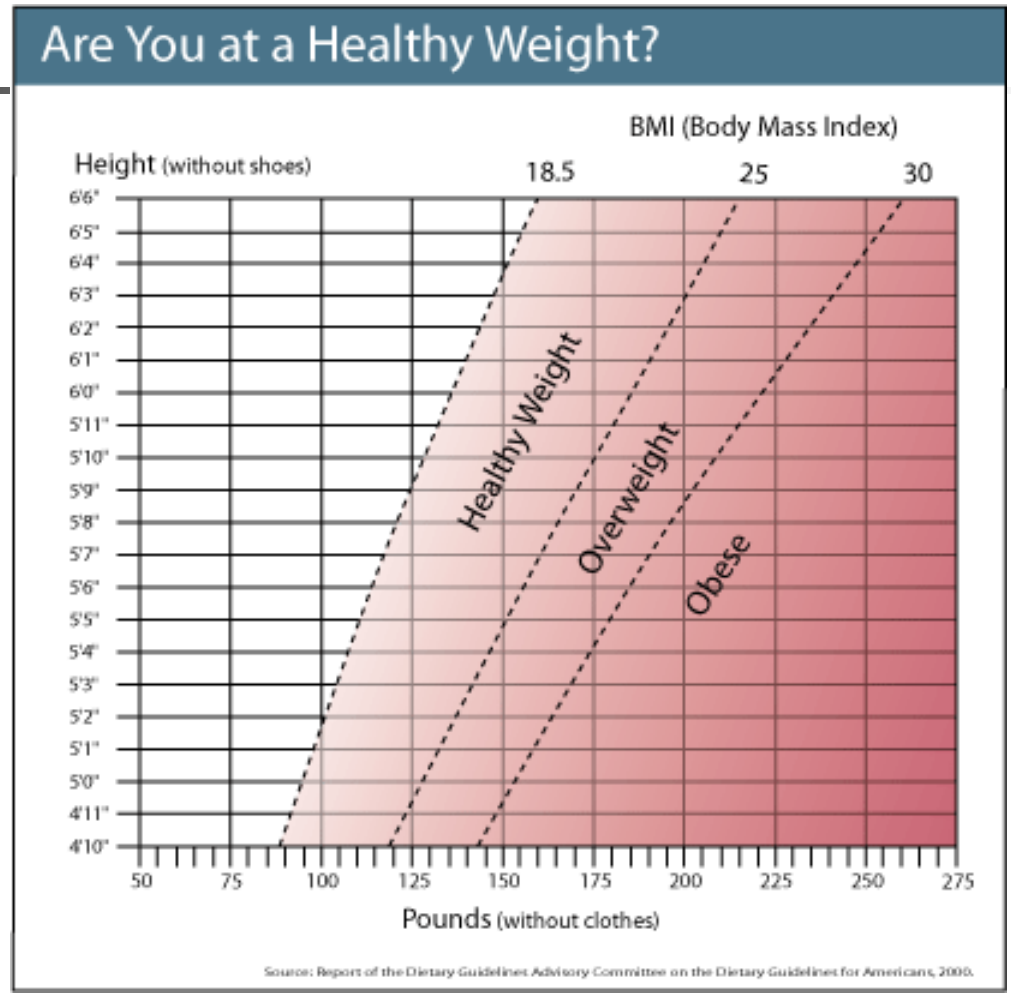
- 1. Weakness
- 2. Difficulty getting exercise
- 3. Fatigue/poor endurance
- **4. Problems with weight**

Craig M. McDonald (2001)

Obesity: A worldwide epidemic



Body Mass Index



INTRODUCTION

- Diminished walking endurance, sedentary lifestyle, and vicious cycle that we have between the disuse atrophy and sedentary lifestyle -
- make people with neuromuscular diseases end up being quite sedentary and
- so, they are prone to overweight.

Body weight- frequently is a problem,

- because it results in an increased muscular demand during movement,

  and
- biomechanical inefficiencies.

WEIGHT PROBLEMS IN NEUROMUSCULAR DISEASE

- 1) **Obesity** (sedentary)
- 2) **Cacchexia** (end -stage)

Obesity

- In more severe NMDs there is an increasing tendency toward obesity, around or after loss of independent ambulation.

- Obesity is common in NMDs, particularly DMD where a 54% prevalence has been reported

Obesity

43 yrs H 173 cm W 105 kg **BMI**(kg/m2)= **35**

Impairments in Duchenne Muscular Dystrophy which reduce physical activity:

1. Weakness
2. Fatigue
3. Cardiopulmonary involvement
4. Contractures
5. **Excessive weight gain**

The consequences of a high adiposity:

1. Increased muscular demand for movement
2. Biomechanical inefficiency
3. Risk factors for coronary artery disease, diabetes and cancer

Reduced Exercise Performance in NMD: Disuse or Disease?

- I) Reduced functional mass (Disease)
- II) Reduced physical activity
- III) Disuse Atrophy

Persons with NM disease represent a very sedentary deconditioned population

They have:

- a) decreased muscle strength and endurance
- b) body composition changes

  (reduced lean muscle mass, increase in fat mass)
- c) lower work capacity, lower peak VO2 capacity
- d) reduced resting energy expenditure, probably on the basis of reduced metabolically active lean tissue

PHYSICAL ACTIVITY IN SLOWLY PROGRESSIVE NMD (Mc Crory 1998)

Total energy expenditure engaged in physical activity NMD subjects,

comparing to control subjects:

a) reduced for male NMD subjects - 30 %

b) reduced for female NMD subjects - 45%

Active minutes per day

a) reduced for male NMD subjects -50%

b) reduced for female NMD subjects - 75%

PHYSICAL ACTIVITY IN SLOWLY PROGRESSIVE NMD (Mc Crory 1998)

- a) The NMD subjects reported spending less time exercising, formally, than controls
- b) When they did exercise, the exercise was at a light level while controls exercised to the moderate level
- **c) Body fat in these NMD subjects was inversely related to minutes active**

WEIGHT GAIN IN NM DISORDERS

Nutritional implications in corticosteroid therapy of Duchenne Dystrophy include:

- **increased appetite**
- **weight gain**
- linear growth suppresion
- decreased absorption of dietary calcium
- fluid retention

Vicious cycle

- People with NM disorders with mobility impairment may expend less calories and be prone to excessive weight gain.

- The resulting obesity may add to the mobility impairment and reduce physical posibilities.

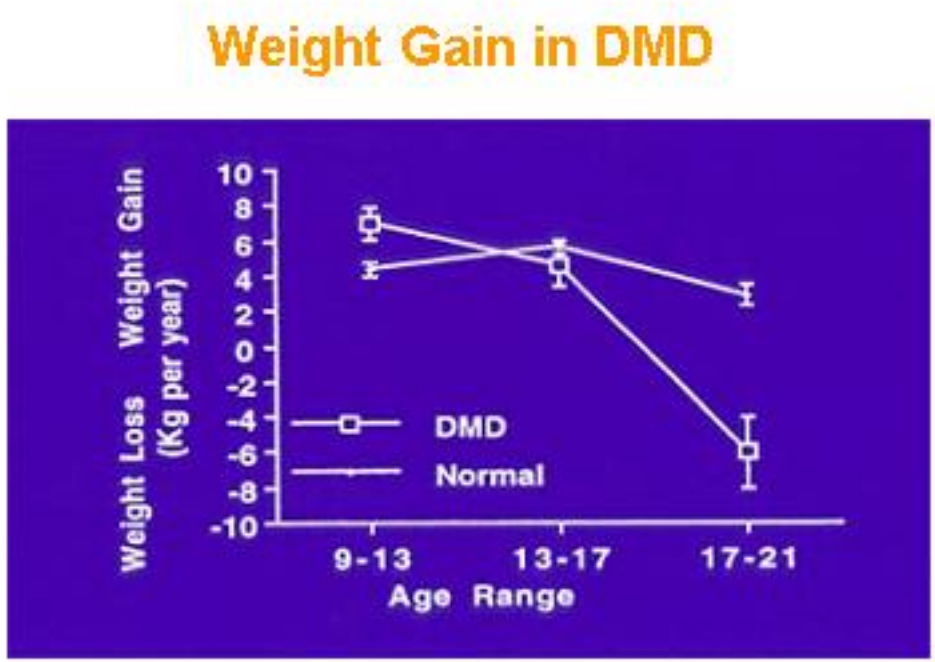
WEIGHT GAIN IN DUCHENNE DYSTROPHY
(Craig M. McDonald, 2001)

- **At 9-13 years** of age, in the transition to the wheelchair, Duchenne Dystrophy boys gain **excessive weight** per year relative to controls.

- **Later, at 17-21 years,** there is actually tremendous **weight loss** in this disorder relative to controls.

WEIGHT GAIN IN DUCHENNE DYSTROPHY

(Craig M. McDonald, 2001)



ADIPOSITAS

Yrs 13 H 151 cm W 51 kg

BMI = 22,4

DMD 10 years

DMD 16 years

Cacchexia

23yrs W 30 kg H 154cm

BMI (kg/m2) = **12,6**

Underweight

BMI (kg/m2) = **13,1**

23 yrs W 38 kg H 170 cm

Nutrition management in NMD:

- Accuracy and consistency of anthropometrics, including body weight and length
- Determination of Ideal Body Weight
- Nutritional implication of corticosteroid therapy
- Osteoporosis and low-impact fractures
- Escalation of nutrition support with progression of muscle weakness
- Obesity with decreased activity
- Nutrition and emotional issues

HELPING AN OVERWEIGHT CHILD- TREATMENT OF WEIGHT PROBLEMS IN EARLY STAGES:

1) The first step:

- TO SET UP A COUNSELING SESSION

2) The second step:

- EXERCISE

3) The third step:

- SUPPORT AND LOVE FROM THE CHILD'S PARENTS

1a) THE FIRST STEP - COUNSELING SESSION

- It is important to counsel both the child and the child's parents

- "Change needs to come in small steps" and should be a combination of: nutrition, exercise and behavioral changes, including encourage physical activity

1b) THE FIRST STEP - COUNSELING SESSION

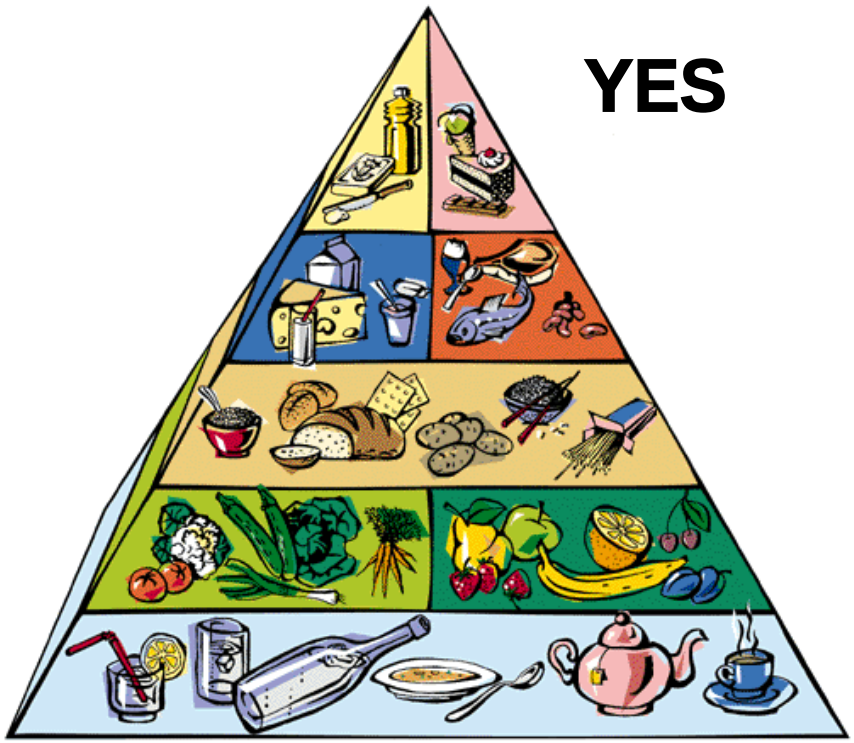
Healthy eating includes:

- establishing a daily eating pattern
- including less fried food
- teaching children about satiety providing healthy snacks, minimizing empty calorie foods,
- and involving the child with grocery shopping and meal preparation

(Broadwater)

1c) THE FIRST STEP - COUNSELING SESSION

when a child with NMD becomes wheelchair dependent



© Société Suisse de Nutrition (SSN)

- Creative ways to reward without the use of food, provide calorie-free or low calorie snacks and adequate after-school supervision- successful management of rapid weight gain
- Nutrition support are: hydration, adequate fiber, protein and energy intake. (Broadwater)

2) THE SECOND STEP IS – EXERCISE !

