



OVERVIEW OF MOTOR REHABILITATION IN NEUROMUSCULAR DISEASES (神经肌肉病的运动康复概述)

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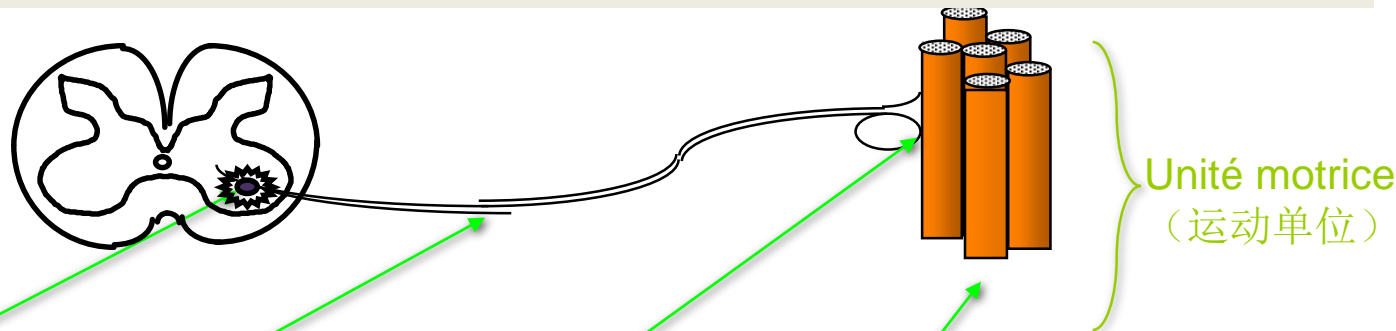
Université Claude Bernard



Lyon 1

Definition : Neuromuscular diseases in children

定义：儿童神经肌肉疾病



Corne Antérieure
前角

Spinal muscular atrophy 脊肌萎缩症

Sclérose latérale amyotrophique
侧索硬化症

Nerf périphérique
(周围神经传导)

Neuropathies
(神经病变)

(CMT disease)
--腓骨肌萎缩症

Jonction NM
(神经受体)

Myasthenia
(重症肌无力)
Myasthenia gravis (假性麻痹重症肌无力)
Congenital myasthenic syndrom (先天性肌无力综合征)

Muscle
(肌肉)

Dystrophies-肌营养不良
Congenital-先天性
Duchenne / Becker- (DMD/BMD)
LGMD-肢带型肌营养不良
Sarcoglycanopathies
Emery-Dreifuss肌营养不良 / FSH-面肩肱型肌营养不良
Myotonic-肌强直
Steinert-远端型肌营养不良
Congenital Myopathy-CMD
先天性肌营养不良²

NMD with an early beginning-神经肌肉疾病通常早期就产生





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■ For DMD (杜氏肌营养不良)

- Steroids-类固醇
- Périndopril 2 mg/day preventive-控制每天2mg的培哚普利哌林多普利
- effect of cardiomyopathy-心肌病的影响
- Exons skipping-外显子跳跃

(Duboc D Am Heart J. 2007 Sep;154(3):596-602)

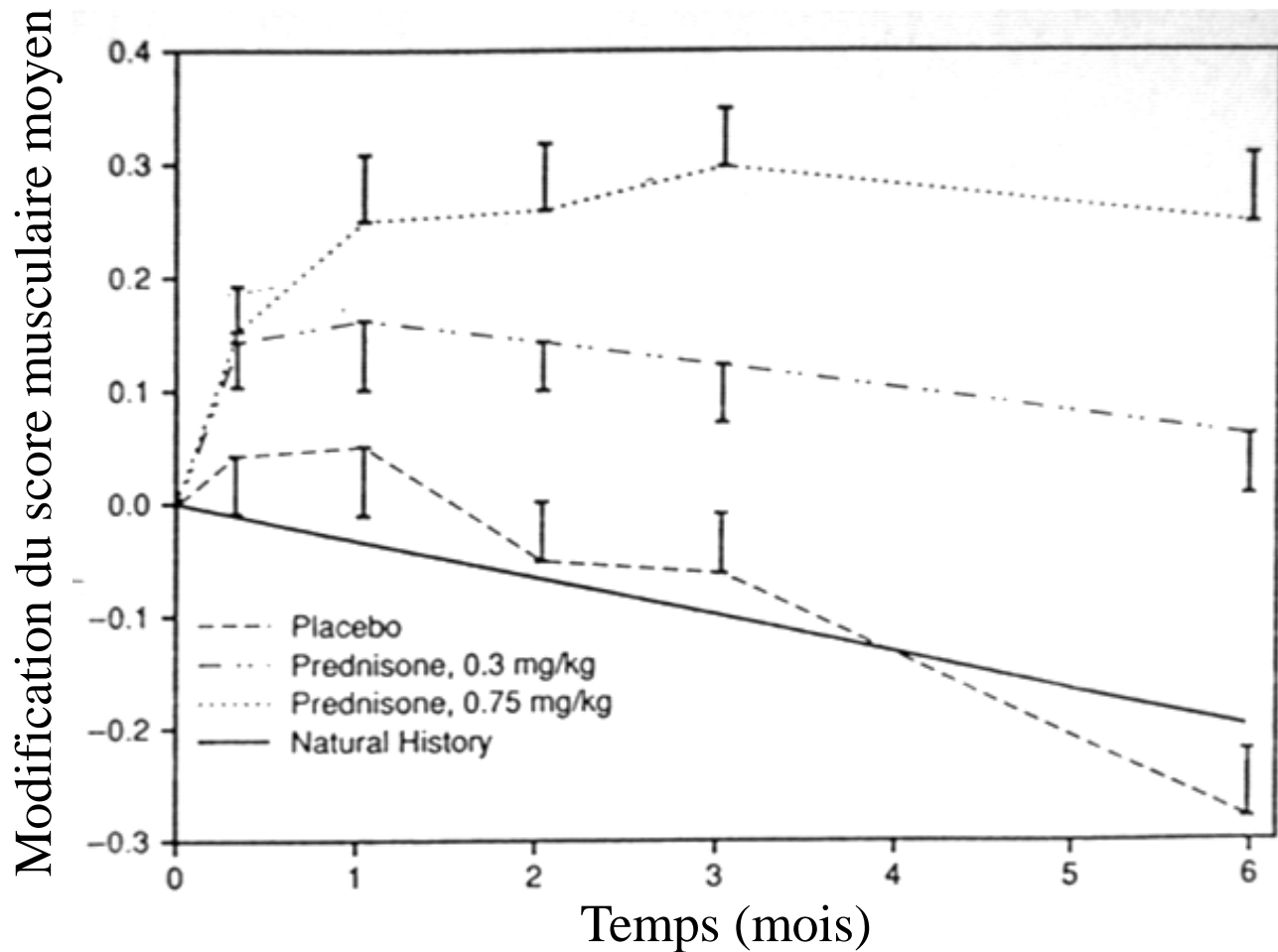
■ For SMA (脊肌萎缩症)

- Salbutamol-舒喘灵
- Olesoxime-奥利索西
- ASO-ASO试验

Duchenne Muscular Dystrophy					
	Programme	Phase 1	Phase 2	Phase 3	
PROSENSA 2PMO	Ex 44	[Yellow bar]			
	Ex 45	[Yellow bar]			
	Ex 51	[Yellow bar]			drisapersen
	Ex 53	[Yellow bar]			
SEREPTA Morpholino	Ex 51	[Yellow bar]		eteplirsen	
	Ex 53	[Red star]			
PTC	Codon stop	[Yellow bar]			Disponible en ATU de cohorte
SANTHERA	ibidenone	[Yellow bar]			ATU de cohorte en cours
Spinal Muscular Atrophy					
	Programme	Phase 1	Phase 2	Phase 3	
ISIS	CS3b	[Blue bar]		[Red star]	
	CS4	[Blue bar]		[Red star]	
Roche	MOONFISH	[Red star]			



DMD et corticostéroïdes (CS): utilisation répandue efficacité démontrée sur la force musculaire



New treatments-新治疗

Does the efficacy clinically meaningful ?