THE BENEFITS OF OF PHYSICAL ACTIVITY AND EXERCISE THAT HAVE BEEN NOTED IN NEUROMUSCULAR DISEASE ARE:

- 1) increase in cardiopulmonary endurance,
- 2) increase of lean mass
- **3) decrease of fat mass**
- 4) increase of muscle strength and endurance
- 5) improves flexibility
- 6) improves health outcomes such as coronary artery disease, type II diabetes
- 7) psychosocial benefits

3) THE THIRD STEP
– PARENTS LOVE AND SUPPORT

WEIGHT LOSS PLAN:
- some possible practical advices

1) Exercises

- moderate exercise, like walking, lifting weights, using a resistance band, yields positive results for people with slowly progressing muscle diseases.
- "The key is to be more active than you are *currently*," and add a few minutes of walking (or other exercise) every day or every other day.

2) Small Victories Count

- set up a reasonable weight loss goal
- if weight isn't yet a big problem, simply holding steady also counts as a victory
- don't embark on a rapid-loss diet strategy

II) TREATMENT OF WEIGHT PROBLEMS IN ADVANCED STAGES OF NM DISORDERS

- Escalation of Nutrition Support

- Patients tend to lose weight due to dysphagia and increased time and energy to eat
- When nutritional intake is inadequate, there is associated acceleration in breakdown of skeletal muscle
- Malnutrition – a significant problem

II) TREATMENT OF WEIGHT PROBLEMS IN ADVANCED STAGES OF NM DISORDERS

- Escalation of Nutrition Support

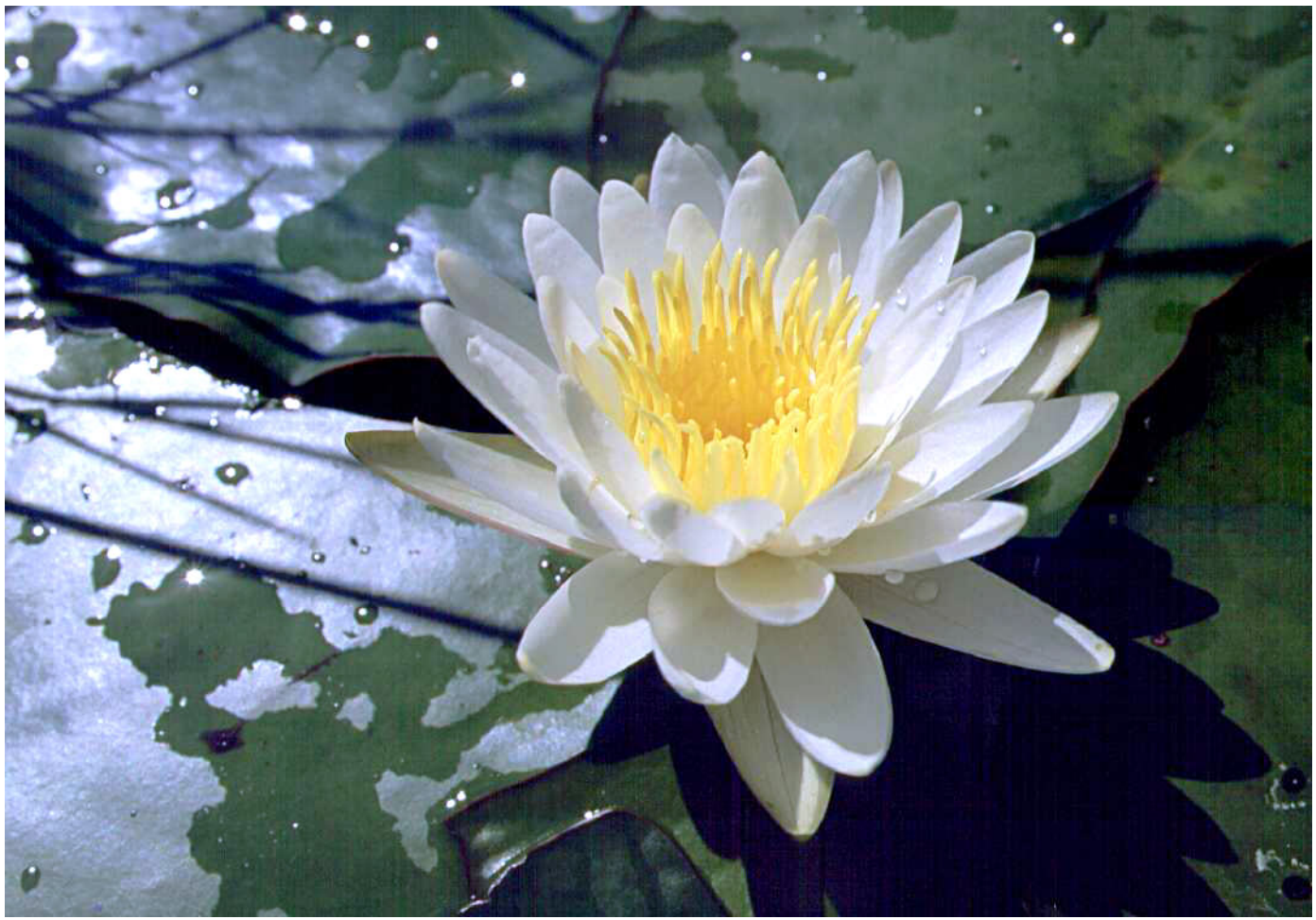
- Careful monitoring of weight and nutritional laboratory parameters to predict when early intervention with nutritional supplements is appropriate
- Nutrition support- hydration, adequate fiber and energy intake and protein intake
- Many patients require the use of oral nutrition supplements or, based on prior patients whishes, enteral feeding with gastric feeding tubes

Conclusions

Problems with (over) weight:

- one of the most frequent problems that negatively impact on quality of life, noted by people with neuromuscular disorders
- great negative impact on the possibillities of physical activity and on rehabilititation posibilities
- it requires adequate attention and treatment
- it would be prudent to involve a nutricionist or dietician, as a part of the rehabilitation team

Thank you for your attention



RENEWAL REHABILITATION PROGRAMS FOR PERSONS WITH NEUROMUSCULAR DISORDERS

Anton Zupan

1993

Unit for rehabilitation of patients with neuromuscular diseases within the Institute for rehabilitation of the Republic of Slovenia

NEUROMUSCULAR DISEASES

- inherited
- chronic
- degenerative
- progressive

DIAGNOSIS

??????????????????????

??????????????????????

??????????????????????

THE GUIDANCE

- patient
- family

THE GUIDANCE

- physical disability
- intellectual capability
- environment

THE GUIDANCE

- childhood
- puberty
- adolescence
- adulthood

CRITICAL PERIODS

- when a child can't start walking
- the loss of walking capability
- respiratory failure and acceptance of a ventilator
- serious cardiac affection
- death

EVERY PATIENT

- **regardless of disability level, must grow up and learn to live on his own**
- **dependence on the help of others !**

NEUROMUSCULAR DISEASES

- muscular weakness
- contractures
- scoliosis
- respiratory insufficiency
- cardiac affection
- nutrition disturbances
- dependence on the help of others
- possible social isolation and psychological problems

PATIENT'S FUNCTIONAL STATE - TEST PROTOCOL (10 tests)

Simple functional movements

- Head and neck
- Upper limbs
- Lower limbs
- Trunk

Complex functional activities

- Intimate d/a
- General d/a
- Locomotion

Vital functions

- Respiratory
- Cardiac

Degree of contractures

- Different joints

MUSCULAR WEAKNESS

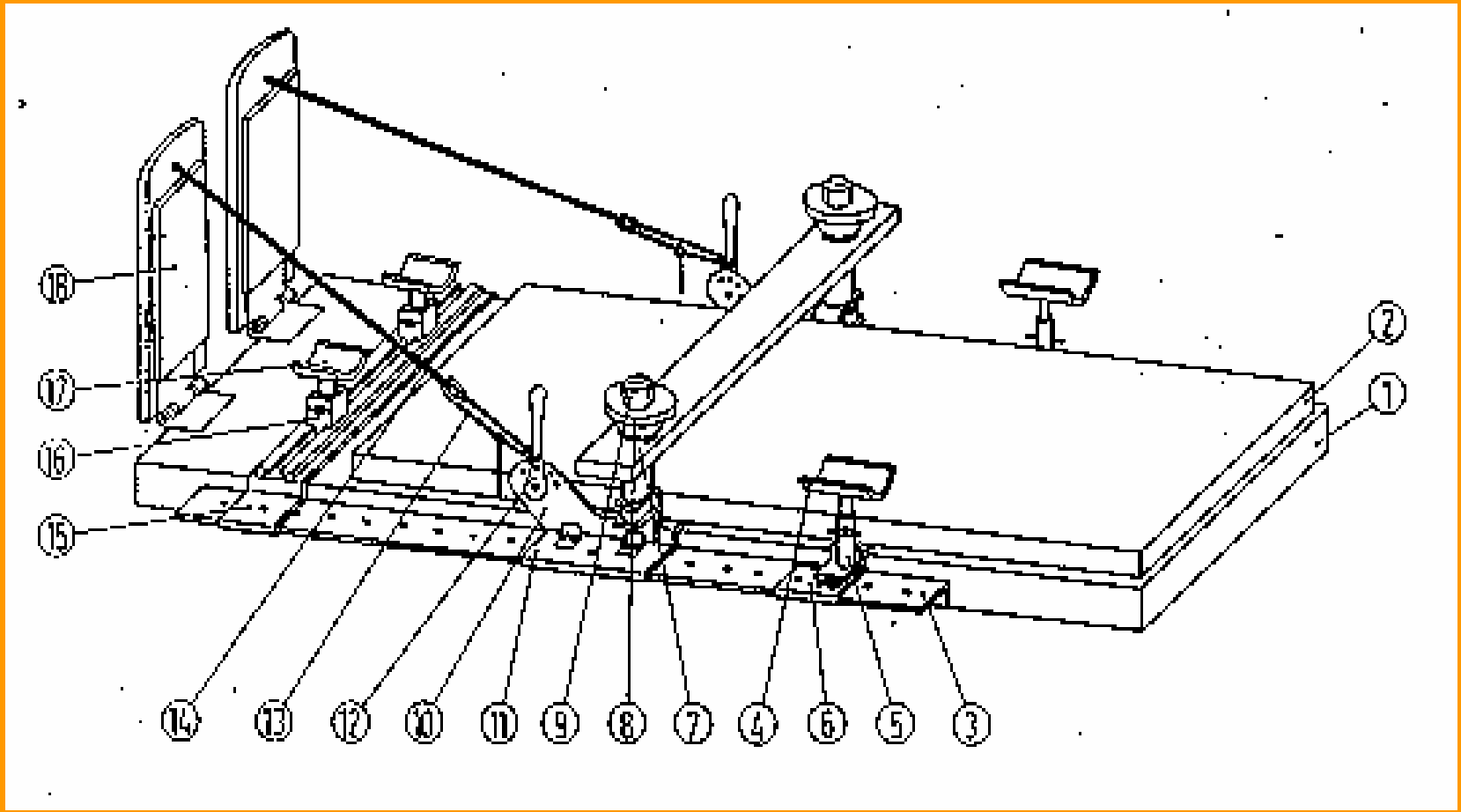
- active or active assisted exercises
- dynamic and isometric
- every day
- in childhood as a game to avoid the feeling of pressure