(新治疗的临床疗效是否会有意义?)

Lancet Neurol. 2014 Oct;13(10):987-96. doi: 10.1016/S1474-4422(14)70195-4. Epub 2014 Sep 7.

Safety and efficacy of drisapersen for the treatment of Duchenne muscular dystrophy (DEMAND II): an exploratory, randomised, placebo-controlled phase 2 study.

Voit T¹, Topaloglu H², Straub V³, Muntoni F⁴, Deconinck N⁵, Campion G⁶, De Kimpe SJ⁶, Eagle M³, Guglieri M³, Hood S⁷, Liefwaard L⁸, Loubakos A⁶, Morgan A⁹, Nakielny J⁷, Quarcoo N⁷, Ricotti V⁴, Rolfe K⁷, Servais L¹⁰, Wardell C¹¹, Wilson R¹², Wright P¹³, Kraus JE¹⁴.

Author information

Abstract

BACKGROUND: Duchenne muscular dystrophy is caused by dystrophin deficiency and muscle deterioration and preferentially affects boys. Antisense-oligonucleotide-induced exon skipping allows synthesis of partially functional dystrophin. We investigated the efficacy and safety of drisapersen, a 2'-O-methyl-phosphorothioate antisense oligonucleotide, given for 48 weeks.

METHODS: In this exploratory, double-blind, placebo-controlled study we recruited male patients (≥5 years of age; time to rise from floor ≤7 s) with Duchenne muscular dystrophy from 13 specialist centres in nine countries between Sept 1, 2010, and Sept 12, 2012. By use of a computer-generated randomisation sequence, we randomly allocated patients (2:2:1:1; block size of six; no stratification) to drisapersen 6 mg/kg or placebo, each given subcutaneously and either continuously (once weekly) or intermittently (nine doses over 10 weeks). The primary endpoint was change in 6-min walk distance (6MWD) at week 25 in patients in the intention-to-treat population for whom data were available. Safety assessments included renal, hepatic, and haematological monitoring and recording of adverse events. This trial is registered with ClinicalTrials.gov, number [NCT01153932](https://clinicaltrials.gov/ct2/show/study/NCT01153932).

FINDINGS: We recruited 53 patients: 18 were given continuous drisapersen, 17 were given intermittent drisapersen, and 18 were given placebo (continuous and intermittent groups combined). At week 25, mean 6MWD had increased by 31.5 m (SE 9.8) from baseline for continuous drisapersen, with a mean difference in change from baseline of 35.09 m (95% CI 7.59 to 62.60; p=0.014) versus placebo. We recorded no difference in 6MWD changes from baseline between intermittent drisapersen (mean change -0.1 [SE 10.3]) and placebo (mean difference 3.51 m [-24.34 to 31.35]) at week 25. The most common adverse events in drisapersen-treated patients were injection-site reactions (14 patients given continuous drisapersen, 15 patients given intermittent drisapersen, and six given placebo) and renal events (13 for continuous drisapersen, 12 for intermittent drisapersen, and seven for placebo), most of which were subclinical proteinuria. None of the serious adverse events reported (one for continuous, two for intermittent, and two for placebo) resulted in withdrawal from the study.

INTERPRETATION: Continuous drisapersen resulted in some benefit in 6MWD versus placebo at week 25. The safety findings are similar to those from previous studies. Ambulation improvements in this young population with early-stage Duchenne muscular dystrophy are encouraging but need to be confirmed in larger studies.

Ataluren (药品) DMD with a stop codon-终止密码

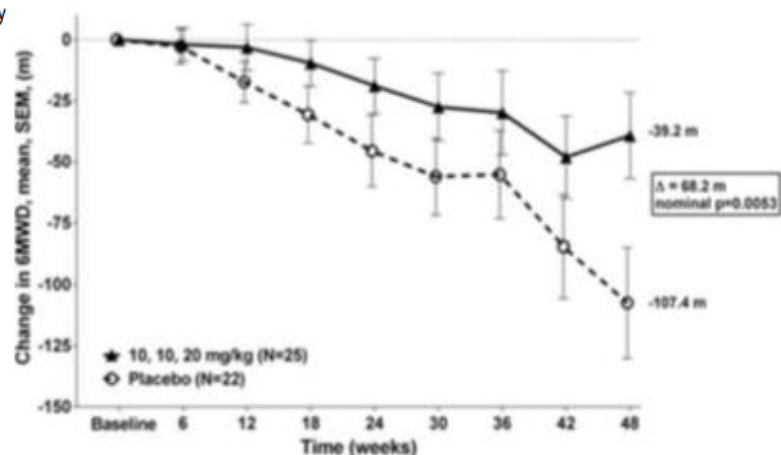


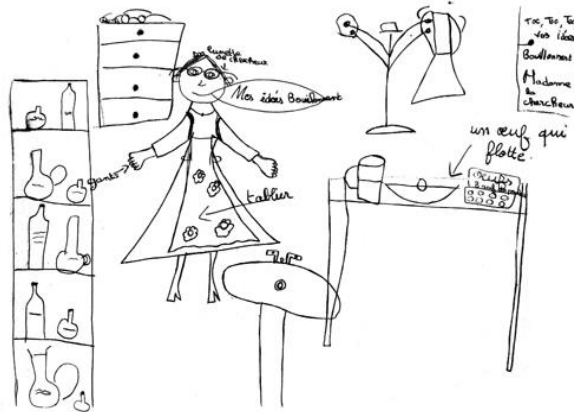
FIGURE 3. Mean change in 6MWD from baseline to week 48 in the < 350 meters 6MWD subgroup.

Drisapersen Exon skipping 51



Need for a longitudinal and multidisciplinary follow up coordinated by a PMR Physician-PMR医生要懂得多维度思考及协调多学科参与

Primary damage
Neurological
impairment – 一级
损伤：神经功能缺损



PREVENTION

Paediatric PMR

Re « education »
(早期教育)

Rehabilitation
(康复)

二级损伤：骨骼、
皮肤、社交并发症

Secondary damage
Orthopedic, cutaneous,
social complications

CURE-治疗

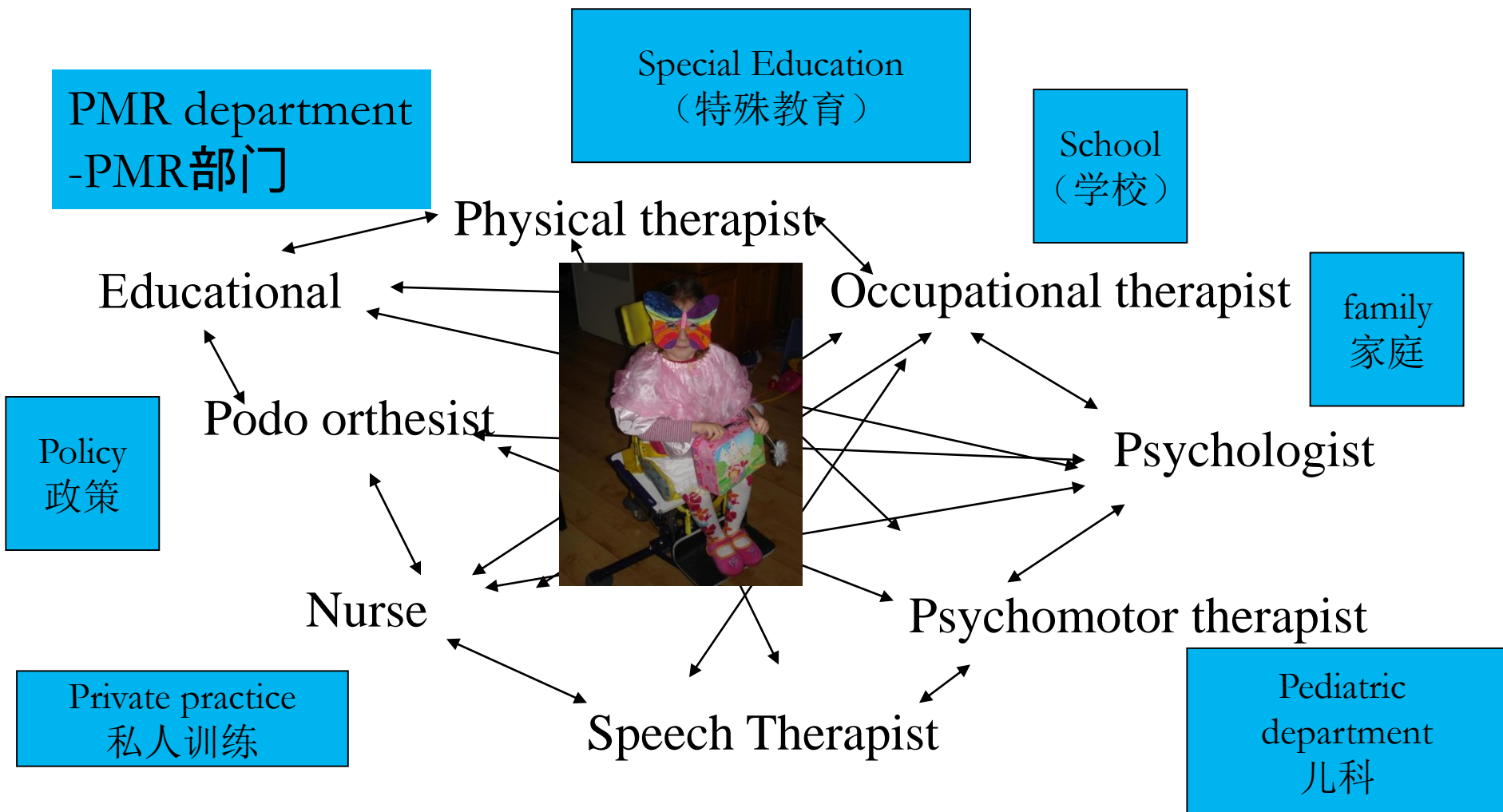
- Orthopedic treatment (cast)-骨科治疗（石膏）
- Surgery (spinal fusion, ankle arthrodesis)-外科手术（脊柱融合术，踝关节融合术）

Tertiary damage-三级损伤
Compensatory movements
or postures used by the
patient because of the
primary damage-因为一级
损伤会让患者出现代偿性动作或者姿势。



PMR Physician

Diagnosis, prevention and treatment of secondary damages caused by the disease (PMR医生: 导致二级损伤的诊断、预防措施及治疗)



Specificities of PMR Consultation

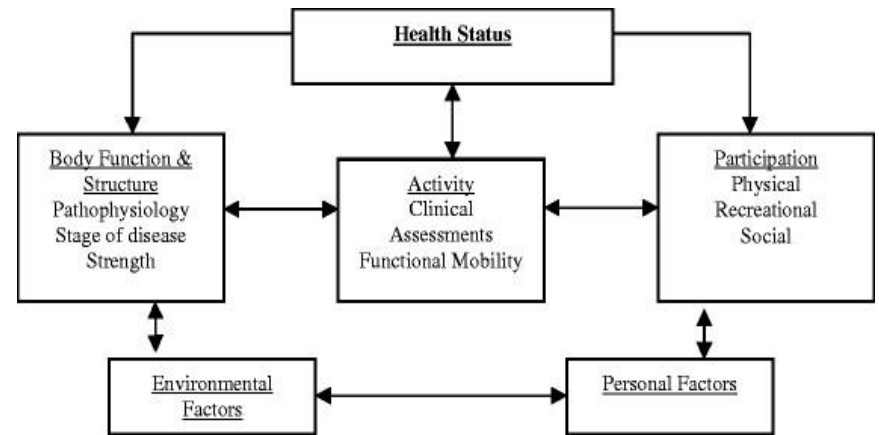
PMR的会诊要点

- First sentence of the consultation-会诊第一要素

“What are you waiting for from us ? What do you need”
(你最想从我们这得到什么? 你需要什么?)

- Taking into account
(考虑因素)

- Health Status-健康状况
- Activity limitation-活动受限
- Environmental and personal factor-环境及个人因素



- A child with NMD is above all A CHILD and THE CHILD of his/her parents-神经肌肉疾病的孩子尤其需要关注的最重要的是孩子本身以及孩子的父母。体现以顾客为中心。



Evaluation scales are for the PMR physician the stethoscope for the cardiologist-评估量表对于PMR医生来说就像听诊器于心脏病科医生一样重要

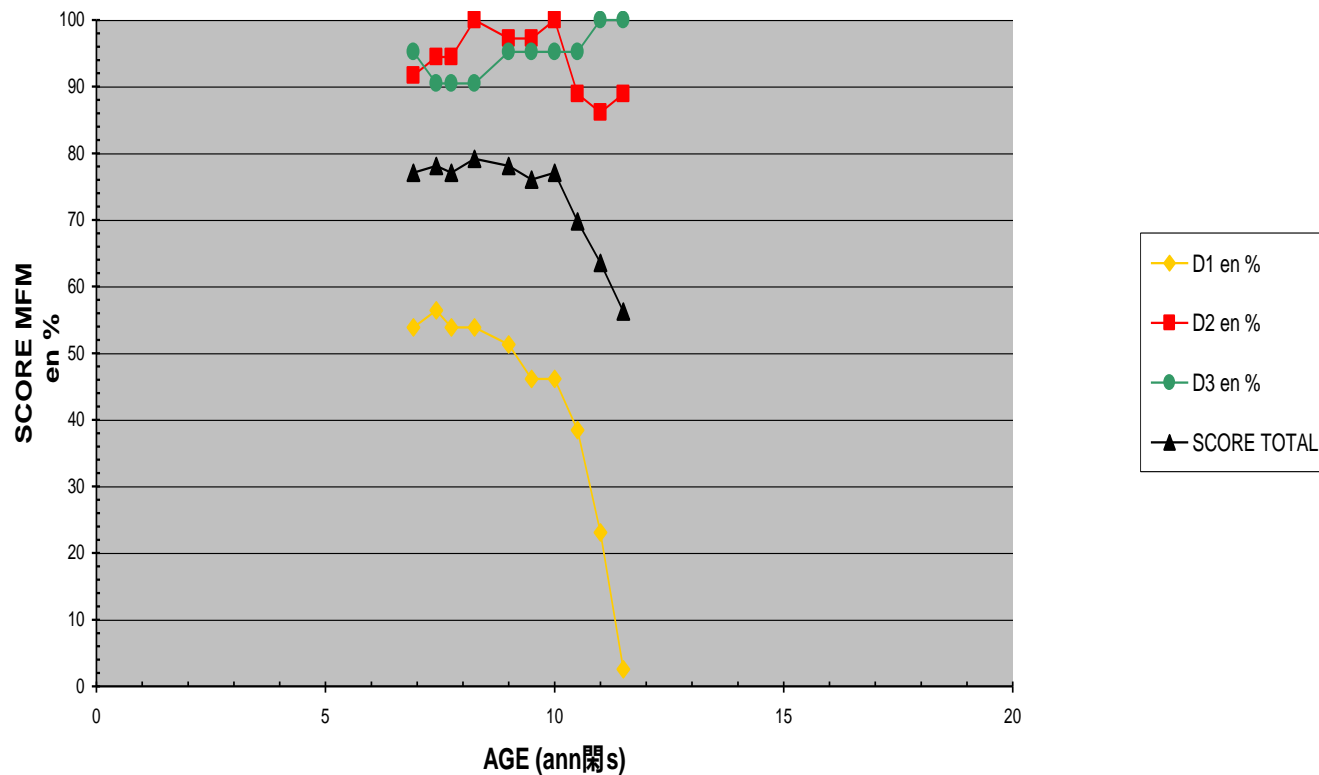
Interests of the evaluation and outcome measures in NMD -NMD的评估要点及结果