CONTRACTURES

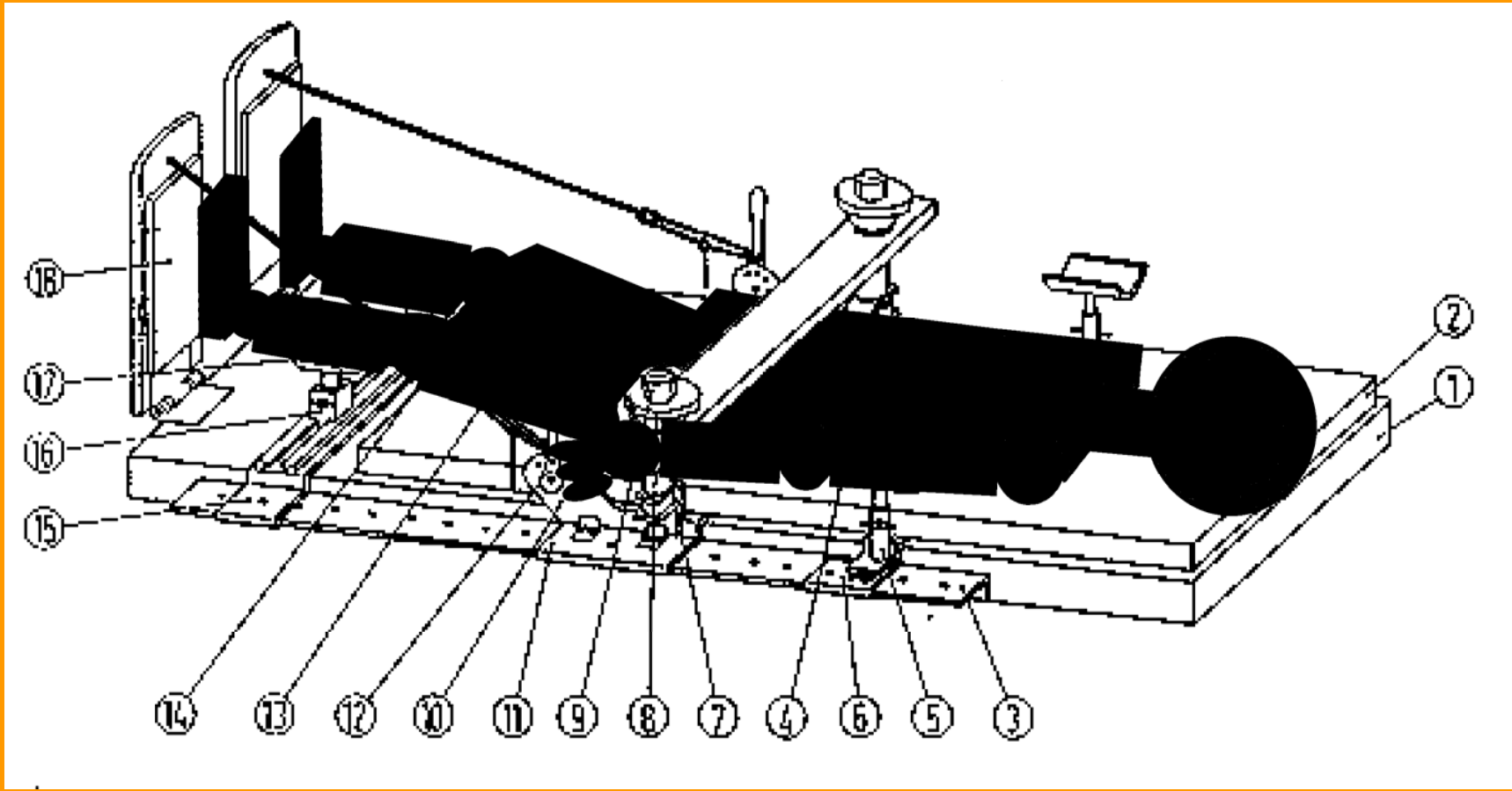
- stretching: every day (2X)
- standing or walking: 3 hours a day
- surgery ?!

manumed

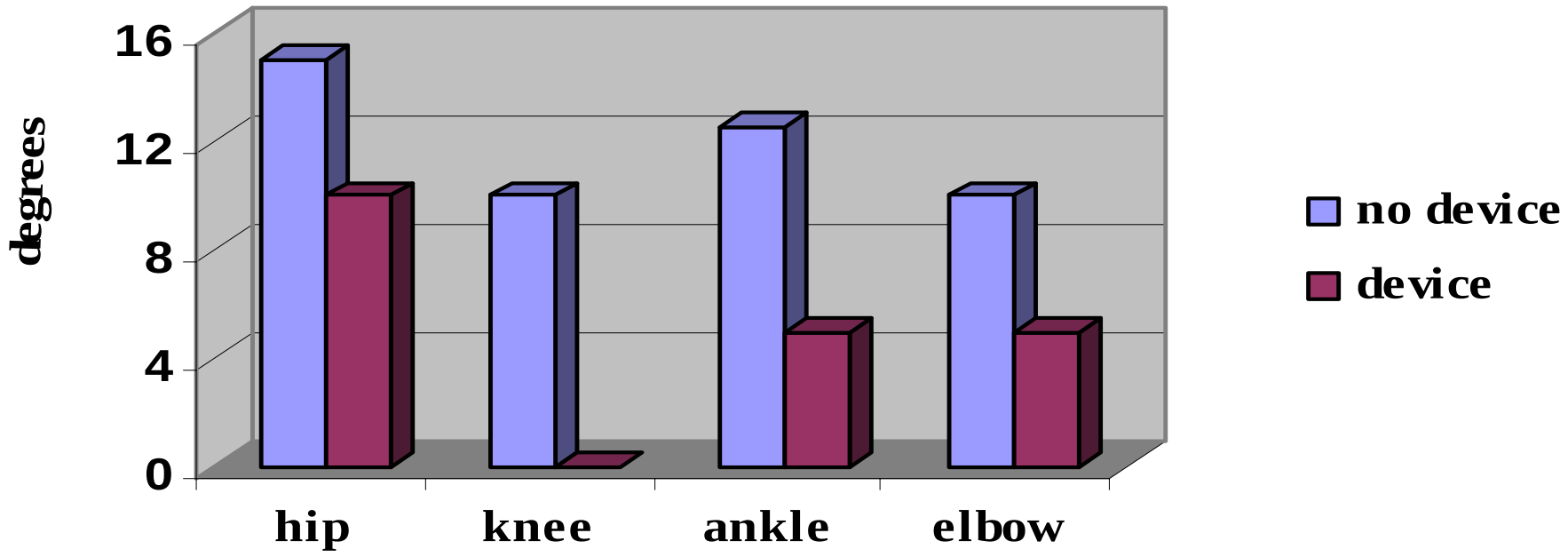
THE DEVICE



POSITION OF PATIENT IN THE DEVICE



AVERAGE PROGRESSION OF CONTRACTURES



SCOLIOSIS

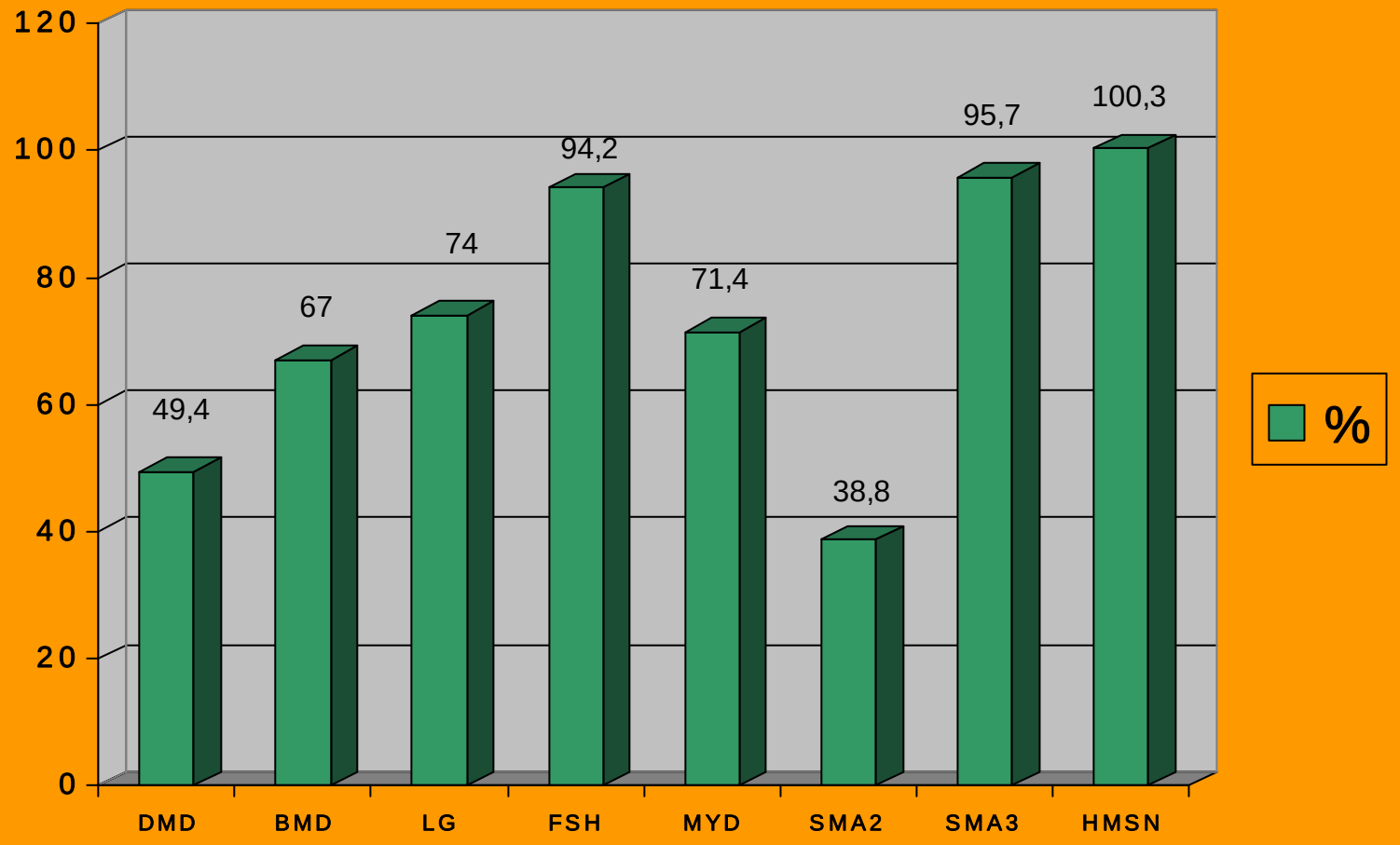
- active exercises
- spinal surgery
- spinal orthosis
- especially made seat

RESPIRATORY PROBLEMS

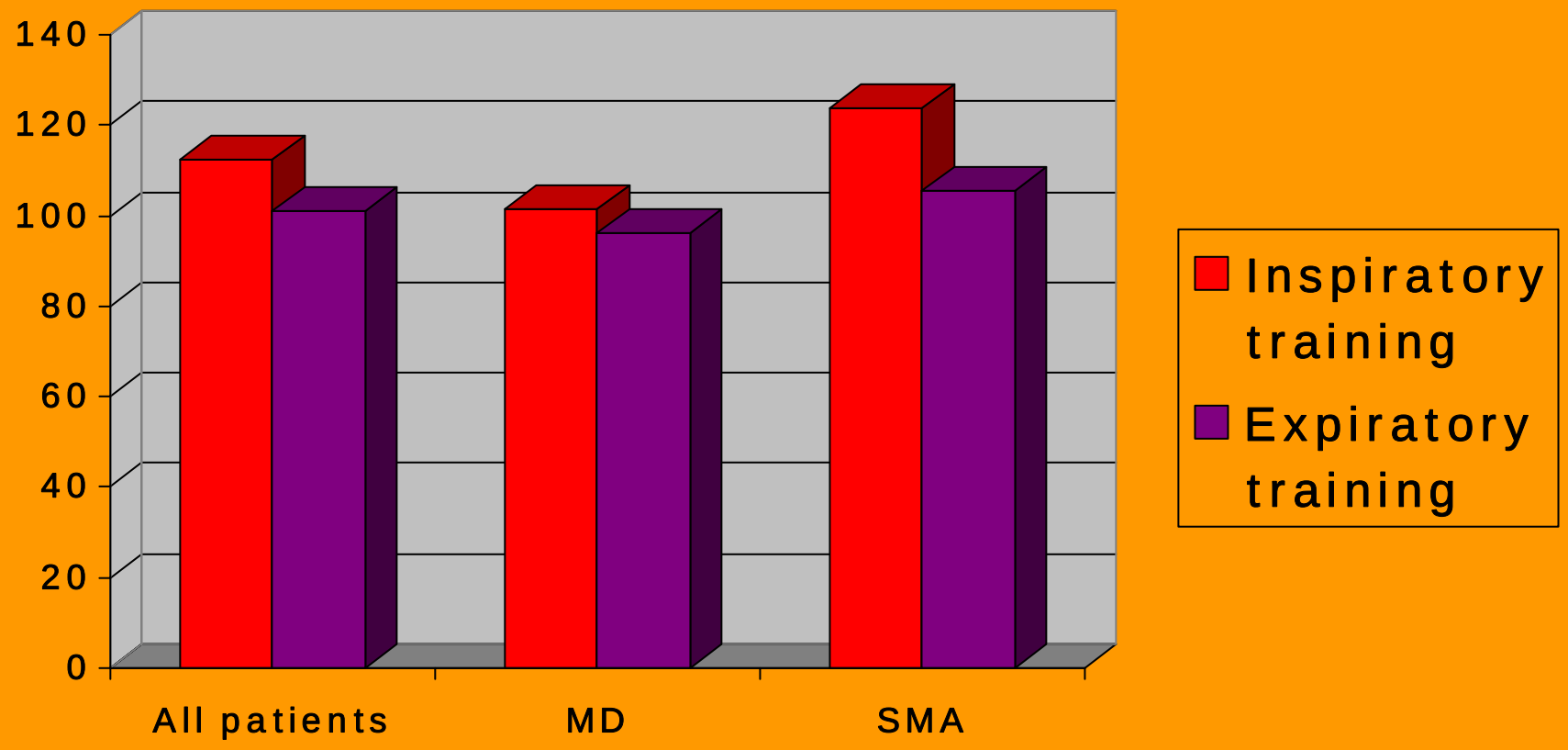
- respiratory exercises
- procedures for cleaning airways (cough machine)
- assisted ventilation