- For an optimal Clinical follow up -临床随访要跟上
- To Define realistic goal of management with patient, family and professional-明确病人、家庭及专业人员对于现实目标的区分
- To prepare important step of the disease (the lost of ability to walk for example *vuillerot et al 2009*)-为该疾病重要的进展做准备：比如不能走路
- To measure efficacy of treatment during clinical trials评估在临床试验中的疗效

Evaluation – 评估

A tool to discuss with patient and family about the evolution of the disease in order to prepare important step 一个可与病人和家庭讨论疾病的进展的评估工具，为重要的进展做准备

MFM de Etienne C.



Need for valid, reliable and responsive tools

需要信效度好的且灵活方便使用的评估工具

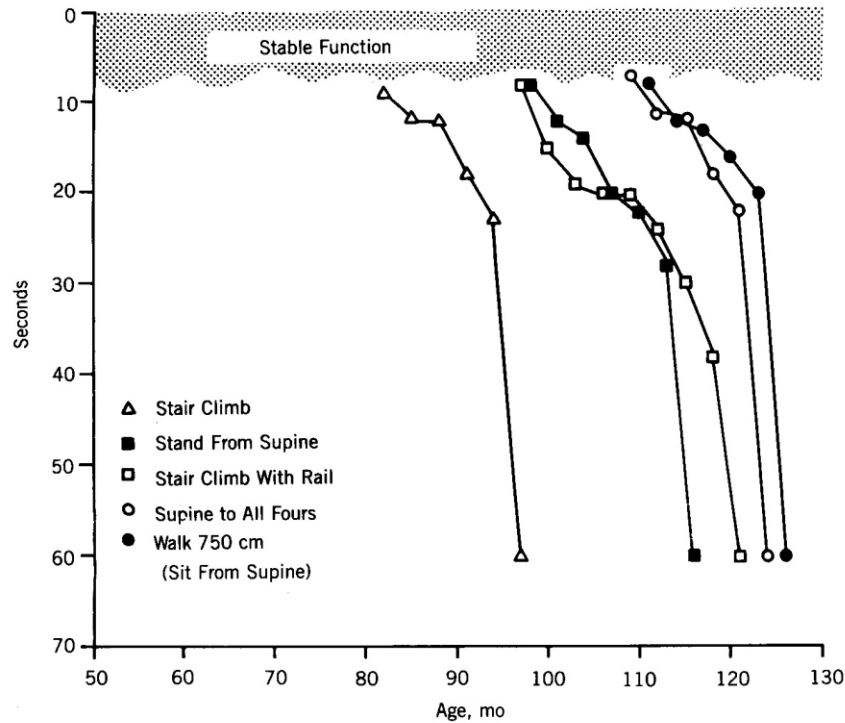


Fig 6.—Composite functional decline in group.

Table 2 Slopes of change in patients with >6 months' follow-up

Variable	SMA Type 2	SMA Type 3 All Ages	SMA Type 3 10–15y
Participants	12	19	10
Age (y)	9.67±3.24 (5.75–16)	21.15±14.14 (8–29)	12.6±1.9 (10–14.58)
Follow-up (mo)	37±20	18.5±14	22±19
D1 score			
Slope (points/y)	-0.18±0.90	-1.95±6.98	-6.22±6.59
SRM	0.2	0.28	0.94
D2 score			
Slope (points/y)	-3.25±2.52	0.71±3.73	-0.72±2.69
SRM	1.29	0.19	0.27
D3 score			
Slope (points/y)	1.93±3.81	-0.13±4.53	-0.52±4.41
SRM	0.51	0.03	0.12
Total score			
Slope (points/y)	-0.86±1.45*	-0.55±3.99*	-2.91±3.33*
SRM	0.6	0.14	0.87

NOTE. Values are n, mean ± SD (range), mean ± SD, or as otherwise indicated.

* Significant changes, SRM>0.6.

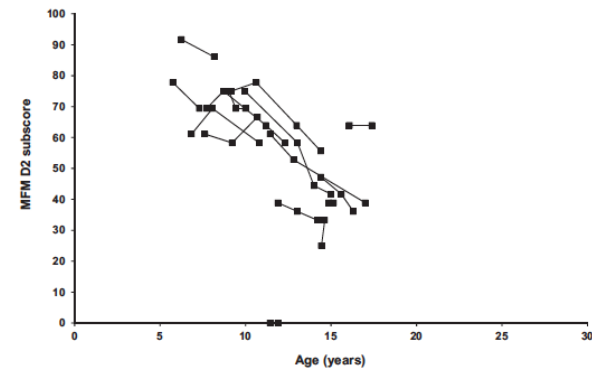


Fig 2 Change in MFM D2 subscore according to age in 14 patients with SMA type 2.

[Vuillerot C et al. Responsiveness of the Motor Function Measure in Patients with Spinal Muscular Atrophy](#)

Arch Phys Med Rehabil 2013 Aug;94(8):1555-61



Classical Orthopedic complications in patients with NMD

神经肌肉疾病在传统矫形治疗下的并发症

- Scoliosis-脊柱侧凸
- Hip luxations-髋关节脱臼
- Contractures +++++-挛缩及挛缩程度
 - All joints are potentially involved : ankles, hip, knees, elbows, shoulders, wrist etc (所有关节都有可能: 肘、髋、膝、踝、肩、腕关节等等)

More than degrees, think about consequences in term of

(不只是损伤程度, 根据以下情况考虑不同结果)

- **Function: walk, hand manipulation, sitting position, ability to wear shoes etc ...**

功能: 行走, 手部控制, 坐姿, 穿鞋能力等等

- **Symptoms : pain, cutaneous**

症状: 疼痛, 皮肤

- **Quality of life**

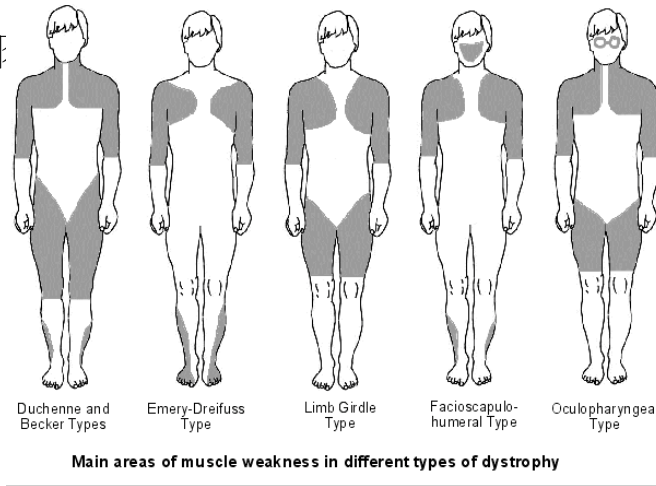
生活质量



A better knowledge of natural history of the diseases help preventing evolution of orthopedic complications