The Human Body

AVERAGE VALUES OF FVC AS A PERCENTAGE (%) OF REFERENCE VALUES



CHANGES OF FVC AFTER INSP. AND EXP. MUSCLE TRAINING AS A PERCENTAGE (%) OF INITIAL VALUES

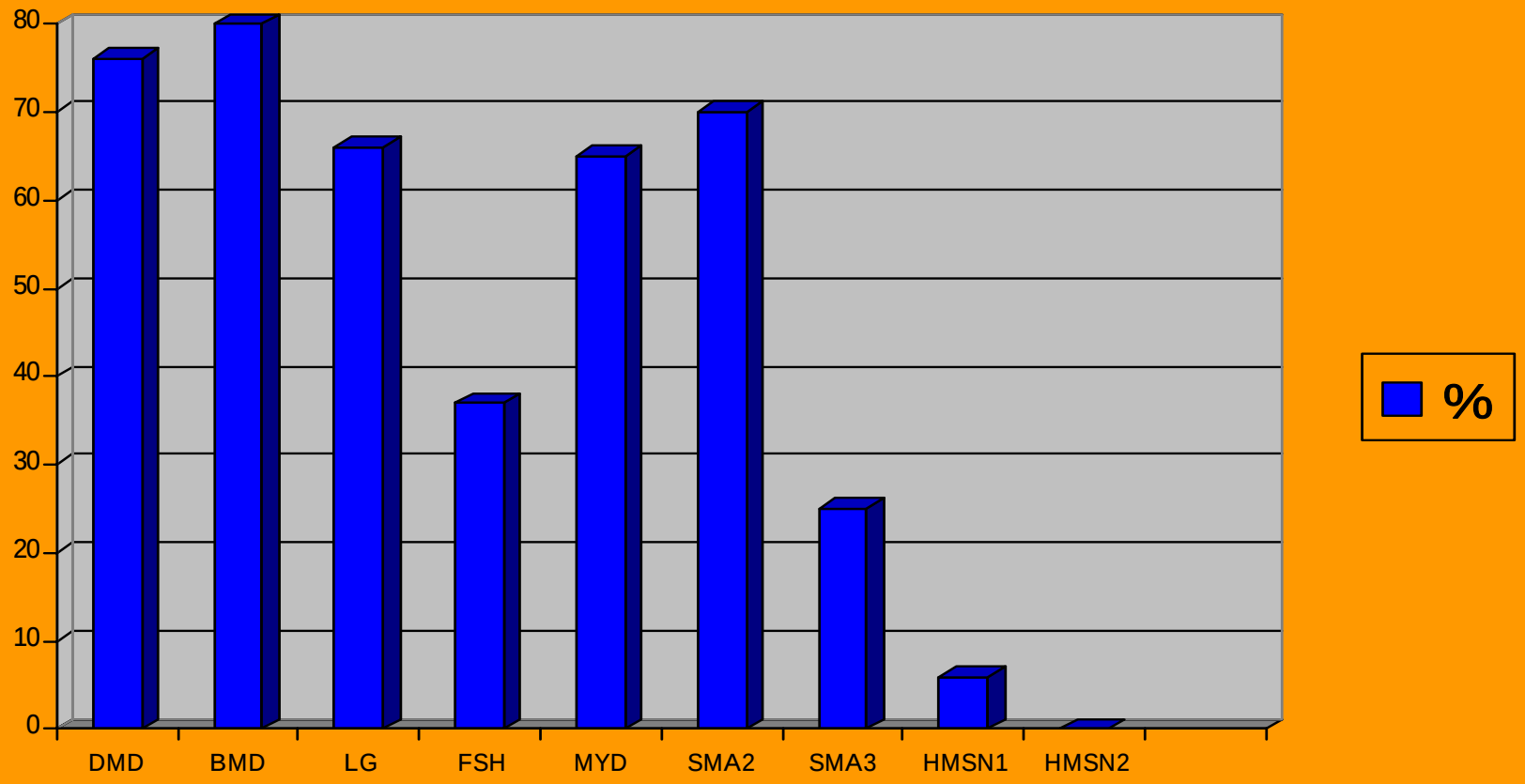


MANUAL CONTROL
EXHALE
INHALE
IN-EXSUFFLATOR

CARDIAC PROBLEMS

- ECG yearly
- Echocardiography
- proper therapy

PERCENTAGES OF ECG CHANGES IN DIAGNOSTIC GROUPS

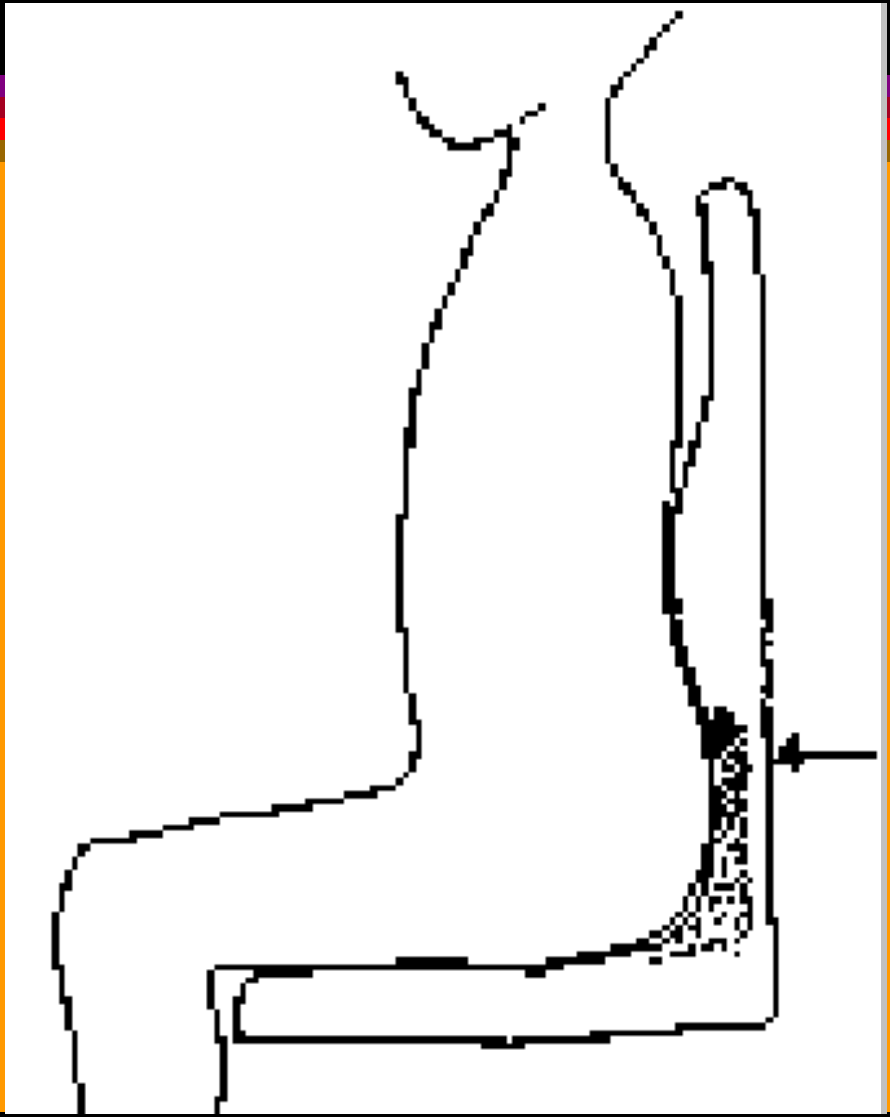


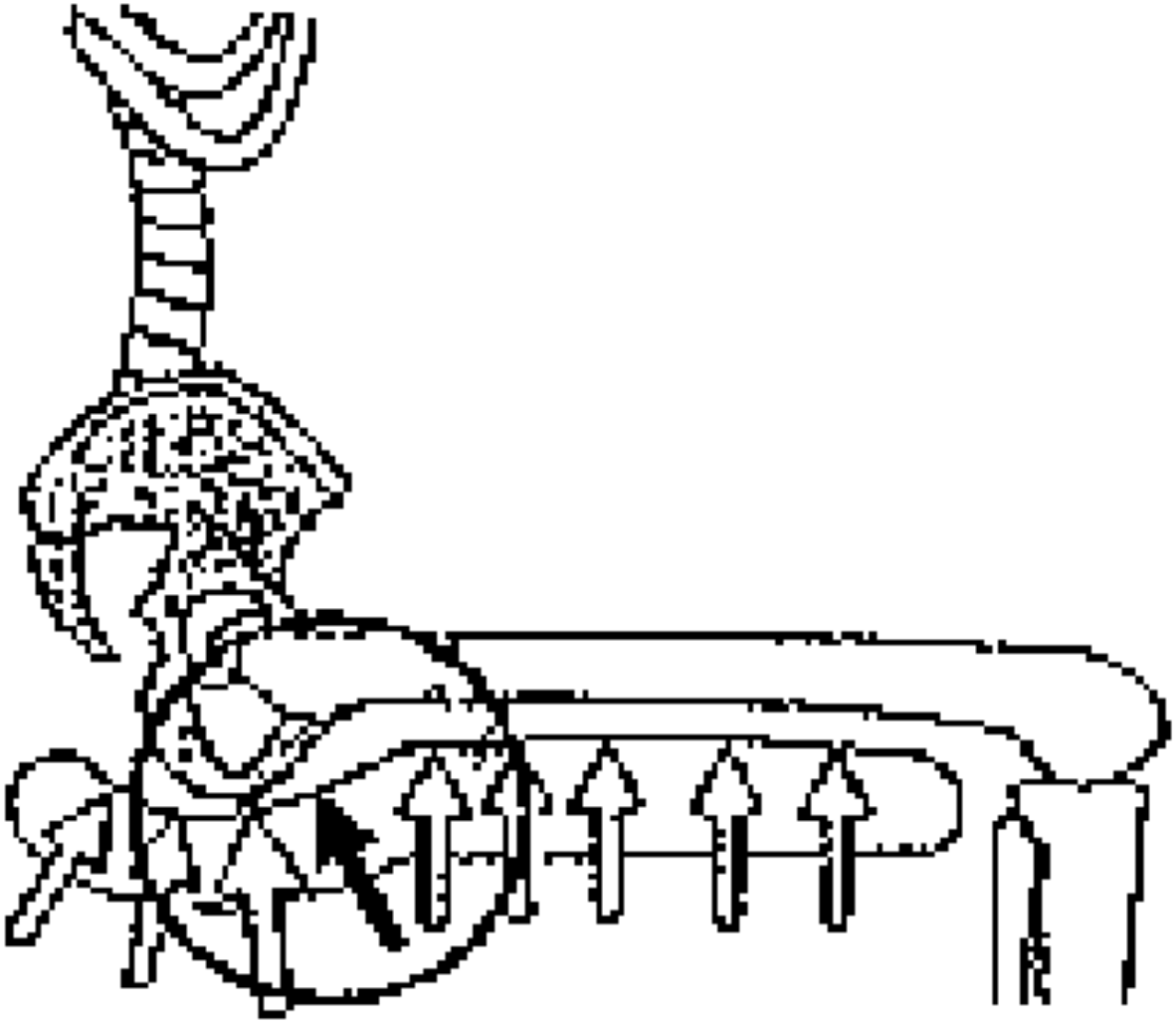
CIRCULATION PROBLEMS

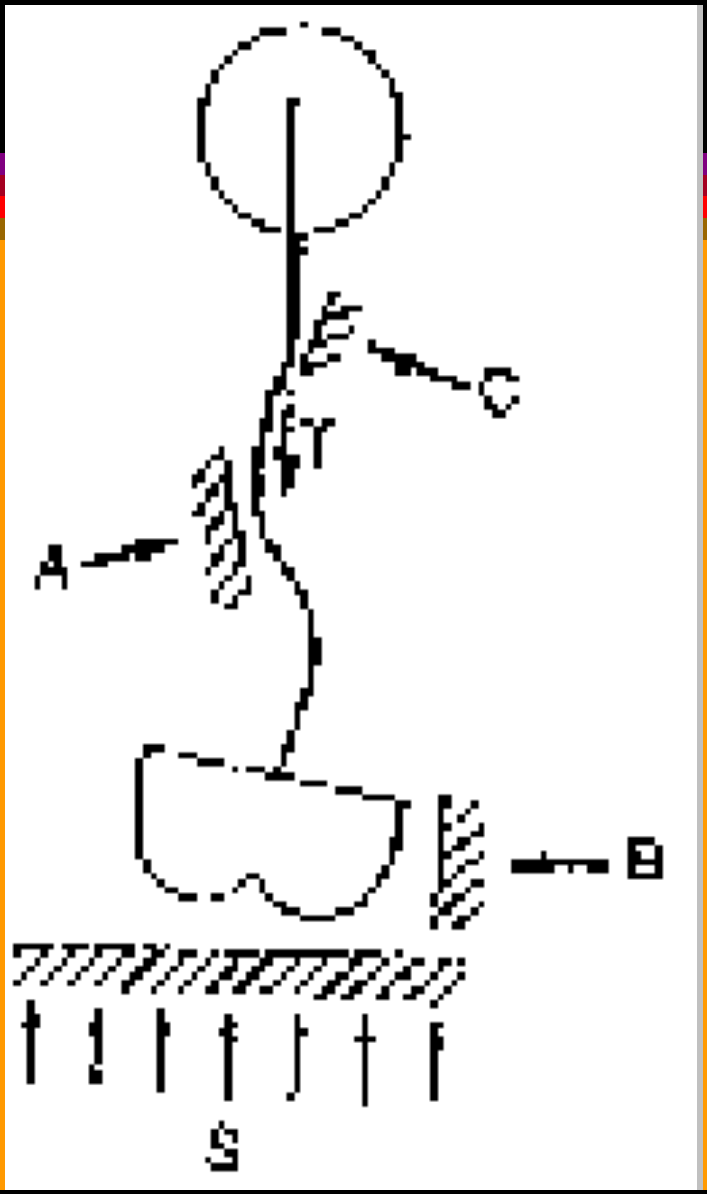
- hypostatic edema
- signs of bad peripheral circulation

PROPER SITTING

- upright
- position of the head (head support)
- position of the spine (hyperlordosis)
- position of the pelvis (anchorage)
- position of the upper limbs (arm supports)
- position of the lower limbs -feet (foot supports)
- special additions



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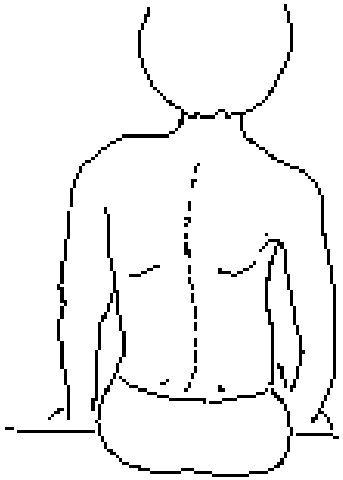
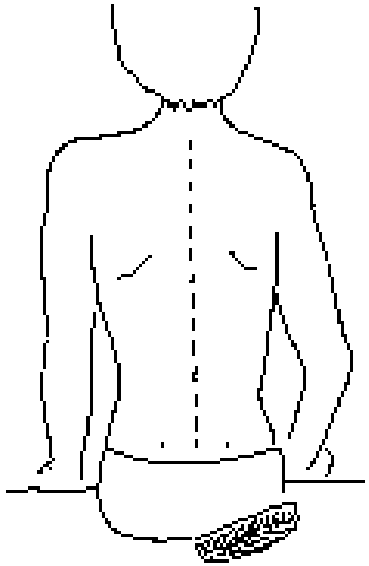




TECHNICAL AIDS

- electric wheelchairs
- equipment for: using computers
  for driving a car

Twister Junior

Twister Junior

DEPENDENCE ON THE HELP OF OTHERS

- book

ANTON ZUPAN

KAKO NUDIMO FIZIČNO POMOČ MIŠIČNO OBOLELIM

DRUŠTVO MIŠIČNO OBOLELIH SLOVENIJE

PRESEDANJE-PRESTAVLJANJE z vozička na stol, posteljo, v avto in obratno

Če si distrofik pri presedanju ne more nič več sam pomagati, ga je treba presesti – prestaviti.

48

En pomočnik (sl. 48)

Pomočnik stopi pred distrofika, obrnjen proti njemu. Voziček mora biti zavrt in odstranjena podnožnika ter naslon za roko na tisti strani, na katero distrofika presedamo. Distrofikovi nogi morata biti trdno na tleh. Nogo na tisti strani, na katero ga presedamo – prestavljamo, mora distrofik (ali pa mu to napravi pomočnik) pomakniti za dolžino stopala pred drugo nogo. Pomočnik s svojima kolenoma objame distrofikovi koleni z zunanje strani, položi njegovi roki preko svojih ramen, kjer ju le-ta sklene in objame pomočnika kar najbolj trdno; za stabilnost lahko distrofik položi brado na pomočnikovo ramo. Pomočnik položi svoji roki okrog distrofikovega hrbta, kar se da nizko, in ju sklene (lahko prime tudi za hlače). Na dogovorjen znak pomočnik stisne koleni, se nagne nazaj, z rokama potegne navzgor in z obratom presede – prestavi distrofika.

Dva pomočnika

49

Način 1 (sl. 49) Pomočnika postavita voziček tako, da so kolesa vzporedna s stranico postelje ali stola. Prvi pomočnik stopi za voziček, za hrbet distrofika, drugi pa pred distrofika obrnjen proti njemu. Prvi pomočnik položi svoji roki od zadaj pod distrofikovima pazduhama in od zgoraj prime njegovi prekrižani roki. Drugi pomočnik pa prime distrofikovi nogi pod kolenoma. Na dogovorjen znak oba pomočnika distrofika dvigneta in ga prestavita.

NUTRITIONAL DISTURBANCES (obesity, malnutrition, constipation etc)

- balanced nutrition with enough proteins and vitamins if necessary laxatives

PAIN

- criomassage
- electrotherapy
- cautious passive exercises
- analgetics

SOCIAL AND PSYCHOLOGICAL PROBLEMS

- a psychologist and a social worker
- numerous social contacts

PERSONS WITH NMD

- **Should live normal life!**
- **Can live normal life!**

Gait patterns in different neuromuscular disorders

Aleš Pražnikar
Rehabilitation institute, Ljubljana

Neuromuscular disorders

- Neuromuscular disorders (NMD) are a heterogeneous group of diseases of motor unit.
- The predominant clinical sign in NMD is muscle weakness.

Gait in patients with NMD

- Difficulties in gait are
  - often presenting sign
  - one of the most frequent complaint and
  - main contributor to disability.

Preserving gait autonomy is a major goal in the management of people with neuromuscular disorders.

Hsu et al. 1993

Epidemiology of NMD

	Prevalence (1/1000000)
Muscular dystrophy	
Duchenne	32
Becker	>7
Facioscapulohumeral MD	20
Limb-girdle MD	<40
Steinert disease	50
Spinal muscular atrophy	12
CMT	100
ALS	57
Miasthenia gravis	80

Emery, 1991

Clinical goals

- to improve and to prolong gait and stance in patients in NMD
- Procedures:
  - surgery
  - bracing
  - physiotherapy
  - pharmacological treatment

Surgery for gait in NMD

The basic principles of management can be described as correction of deformity, stabilization of unstable joints and balance of muscle power.

Early surgery in DMD

- 32 patients with DMD 6.1 years old and follow-up 3.4 years – all ambulate with less contractures

Goertzen et al, 1995

- 87 with DMD 6.56 years old and follow-up 5.4 years and control group of 100 non-operated patients with DMD
  - loss of walking ability occurred in the control group at an average of 9.29 years and in the operated group at an average of 10.55 years

Forst, 1999

Surgery for gait prolongation

Prolongation of ambulation in children with Duchenne muscular dystrophy by subcutaneous lower limb tenotomy.

- 29 patients with hip, knee, and ankle tenotomies continued ambulation in long-leg braces to a mean age of 12 8/12 years
- control group of 25 ceased ambulating at a mean age of 10 years
- Conclusion: s.c. tenotomy is effective in allowing braced ambulation well beyond what the natural history would allow.

Smith et al, 1993

Bracing for gait prolongation

- Lightweight, modular knee-ankle-foot orthosis for DMD: design, development, and evaluation.
  - 9 with knee-ankle-foot orthotics of 26 patients
  - 23% weight saving, resulting in a 10% energy saving during ambulation and an 8% increase in walking speed
  - They were easier to don/doff and were preferred by all involved

Taktak et al., 1995

Bracing for gait ambulantion

- The value of orthoses for patients with DMD
  - 17 consecutive patients with KAFO
  - 7 (41%) became effective ambulators
  - 4 (23%) had borderline results
  - 6 (35%) were considered failures
- We believe orthoses are indicated for many but not all patients with Duchenne muscular dystropy.

Ziter et al., 1979

Bracing for gait ambulantion

The effects of knee-ankle-foot orthoses in the treatment of Duchenne muscular dystrophy: review of the literature.

- 9 studies relevant
- operations on the lower limbs were performed on most patients

The scientific strength of the studies reviewed is poor. It seems that the use of knee-ankle foot orthoses can prolong assisted walking and standing, but it is uncertain whether it can prolong functional walking. The boys that benefit most have a relatively low rate of deterioration, are capable of enduring an operation and are well motivated.

Bakker et al., 2000

Physiotherapy for gait prolongation

Results of the treatment of Duchenne de Boulogne myopathy by early physiotherapy. A comparative study with untreated subjects.

- 163 patients in 2 groups
- gait prolongation 3 to 4 years !!

Demos, 1983

Physiotherapy for gait prolongation

Treatment of Duchenne's myopathy with early physiotherapy. Critical analysis

- 35 patients treated with early intensive physiotherapy were compared with 28 untreated ones.
- prolongation of walking was 8 months.

- the efficiency of a therapeutic modality in Duchenne Muscular Dystrophy (DMD) is difficult to judge

Rideau, 1985

Surgery and physical medicine

Management of muscular dystrophy. Pharmacological and physical aspects.

- Tenotomy for release of contractures in DMD is of practical value only in the context of fitting callipers and has no place in isolation in the management of the disease.
- How much exercise is optimal in NMD?
- Prolongation of up to 3 years?

Dubowitz, 1980

Pharmacotherapy in DMD

Practice parameter: corticosteroid treatment of Duchenne dystrophy: report of the Quality Standards Subcommittee of the American Academy of Neurology and the Practice Committee of the Child Neurology Society.

Gait prolongation

- Deflazacort 13 months
- Prednisone 24 months

Moxley et al, 2005

And the rest of NMD?

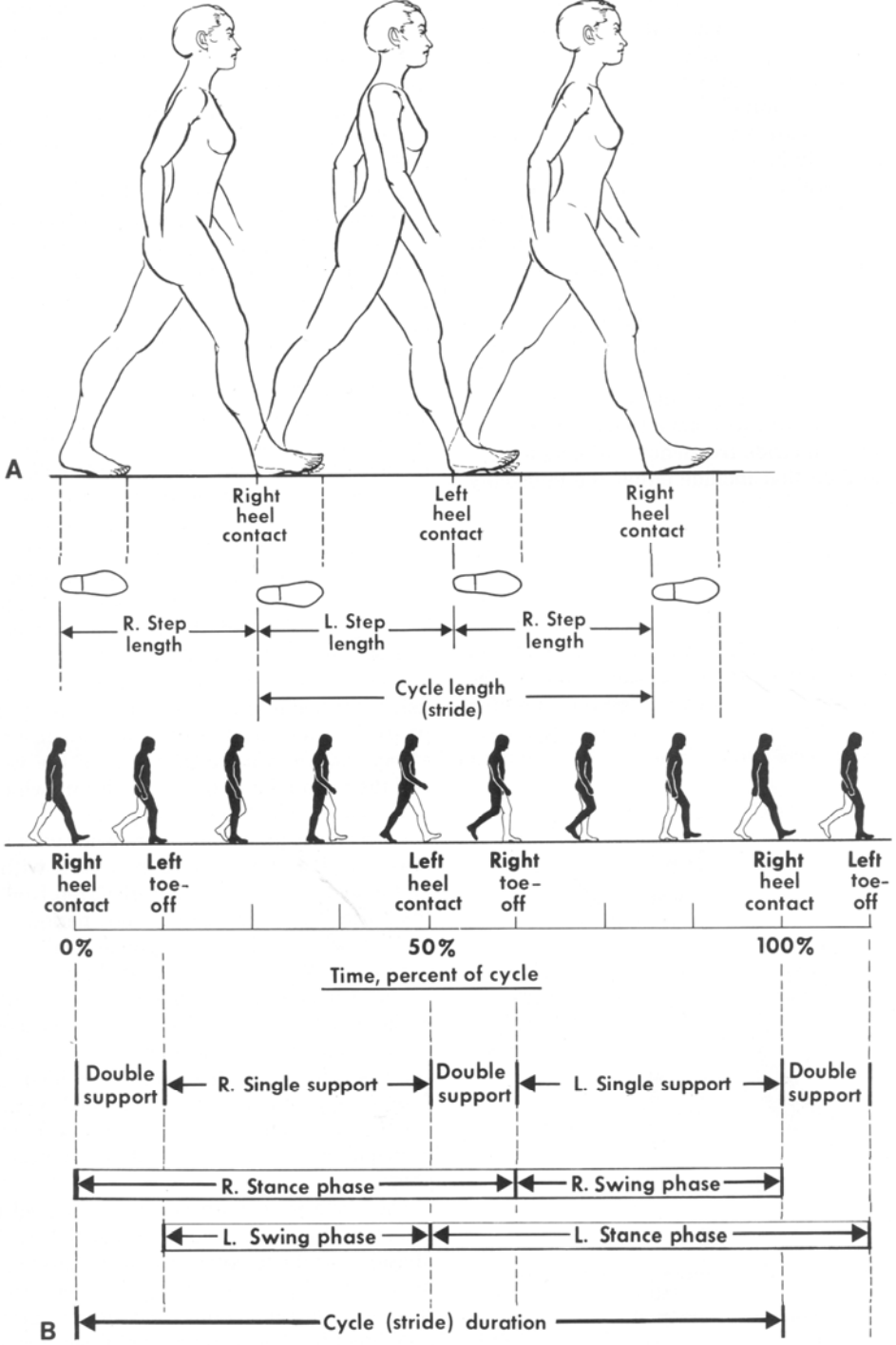
- Much less clinically useful information
- Must apply knowledge of DMD to other forms of NMD
- but growing interest of pathophysiology in recent years.