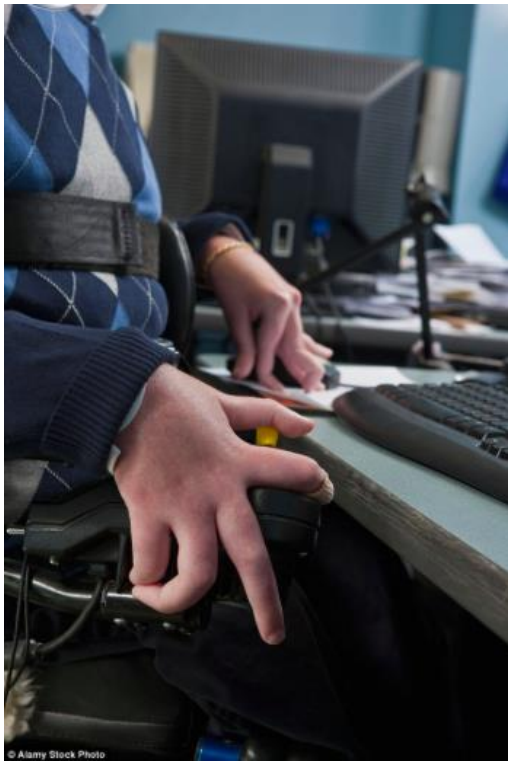
了解该疾病的自然发展史有助于预防骨科畸形的一些并发症

- Some patients are more affected by contractures than others (DMD, Col6RD)-一些病人相较于其他因素而言更容易受到挛缩因素的影响
- Weakness varies from one muscle to another-肌肉的无力往往是从某一块肌肉慢慢的影响到另外的肌肉



- Always an improvement of contractures after the loss of ability to walk-行走能力的丧失往往会使得挛缩的情况进一步恶化
- Help defining the objectives of PMR management and follow up-帮助确定PMR管理和随访的目标





Disease ? (疾病?)

What type of orthopedic deformation is the most important to prevent in this disease ?

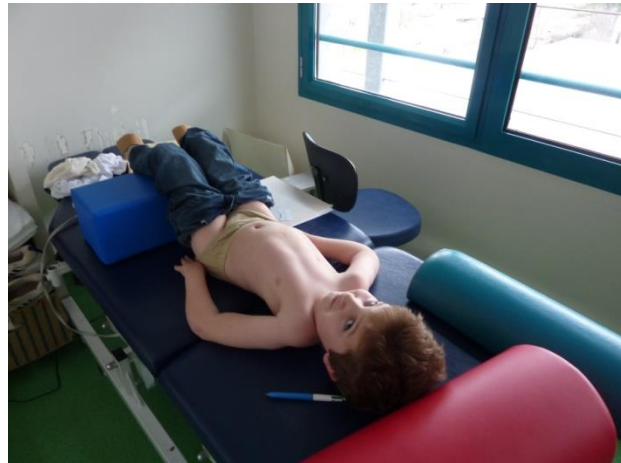
对于这类疾病来说最重要的需要预防的骨骼畸形是什么？



Myopathie de Duchenne



Bushby K et al. Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and pharmacological and psychosocial management. Lancet Neurol 2010; 9:77-93 Lancet Neurol 2010; 9: 177-89



Disease ? (疾病?)

What type of orthopedic deformation is the most important to prevent in this disease ?
对于这类疾病来说最重要的需要预防的骨骼畸形是什么？



Orthopedic management-骨科矫形

Muscle Weakness, Immobility-肌无力、活动能力下降



CONTRACTURES-挛缩



Orthopedic management-骨骼管理

- Harmonious growth is performed by an adequate traction of different balanced muscles on bones-骨骼的协调生长有赖于适当的牵引力，而这些牵引力是有附着在骨骼上不同互相平衡的肌肉产生。
- 2 consequences in NMDs-2种结果
 - Muscle Weakness （肌无力）
 - Immobility （活动能力下降）
- The best compromise between:最好的折中解决办法
 - The largest possible mobilization for-最大关节活动度
 - Keeping a good trophic status-保证良好的营养
 - Decreasing pain associated with contractures -挛缩牵伸时减少疼痛
 - Osteoporosis-骨质疏松症
 - Immobilization in good position to-保持良好的固定体位
 - Avoid retractions-避免皱缩
 - Prevent deformities-预防畸形

Orthopedic deformities-骨骼畸形

- Immobility with joints consequences
(活动下降导致的关节后果)
- Muscle imbalances around the joint :
(关节周围肌群肌力不平衡)
 - Hamstring/4iceps : Knee flexum
(腘绳肌/股四头肌:膝反张)
 - Triceps/Tibialis anterios: Varus Equinus
小腿三头肌/胫骨前肌: 马蹄内翻足
 - Tensor fascia Latae muscle /hip abductor: frog leg position
(阔筋膜张肌/髋外展肌-臀中肌: 蛙式位)
- Installation-配置
- Variability caused by different disease and retractil potential
(不同疾病和肌肉伸缩导致的易变性)
 - +++ Duchenne-杜氏+++
 - Congenital Muscular Dystrophy-先天性肌营养不良



PASSIVE PHYSICAL THERAPY(被动的物理治疗)

■ Passive Mobilizations-被动关节活动

- All joints-全关节
- Complete range of motion (全范围关节活动)
- Short hand taking
- ++ **Spinal Deformities** (++ 脊柱畸形)



■ Stretching-牵伸

- +++ **Hip, Trunk** for non ambulant-髋、躯干，不可行走的患者
- ++++ **Foot, Knees** for ambulant-足踝及膝盖，可行走的患者
- Don't forget upper limb before and after loss of ability to walk (行走功能丧失前后都不能忘了上肢)
- Carefully because of hypotonia and pain susceptibility-注意肌张力低下及疼痛敏感型患者

ACTIVE PHYSICAL THERAPY

主动的物理治疗

Consider adapted physical activity as a way of life
将调整后的体能活动当成生活的一部分

- Spending time, creating the need
并为此花时间, 创造需求
- Imagine the possibilities and access
想象可能完成的活动并且努力的去完成
- Diversified practices = social role
从自己所扮演的社会角色中获取各种各样的活动锻炼

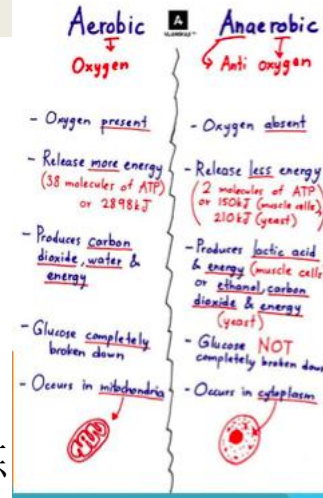
Set clear rules and realistic goals 给自己定下明确的规则和实际的目标

- Palliative treatment \neq curative - 保守治疗 (药物治疗) \neq 有疗效
- Fight against the progression of the disease \neq gain in strength - 延缓疾病进展 \neq 增强力量
- Stabilize and improve function with consequences in terms of autonomy - 自我坚持锻炼治疗并且努力改善功能状况
- Raised in progressive load – 逐渐增加运动量及运动强度

Program and personalized adjustments - 运动方案和个性化的调整

- **Dynamic and concentric work** - 动态及向心的运动
- **Low initial intensity** (balneotherapy < active aid < weight against < resistance) – 低强度开始 (水疗 < 减重 < 协助主动 < 抗重力 < 抗阻力)
- **High frequency** (3 times / week with a day off between) – 高频率 (3次/周 隔天1次)
- **Oriented on the important functions to be preserved** (equilibrium, walking, transfers, turning)
维持重要的功能 (平衡、步行、转移及转向)

EXERCISE AEROBIE VS EXERCICE ANAEROBIE



Aerobic exercises

- Presence of oxygen
- Moderate intensity
- Long duration
- Develops stamina
- Burns calories during the activity



Anaerobic exercises

- Absence of oxygen
- High intensity
- Short duration
- Develops force
- Burns calories even when the body is at rest