Gait in patients with NMD

- Descriptions of gait in specific NMD is mostly lacking
- Pathophysiology of emergence of gait problems:
  - muscular weakness,
  - contractures,
  - impaired function of muscle spindles in dystrophic muscle
  - systemic features and
  - others
- Can we distinguish the causes and the compensatory mechanisms?

QUALITATIVE DESCRIPTION OF WALKING

- KINEMATICS (SEGMENTS & JOINTS ANGULAR AND LINEAR DISPLACEMENT)
- KINETICS (FORCES, MOMENTS, POWERS, ENERGY)
- MUSCULAR ACTIVITY (PROPULSION, BALANCE)


A
Right heel contact
Left heel contact
Right heel contact
R. Step length
L. Step length
R. Step length
Cycle length (stride)
Right heel contact
Left toe-off
Left heel contact
Right toe-off
Right heel contact
Left toe-off
0%
50%
100%
Time, percent of cycle
Double support
R. Single support
Double support
L. Single support
Double support
R. Stance phase
R. Swing phase
L. Swing phase
L. Stance phase
B
Cycle (stride) duration

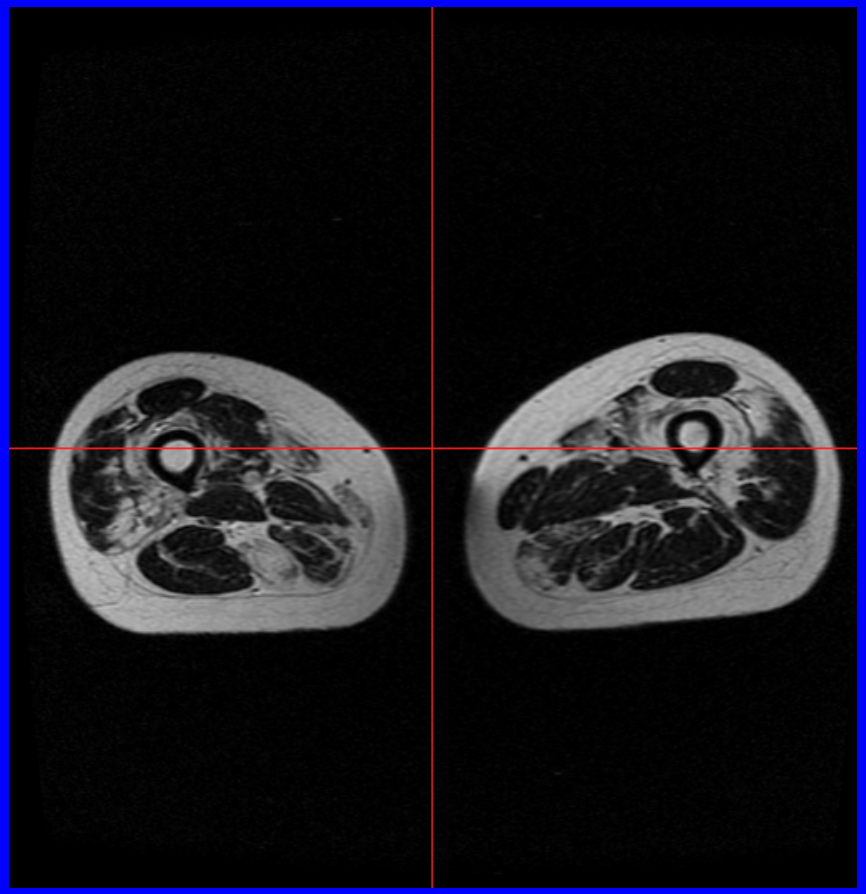
	DMD	DM1	SMA III
Kinematics	Constant plantar flexion, marked flexion in hip and knee, adducted hips, knee valgus	Anterior pelvic tilt, lack of dorsiflexion of foot	Anterior pelvic tilt, decreased flexion of knees, abducted hips
Kinetics	Generation of power in hips in ab/adduction	Flattened proximally, disturbed generation of power in knees	Lack of extension moments
EMG	Flattened EMG envelops with marked activity proximally at take-of	Activity of erectors spinae, bursts at take-off, no activity distally	Lack of activity in body extensors, disturbed activity of flexors and abductors of thigh and knee

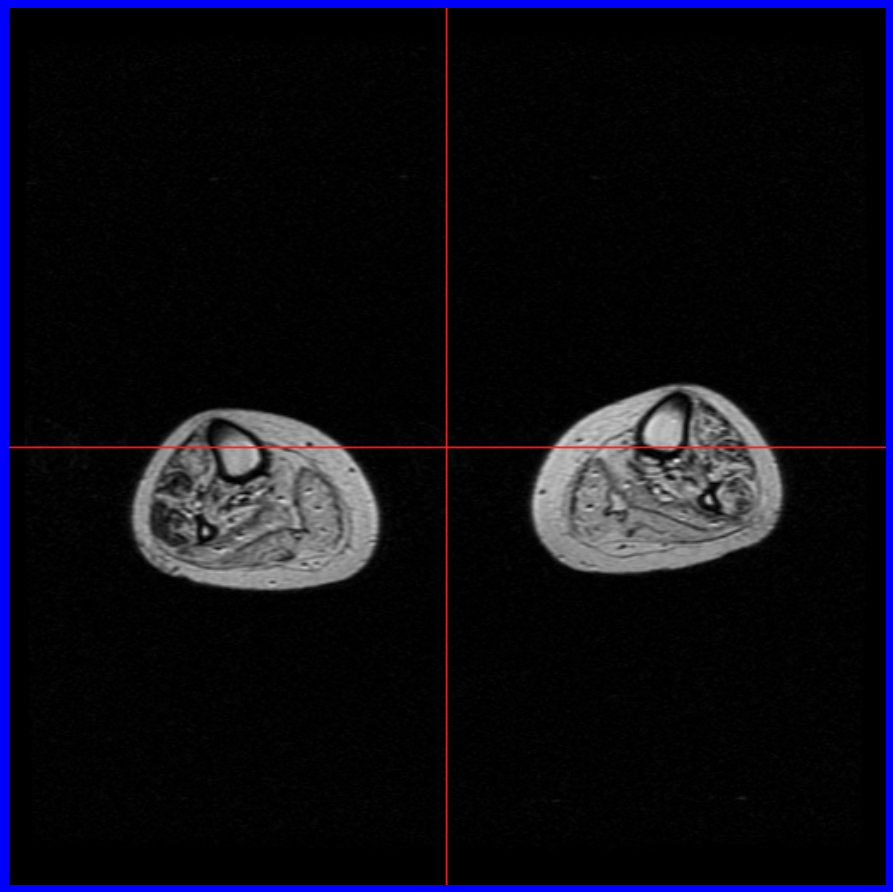
Isokinetics

FLE/EXT	NORMAL	SD	SMA
HIP	268/459	43-help/101	x/50
KNEE	262/122	24/44	11/9
ANKLE	93/30	5/5	14/9

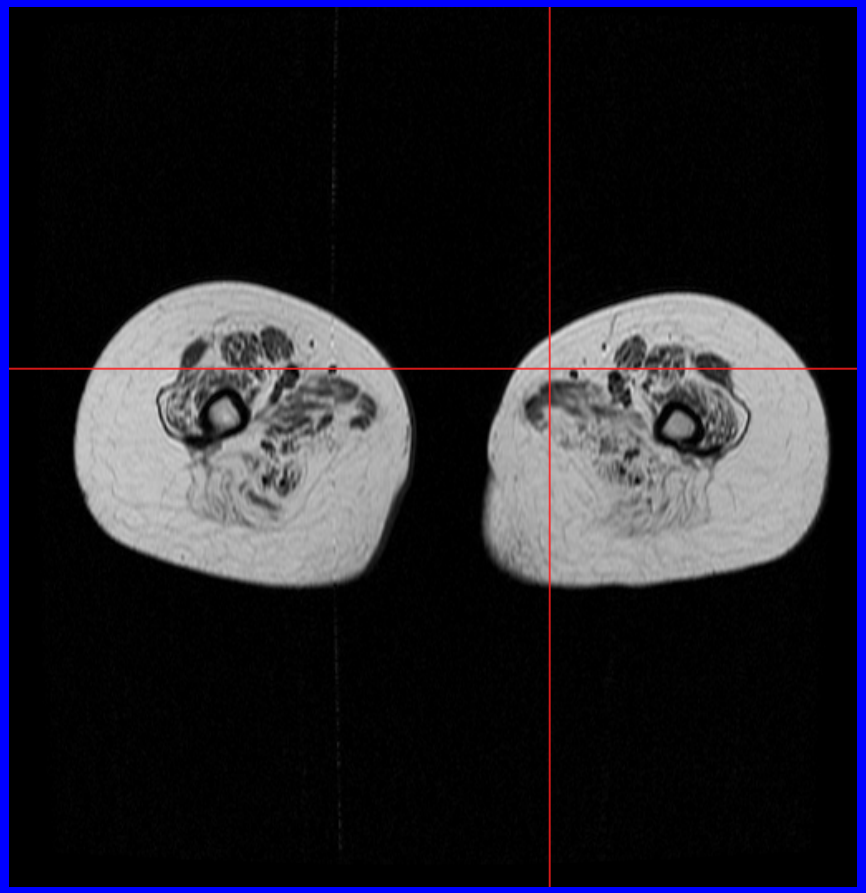
(Nm), concentric muscle activity at 60 º/s, hip at 30 º/s.

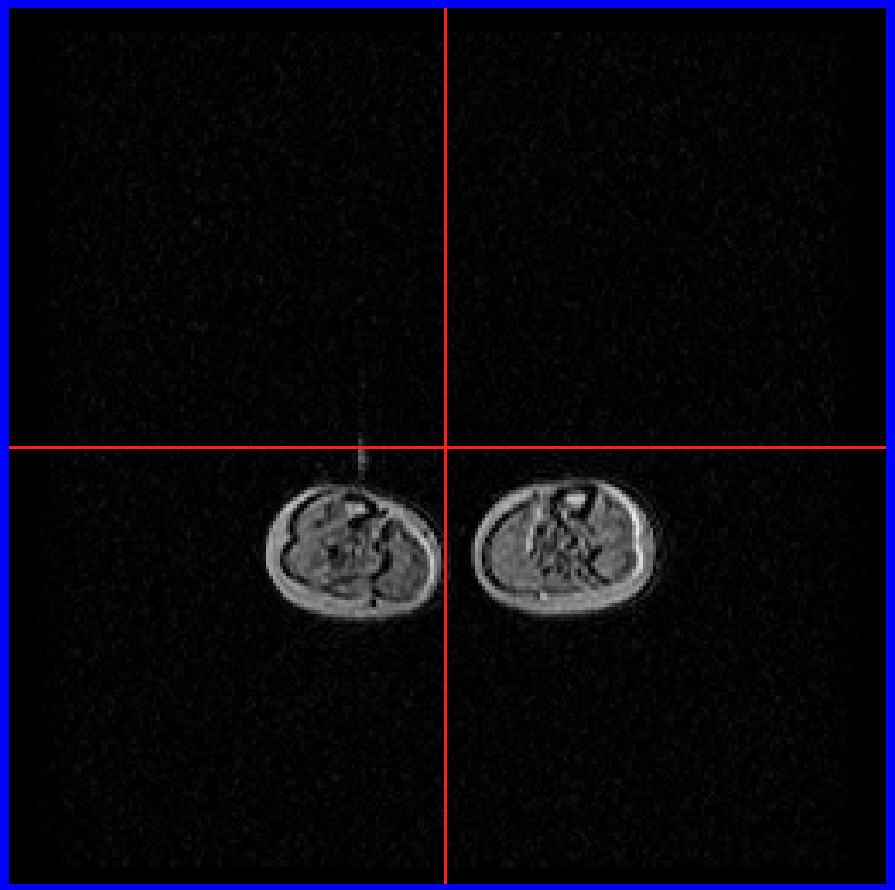
MRI - muscles (DM1)





MRI - muscles (SMA III)





What do we expect?