ACTIVE PHYSICAL THERAPY

主动的物理治疗

■ **Regular Evaluations** toward a progressive autonomisation of the patient - 针对患者的变化要做定期的评估

■ **Progressive Demedicalisation with warnings**
主动运动过程中的注意事项

Attention to exercises of high intensity or duration

注意高强度运动及时间

Attention to overuse syndrome

注意超负荷症状

Recognize the threshold of fatigue not to be crossed


记住疲劳的阈值，不能训练过度

EXERCISE AEROBIC VS EXERCISE ANAEROBIC

Aerobic	Anaerobic
<p>Oxygen</p>	<p>Anti oxygen</p>
<p>- Oxygen present</p>	<p>- Oxygen absent</p>
<p>- Release more energy (38 molecules of ATP or 2898kJ)</p>	<p>- Release less energy (2 molecules of ATP or 150kJ (muscle cells), 210kJ (yeast))</p>
<p>- Produces carbon dioxide, water & energy</p>	<p>- Produces lactic acid & energy (muscle cells) or ethanol, carbon dioxide & energy (yeast)</p>
<p>- Glucose completely broken down</p>	<p>- Glucose NOT completely broken down</p>
<p>- Occurs in mitochondria</p>	<p>- Occurs in cytoplasm</p>


Aerobic exercises

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Anaerobic exercises

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Physical activity as efficient on strength loss, fatigability, pain, quality of life

“Recent studies have reported that progressive resistance strengthening programs for lower extremities are feasible, safe, beneficial, and improve exercise intolerance and undue fatigue in patients with NMD can improve walking function, activities of daily living, and subjective perception of pain and fatigue in patients”

CMT [Tajima F Brain 2016] [El Mhandi L 2003]

DMD [de groot 2015] [Jansen M 2013]

FSDH [Andersen G 2015]

[Neurorehabil Neural Repair](#). 2013 Nov-Dec;27(9):816-27. doi: 10.1177/1545968313496326. Epub 2013 Jul 24.

Assisted bicycle training delays functional deterioration in boys with duchenne muscular dystrophy: the randomized controlled trial "no use is disuse".

[Jansen M¹](#), [van Alfen N](#), [Geurts AC](#), [de Groot LJ](#).

⊕ Author information

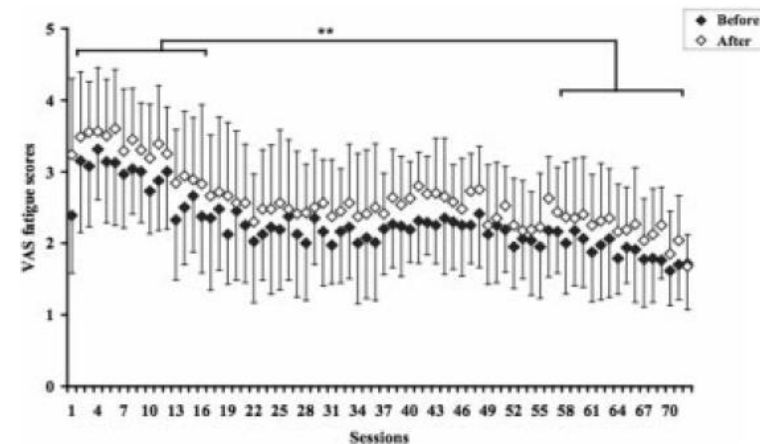
Abstract

BACKGROUND: Physical training might delay the functional deterioration caused by disuse in boys with Duchenne muscular dystrophy (DMD). The "No Use Is Disuse" study is the first explorative, randomized controlled trial in boys with DMD to examine whether assisted bicycle training is feasible, safe, and beneficial.

METHODS: Ambulatory and recently wheelchair-dependent boys with DMD were allocated to the intervention or control group. The intervention group received assisted bicycle training of the legs and arms during 24 weeks. The control group received the same training after a waiting period of 24 weeks. The primary study outcomes were the Motor Function Measure (MFM) and the Assisted 6-Minute Cycling Test (A6MCT). Group differences were examined by an analysis of covariance.

RESULTS: Thirty boys (mean age 10.5 ± 2.6 years, 18 ambulant and 12 wheelchair-dependent) were allocated to the intervention (n = 17) or the control (n = 13) group. All boys in the intervention group (except one) completed the training. After 24 weeks, the total MFM score remained stable in the intervention group, whereas it had significantly decreased in the control group ($\Delta = 4.9$, 95% confidence interval = 2.2-7.6). No significant group differences were found for the A6MCT. No serious adverse events were observed.

CONCLUSIONS: Our results suggest that assisted bicycle training of the legs and arms is feasible and safe for both ambulant and wheelchair-dependent children and may decline the deterioration due to disuse. Progressive deterioration, however, may compromise the design of trials for DMD.



[El Mhandi L 2003]



Physical activity as efficient on strength loss, fatigability, pain, quality of life 针对如何增强肌力, 改善疲劳状况、疼痛和生活质量的物理治疗活动

最近研究报道得出针对下肢的患者使用渐进式强化疗法是可行的, 安全的, 有效的。同时此疗法可改善患者运动耐受不良及疲劳状况, 对行走功能, ADL能力的提高, 疼痛敏感和疲劳状况都有一定的疗效。

CMT [Tajima F Brain 2016] [El Mhandi L 2003]

DMD [de groot 2015] [Jansen M 2013]

FSHD [Andersen G 2015]

[Neurorehabil Neural Repair](#). 2013 Nov-Dec;27(9):816-27. doi: 10.1177/1545968313496326. Epub 2013 Jul 24.

Assisted bicycle training delays functional deterioration in boys with duchenne muscular dystrophy: the randomized controlled trial "no use is disuse".

[Jansen M¹](#), [van Alfen N](#), [Geurts AC](#), [de Groot LJ](#).

⊕ Author information

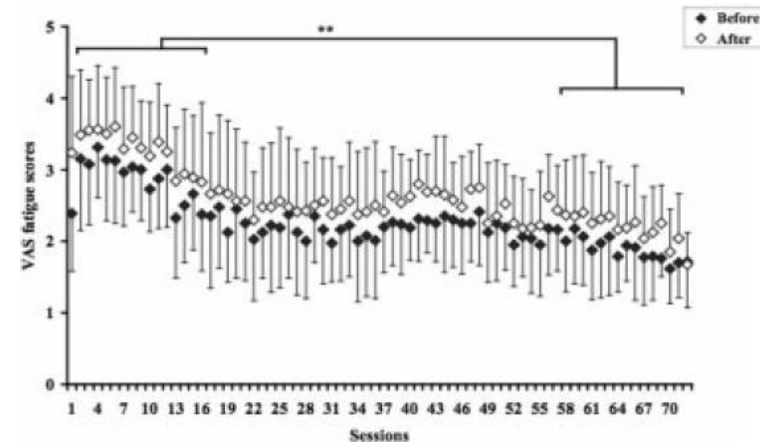
Abstract

BACKGROUND: Physical training might delay the functional deterioration caused by disuse in boys with Duchenne muscular dystrophy (DMD). The "No Use Is Disuse" study is the first explorative, randomized controlled trial in boys with DMD to examine whether assisted bicycle training is feasible, safe, and beneficial.

METHODS: Ambulatory and recently wheelchair-dependent boys with DMD were allocated to the intervention or control group. The intervention group received assisted bicycle training of the legs and arms during 24 weeks. The control group received the same training after a waiting period of 24 weeks. The primary study outcomes were the Motor Function Measure (MFM) and the Assisted 6-Minute Cycling Test (A6MCT). Group differences were examined by an analysis of covariance.