- To recognize causes of disturbed gait in patients with NMD.
- To recognize compensatory mechanisms developed by patients to negotiate the gait problems
- To develop treatments to support the natural compensation and doing so prolong the ambulation in patients with NMD

EUROPEAN UNION
& Physical and Rehabilitation Medicine

EUROPEAN SECTION MPR

Union Européenne des Médecins Spécialistes UEMS

Hannu Halila

Bernard Maillet

Physical and Rehabilitation Medicine Section

“PRM is the medical specialty that coordinates, performs and evaluates interventions and programs based on the diagnosis of health conditions and the assessment of functioning to enable people with/or at risk of disability to achieve and maintain optimal functioning in the interaction with the environment in all situations from the acute hospital to the community.”

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Pr J.M Viton

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EXAMINATION

Director examination (BC)

Pr Xanthi Michail

EXAMINATION 2004

23 trainees passed

INITIAL TRAINING

Next Examination

Saturday 26 Novembre 2005

INITIAL TRAINING

Site Visit Committee

Pr Alex Chantraine

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criteria trainers

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(re)accreditation

Site Visit Mûnich 04 Pr G.Stucki*

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EUROPEAN TEACHING PROGRAMS

EUROPEAN SCHOOL MARSEILLE

"Motor Disabilities: posture and movement analysis, Rehabilitation, Neurophysiology"

PI Erasmus Socrates Juillet 2005

A.Bardot, A.Delarque, JM.Viton,
L.Bensoussan V.Debovis Milhe

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Department PMR

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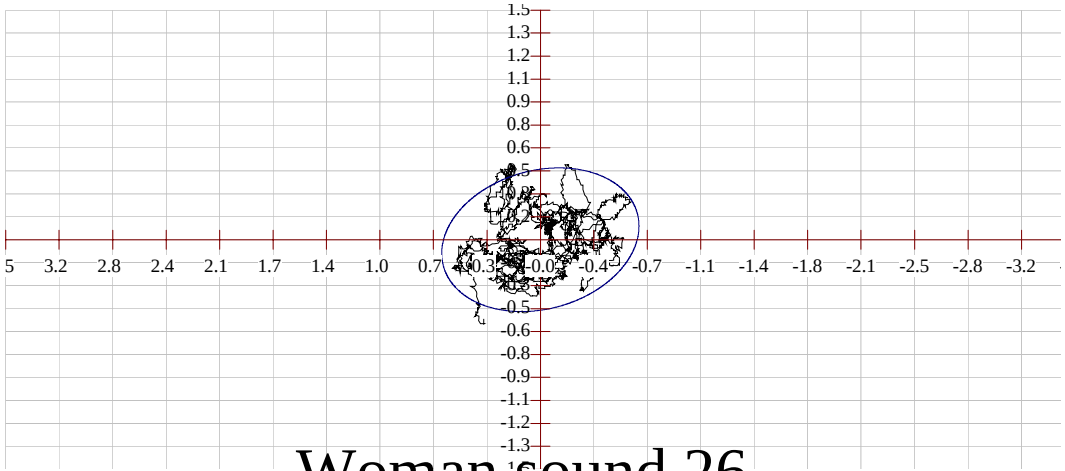
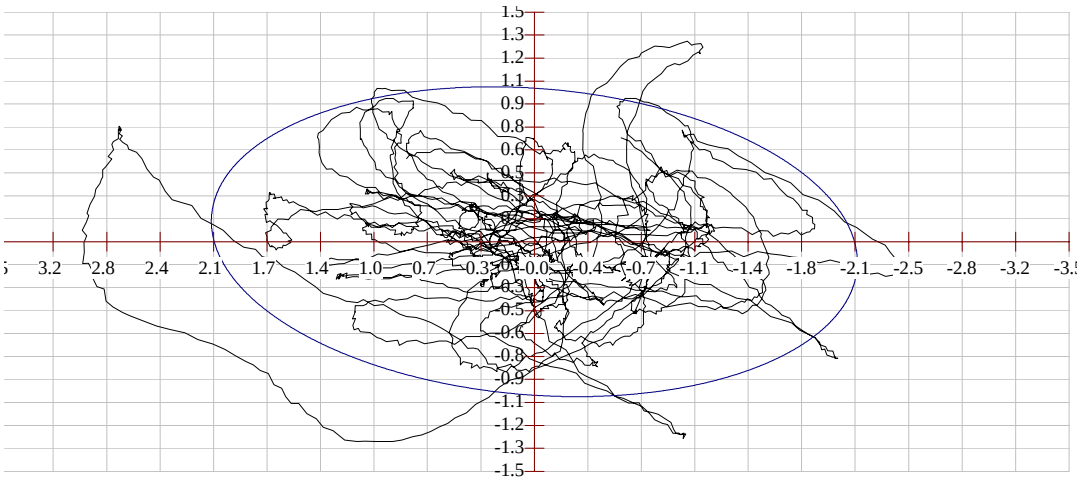