RESULTS: Thirty boys (mean age 10.5 ± 2.6 years, 18 ambulant and 12 wheelchair-dependent) were allocated to the intervention ($n = 17$) or the control ($n = 13$) group. All boys in the intervention group (except one) completed the training. After 24 weeks, the total MFM score remained stable in the intervention group, whereas it had significantly decreased in the control group ($\Delta = 4.9$, 95% confidence interval = 2.2-7.6). No significant group differences were found for the A6MCT. No serious adverse events were observed.

CONCLUSIONS: Our results suggest that assisted bicycle training of the legs and arms is feasible and safe for both ambulant and wheelchair-dependent children and may decline the deterioration due to disuse. Progressive deterioration, however, may compromise the design of trials for DMD.



[El Mhandi L 2003]



Braces-辅具

- Progressively function of age and disease progression
(根据患者的年龄和疾病进展情况作出可靠的预期目标设定)
- Objectives-目标
 - Installation patient comfort-使患者舒适
 - Orthopedic prevention : splint-预防挛缩：支具
 - Function Improvement : FAO (Foot ankle orthosis)-功能性辅助器具：踝足矫形器
- Preventing deformities with functional consequences
Preservation Walking, sitting Station, FRE of Conduct, dressing ..
通过功能性活动预防畸形，如保持行走、坐姿、FRE行为、穿衣等
- All joints could be concerned by braces :Foot, Arms and wrists
Hips and pelvis, Spine
所有关节均可以考虑使用支架保护：足踝、手臂、手腕、髋、骨盆及脊柱



SPLINT

Use and tolerability of a side pole orthosis in preventing deformation in the frontal plane of the children foot with neurological disorders

支具：单侧支持矫形器的使用及耐受性对神经肌肉疾病的患者在预防足踝部额状面的变形有非常重要的作用



[Delvert 2016 J of ISPO]

Risk of hip luxation and contractures

髋关节脱位及关节挛缩的风险

- Adequate position of lower limb during lying position (卧位时下肢的良好体位摆放)
- Avoid frog leg position : contractures in TFL et anterior luxation of the hip-避免蛙式体位：阔筋膜张肌挛缩，髋关节前脱位
 - ➔ Severe consequences in terms of Sitting position, Pain and quality of life-导致坐姿、疼痛、生活质量等方面的严重后果

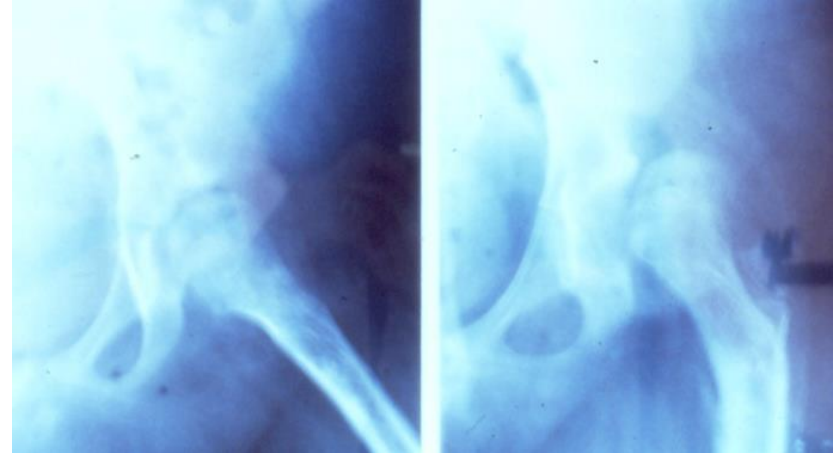


Standing position for non ambulant patients

无行走功能的患者的站立维持

■ Advantages-优点

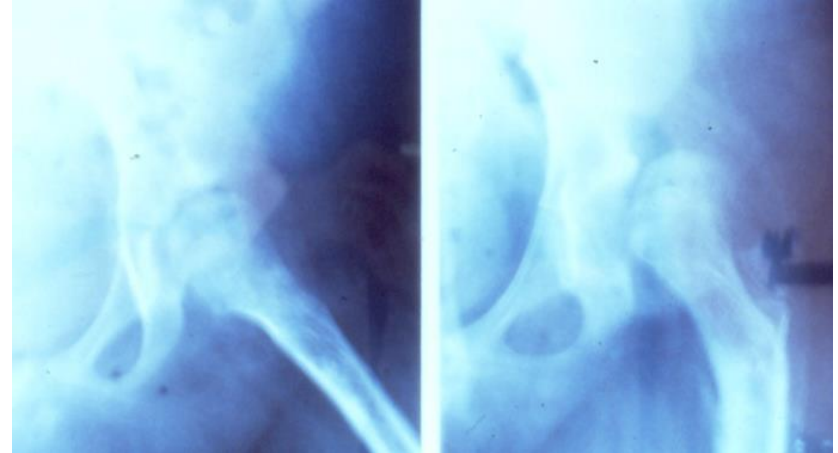
- Orthopedic deformities : Prevention of hip luxation ? Stretching-骨骼畸形：预防髋关节脱位？牵伸
- Preventing osteoporosis [Glickman, Geigle and Paleg, 2010] {Pin, 2007}-预防骨质疏松症
- Effects on pulmonary function and digestion-影响肺功能和消化功能
- Better Social position to improve interaction with others更好的功能体位更加有助于患者于他人的交流



Standing position for non ambulant patients

无行走功能的患者的站立维持

- more frequent standing/loading several times per day may be more osteogenic than less frequent-每天多站或负重会减少骨质疏松
- for standing in children has given further clarification of the best evidence for standing duration and frequency (Paleg, Smith, & Glickman, 2013)-对于儿童站立的时间和频率已经有很好的证据
- five days per week is necessary.-一周五次是有必要的
- During each standing session-每次站立时要求
 - a minimum of 60 minutes to affect bone density and hip stability.-每次至少60分钟以上才能影响骨骼生长和髋关节的稳定
 - a minimum of 45 minutes to improve Range of motion in the lower extremities,-站立45分钟可以改善下肢关节的活动度
 - while spasticity reduction may occur with as little as 30 minutes per session of standing-要想减少痉挛每次至少站立30分钟。



Orthopedic lower limbs surgery ? 下肢的骨科手术

Ambulant Patients : Rare indications of orthopedic management to avoid premature loss of ambulation-可步行的患者：有明显的骨科手术指征才可以做手术以避免过早的丧失步行的能力

■ Promote Casts instead of foot surgery-提倡以打石膏替代足部的骨科手术

The results of this pilot study suggest that serial casting of the TAs can be considered in DMD patients with moderate TA contractures and in whom there are no significant iliotibial band (ITB) tightness, although this intervention is probably not as effective as surgery in maintaining long-term TA range

[Main M1, Mercuri E, Haliloglu G, Baker R, Kinali M, Muntoni F](#). Serial casting of the ankles in Duchenne muscular dystrophy: can it be an alternative to surgery? *Neuromuscul Disord*. 2007 Mar;17(3):227-30. Epub 2007 Feb 15.

[Glanzman AM1, Flickinger JM, Dholakia KH, Bönemann CG, Finkel RS](#). . Serial casting for the management of ankle contracture in Duchenne muscular dystrophy. *Pediatr Phys Ther*. 2011 Fall;23(3):275-9

■ Discouraging Long term effect-长期效果不肯定

Hip and ankle surgery can release contractures. In the knees, surgery should be restricted to cases with knee flexion greater than 30°. The best results are obtained with extensive m. tensor fascia lata tenectomy and posterior tibial tendon transfer. Achilles tendon tenotomy results in better recovery of dorsal flexion. Surgery is indicated when contractures are severe and the patient is no longer able to walk. The aim should not be to continue walking but to release contractures, so that the patient can continue assisted upright positioning as long as possible. Especially in cases of asymmetric contractures, surgery can help slow the progression of scoliosis.