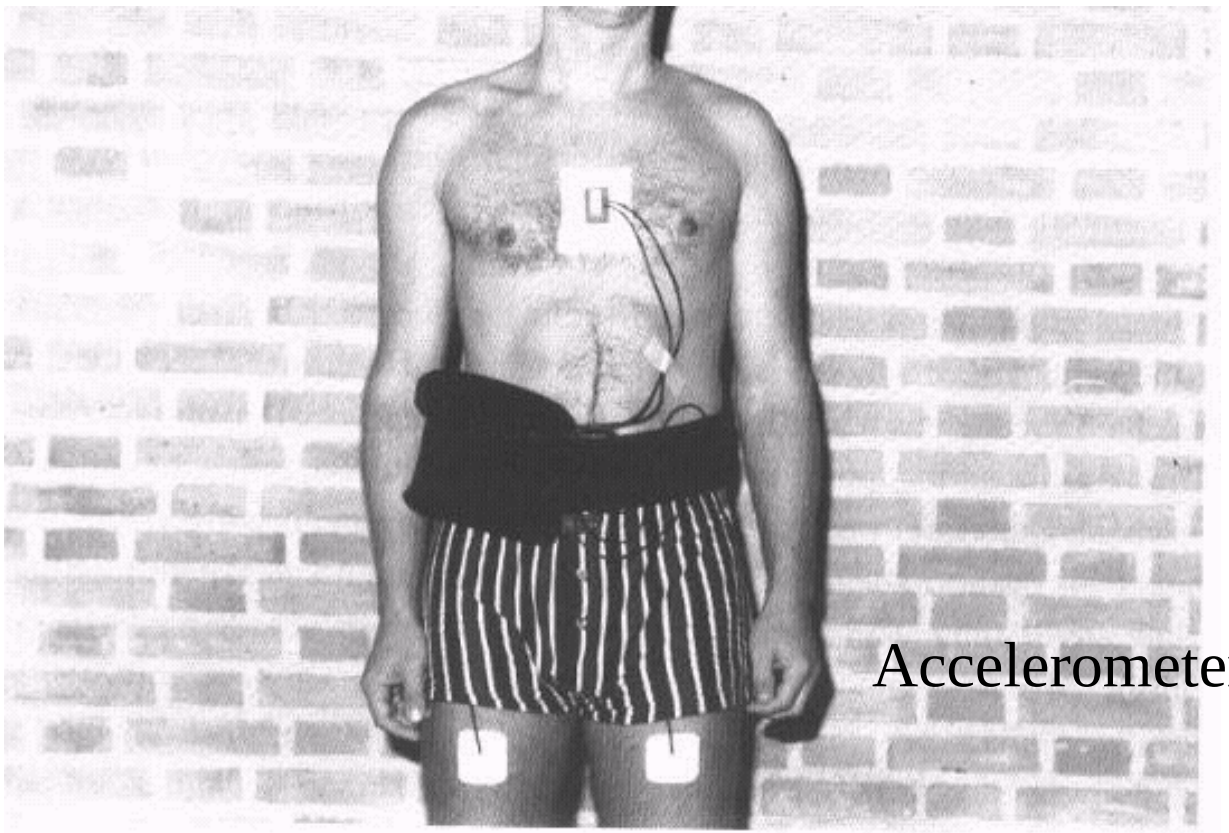
Woman sound 26



Woman Cerebellar syndrom 20

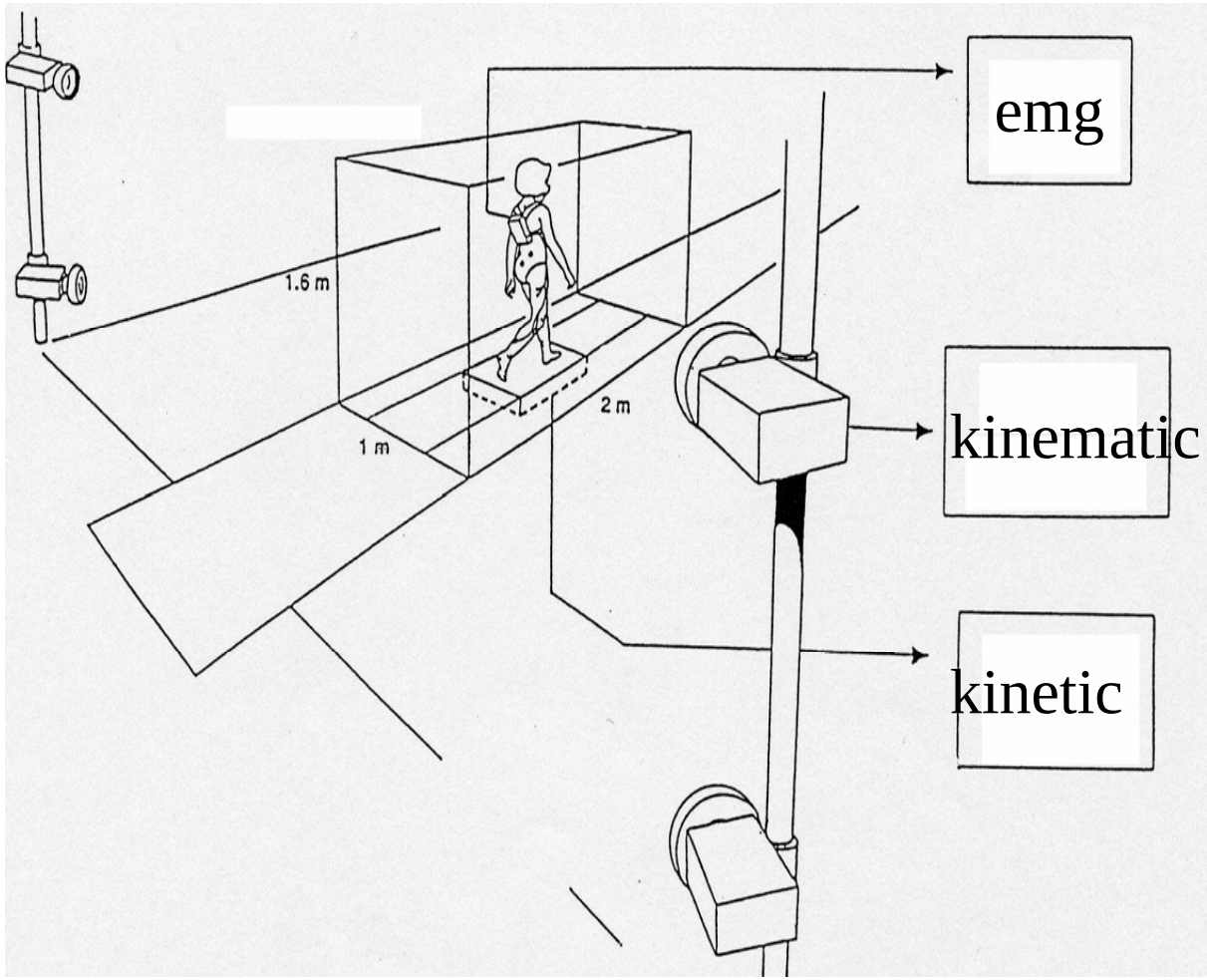
Back home

Ambulatory monitoring

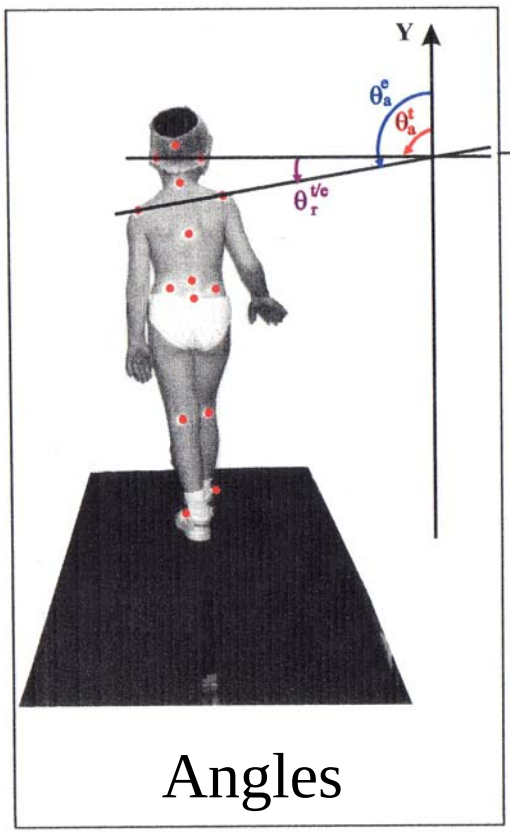


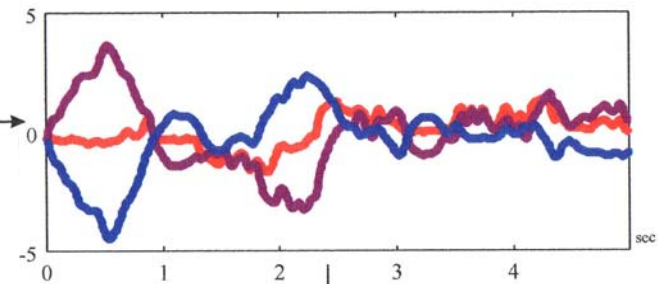
Accelerometers

Laboratory

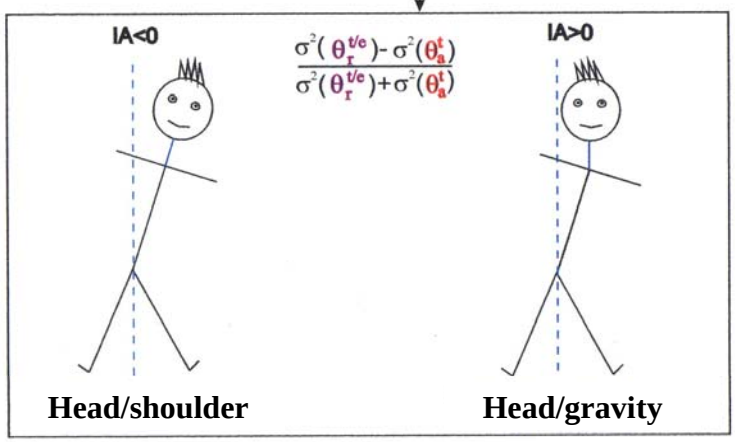


Christine Assaïante





Anchoring index



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60 trainees

http://www.mediterranee.univ-mrs.fr/esm

ESM 05

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www.euro-prm.org

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  grants EB registered trainees

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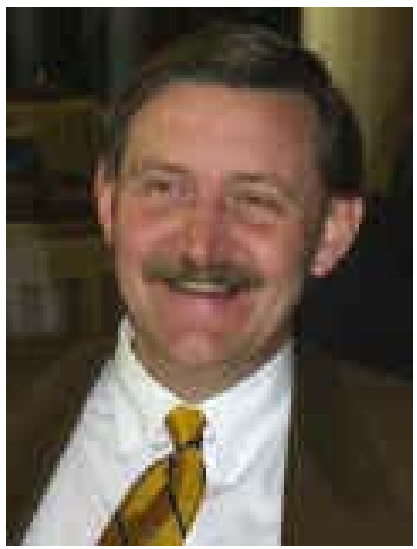

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- **Disabled persons associations**
- **European Bodies**

European Neuro Muscular Centre

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European Council

Strasbourg

1 V. Boltho, Head Health & Social Affairs

2 T. Afflerbach, Coord Rehab & Integration Disabled Persons

European Commission

Brussels

Ms Wallis Goelen

General Direction Employment & Social Affairs

Integration Disabled Persons

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Stadt Linz

Legends to illustrations:

Slide 4. Ysbrand Poortman, initiating founder and long-standing vice-president of EAMDA

Slide 5. Early Yugoslavian delegates: Gordana Rajkov (Serbia, seated with helper standing behind her) and Prof. Osman Sinanovic (Bosnia & Herzegovina, background)

Slide 11. Board of EAMDA 1987-89: seated, from left to right: Prof. Reinhardt Rüdel (president), Gordana Rajkov, Derek *; standing: Ysbrand Poortman, Patricia Melsom, Judy Windle (vice-presidents)**

Slide 15. Chartering of the European NeuroMuscular Centre (ENMC). From left to right, seated: Joop Bars (president VSN); the chartering notary; Prof. Giovanni Nigro (representative U.I.L.D.M.) Prof. Reinhardt Rüdel (president EAMDA); standing: Nelleke van den Bergh (secretary EAMDA); Michael Rutgers (manager ENMC); Ysbrand Poortman (vice-president EAMDA)

The History of EAMDA:

The European Muscular Disease Associations United Long Before the Politicians Generated the European Union

Reinhardt Rüdel, PhD
Deutsche Gesellschaft für Muskelkranke (DGM)

Zagreb, 23 September 2005

The Foundation of EAMDA

- EAMDA was founded in 1971 in London

  following incentives by the Dutch MDA (VSN) one year earlier. The sparking figure was Ysbrand Poortman.

- The founding associations were from (in alphabetical order):

  Belgium, Denmark, Germany (FRG), France, Great Britain, Italy, The Netherlands, Sweden and Yugoslavia.

- A constitution was accepted with the following major aims:

  1. To develop help for all people who suffer from muscular dystrophy or a related disorder.
  2. To improve their physical and mental conditions, both with regard to their social and professional possibilities.
  3. To support research into the causes, treatment and cure of this group of diseases.

Present Memberships of EAMDA

- EAMDA was first officially registered in Brussels in 1972.
  The seat was later transferred to London, UK, and has since 2002 been in Malta.

- Later joining or applying associations:
  Austria, Bulgaria, Belgium, Croatia, Cyprus, Czech Republic, Denmark, Estonia, Finland, German Democratic Republic (united with FRG), Greece, Hungary, Ireland, Kosovo, Lithuania, Malta, Moldavia, Norway, Poland, Portugal, Romania, Russia, Serbia & Montenegro, Slovakia, Slovenia, Spain, Switzerland (German & French), Turkey, Ukraine.

- Associations that have left EAMDA:
  Austria, Bulgaria, Estonia, France?, Greece, Hungary, Moldavia, Poland, Spain, Ukraine

EAMDA has at present 22 members.

Common Language of EAMDA

- In contrast to the babylonic language confusion within the EU, EAMDA adopted English as the only lingua franca right from the beginning.

  The reasons were that English

  – is the language of science that is generally used by all doctors and scientists dealing with muscular disorders.

  – is world-wide the language of computers. All the younger patients with neuromuscular disorders very quickly recognised the value of this tool as a means of communication when personal computers became available…

A Common Currency for EAMDA

- In the early days of EAMDA, the official currency was the British Pound

  because the seat of EAMDA was in London.

- In the late 1980s, an often used currency was the Irish Pound

  because the long-standing treasurer, Judy Windle, was from Eire and thus the accounts were kept in Dublin.

- Also, the American Dollar was often used in early EAMDA balance sheets

  to avoid the confusion with British and Irish Pounds; and because for many members, the Dollar was the best known international currency.

A Common Currency for EAMDA

- At the beginning of the 1990s, the ECU was adopted as the only currency to be used in official EAMDA documents

  although it was only a nominal currency and by far not as familiar as the US Dollar.

- With the introduction of the Euro, this currency was of course immediately agreed on by EAMDA.

  Euro is just another name for the ECU whose value remained unchanged.

Specific Problems for EAMDA

- Being an umbrella organisation, EAMDA has not its own territory where it could easily go fund-raising.

  EAMDA should try to win an international pharmaceutical company as permanent sponsor.

- Incongruent opinions on how EAMDA should split its efforts between Medical and Social Affairs have severely drained on EAMDA's strength and credibility.

  Supporters of Social Affairs tend to neglect the fact that potent sponsors are more easily found for Medical affairs.

  Supporters of Medical Affairs tend to underestimate the wish of adult affected persons to profit from EAMDA actions during their life-time.

- The financial possibilities of the members vary considerably.

Patronages Created by EAMDA

- When the East-European States had regained their sovereignty in the early 1990s, their newly founded MDAs immediately wanted to join EAMDA,

  but they did not have the money to pay the membership fees.

- Thus, EAMDA created patronages of established members to support financially a specific new member:

Denmark	for	Estonia
France	for	Romania
Germany	for	Czechoslovakia
Great Britain	for	Poland
Netherlands	for	Bulgaria
Switzerland (French speaking)	for	Croatia
Switzerland (German speaking)	for	Hungary

This system worked out very well.

Unified European Muscle Resarch

- One of EAMDA's three major aims at its foundation was to support research into the causes, treatment and cure of muscle diseases.

  In 1989, making use of substantial Telethon money donated to EAMDA by the French AFM, EAMDA founded the European Neuromuscular Centre (ENMC).

  The task of ENMC is to organise and finance workshops for European muscle experts in order to ally forces, to facilitate the exchange of knowledge and to stimulate new ideas.

- From its beginning, ENMC was so successful that after three years of operation it could be transformed into an independent institution with seat in London.

EAMDA Youth Organisation

- Another one of EAMDA's three major aims is to improve the muscle patients' physical and mental conditions, both with regard to their social and professional possibilities.

  In the late 1980s, the EAMDA Board decided that this was particularly important for young muscle patients.

- They therefore started to organise regularly EAMDA Youth Camps in varying member countries.
- They founded the EAMDA Youth Organisation (EYO) as a subdivision with its own budget and a representation on the Board.
- During the Yugoslav war many young muscle patients from there were received by other member countries.

Conclusion

- Although EAMDA started a decade later (1971) than the European Common Market (EC,1958), major steps towards a harmonisation of the European MDAs were performed faster than comparable steps occurred in the grand politics.

- Constitution
- Common language
- Common currency
- Supported memberships of East Europeans
- Foundation of a European Muscle Research Institution
- Special care for its youth

- The European muscle patients can be proud of the achievements of their representatives. The visions and the actions of Ysbrand Poortman must be acknowledged.

however…

- With the EU now being stronger established, EAMDA should try and find a long-standing European sponsor.

  For this, a reasonable relation with doctors and medical research must be a matter of course.

- EAMDA should strife at being recognised at Brussels as being the European representative of all muscle patients.

  EAMDA should make use of the experience of successful organisations such as VSN or ENMC.

- The rich members must again be convinced of their responsibility for the less affluent fellow members.

  This task has been better solved in the grand politics.

- The youth must be fostered again…

EAMDA ACTIVITIES 2005-2006

Cure, Research and Development of Medicines

~~Pos~~sibilities for drug development

- Advanced technology
- Advanced knowledge
- EU-programmes (FP6, FP7)
- Trial Unit ENMC
- Orphan Drug regulation (Industrial interest)
- Increase of Neuromuscular expertcentres
- Development of Patient Organisations

Trials & Developments

- SMA
- ALS
- Duchenne
- FSHD
- Pompe’s disease
- Multifocal Motor Neuropathy
- Friedreichs’ Ataxia
- Oculopharyngeal Muscular Dystrophy
- Myotonic Dystrophy
- Guillain Barré
- Inclusion Body Myositis
- Myasthenia Gravis

What EAMDA needs

- Representation by EAMDA on the European level to put Neuromuscular Disorders on the (EU) map, and to voice our needs and opinions
- Exchange of information on both medical scientific developments and EU-policy matters
- Development of national activities favourable for therapy development
- Joint effort of all EAMDA members
- Expanding our network (throughout Europe)

Activities by Peter Streng and the EAMDA secretariat

- Representation in Eurordis, ENMC, EPF, EPPOSI, WANDA
- Contacts with Industry (Genzyme, Glaxo, Santhera, Trophos)
- Inventarisation of European Neuromuscular patient organisations (number of patients, national expertcenters)
- Exchange of information by www.EAMDA.net (EU-policy, scientific developments)
- Obtaining funding for these activities (EU, Industry, ...)
- Every 2 years, an EAMDA conference on therapy development (in conjunction with WMS or IC-NMD congres)?

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