Griffet J, Decroq L, Rauscent H, Richelme C, Fournier M. **Lower extremity surgery in muscular dystrophy**. *Orthop Traumatol Surg Res*. 2011 Oct;97(6):634-8



Lower limbs surgeries : Rarely

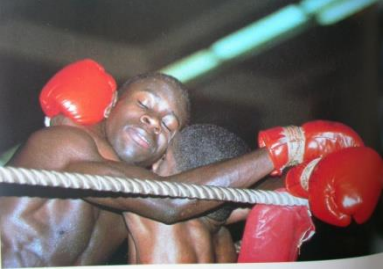
下肢手术：极少

Non ambulant patients : Sometimes-不能行走的患者：有时

**Prevention +++ to avoid Pelvis Obliquity (避免骨盆倾斜)
, hip luxation (髋关节脱位) and difficulties for sitting
position (坐姿维持困难)**

Pelvis obliquity-骨盆倾斜

- Low Origin 下部原因
 - Contractures of adductors, Fascia Lata Tensors-内收肌及阔筋膜张肌的挛缩
- High origin上部原因
 - Lumbar scoliosis-腰椎侧弯
- Both origins +++两者



Foot surgery for non ambulant patients with NMD: almost never

神经肌肉疾病不能行走的患者进行足部手术的：几乎没有

Controverse : Faut-il opérer les pieds dans la dystrophie musculaire Duchenne de Boulogne ?

V. CUNIN, P. WICART

Introduction

Les principales préoccupations médicales face à un enfant ayant une dystrophie musculaire congénitale de Duchenne de Boulogne (DDB) sont la dégradation de la fonction cardio-respiratoire et l'apparition puis l'aggravation d'une déformation du rachis. Cependant, les déformations des membres inférieurs et particulièrement des pieds, avec un équin et des composantes plus ou moins importantes de varus et d'adduction [1, 2], sont intriquées avec la perte progressive de la marche (fig. 1). Les indications et modalités thérapeutiques des déformations des pieds ne sont pas consensuelles. Philippe Wicart expose des éléments en faveur du traitement chirurgical des déformations des pieds alors que Vincent Cunin argumente l'inverse.

Arguments en faveur de la chirurgie

Il existe des indications à la chirurgie des déformations des pieds dans la dystrophie musculaire congénitale de DDB. Au préalable, il convient d'avoir établi le diagnostic avec certitude.



Fig. 1 : Déformation typique des membres inférieurs chez un enfant âgé de 8 ans venant de perdre la marche.

Very carefully-警惕

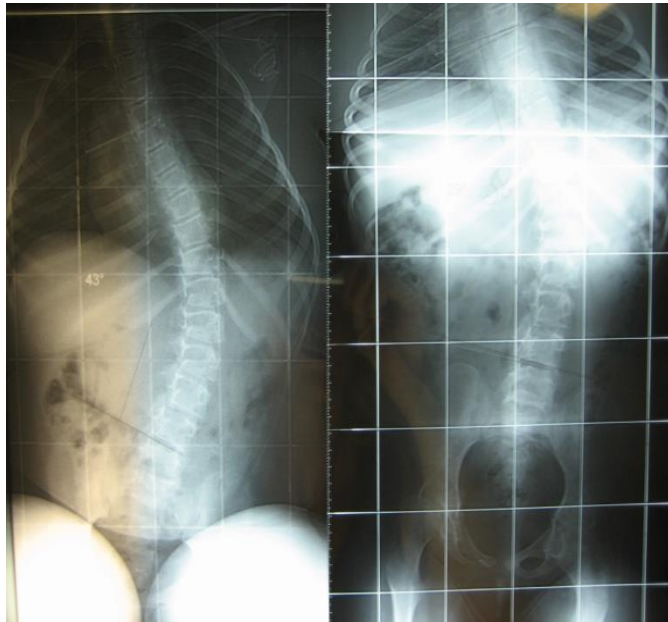
-Because no evidence in international littérature on long term benefice after surgery in term of comfort, pain and quality of life
还没有针对此种手术过后有关如何摆放舒适体位，改善疼痛和提高生活质量的国际共识

-Recurrence +++-反复出现



Spine deformities : Neurological scoliosis 脊柱畸形：神经源性脊柱侧凸

- Early beginning in infancy (SMA, DMC)
早期婴幼儿发育时期就会产生 (SMA,DMC)
- Continue to worsened even after puberty
青春期后会持续恶化
- Consequences in term of pulmonary function
会导致肺功能障碍
- Consequences in term of sitting position, comfort, and pain
最终会出现影响坐姿，舒服度和产生疼痛



Disease progression and effect of spine deformation

疾病进展和脊柱畸形产生的影响

Dev Med Child Neurol. 2006 Jun;48(6):513-8.

Management of scoliosis in Duchenne muscular dystrophy: a large 10-year retrospective study.

Kinali M, Messina S, Mercuri E, Lehovsky J, Edge G, Manzur AY, Muntoni F.

Department of Paediatrics, Dubowitz Neuromuscular Centre, Hammersmith Hospital Campus, Imperial College London, UK.

- **DMD:** 75 à 90% before steroids
15 à 25% < 30°
- Scoliosis-脊柱侧凸
 - ≥ 10° 在 85/88 patients (97%)
 - ≥ 20° 78/88 patients (89%)
 - ≥ 30° 在 66/88 patients (75%)



■ SPINE

Progression of spinal deformity in wheelchair-dependent patients with Duchenne muscular dystrophy who are not treated with steroids

F. Shapiro,
D. Zurakowski,
T. Bui,
B. T. Darras

CORONAL PLANE (SCOLIOSIS) AND SAGITTAL PLANE (KYPHOSIS, LORDOSIS) DEFORMITY

Effect of Cortico steroids on scoliosis prognosis : 90% vs 20%

皮质激素对脊柱侧凸患者预后的影响

[Biggar WD et al. *Neuromuscul Disord* 2006]

[Manzur AY et al. *Cochrane Database Syst Rev* 2008]

[Moxley RT et al. *J Child Neurol* 2010]

- High prevalence in non ambulant **SMA** : 98,5% des cas
- 在不能步行的SMA患者中是高发生率

Rachis. 1991, vol. 3, n° 5, p. 439 - 447

ARTICLE ORIGINAL

AMYOTROPHIE SPINALE INFANTILE
ET SCOLIOSE

INFANTILE SPINAL MUSCULAR ATROPHY
AND SCOLIOSIS

PH. COSSON, L. MILADI, R. SERINGE, J. DUBOUSSET

Spinal deformities-脊柱畸形

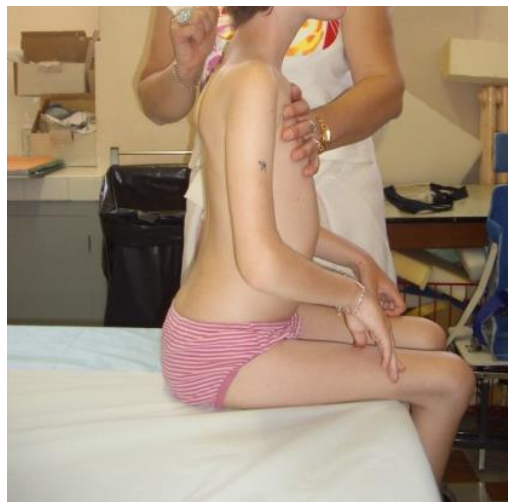
Thoraco lumbar Sagital deformities : reductibility, flexibility of the deformities-胸腰椎矢状位的脊柱畸形：可还原性和可塑性

Lordosis: Contracture in paraspinal muscles

前凸：脊椎旁肌群肌肉挛缩

Kyphosis : Weakness in paraspinal muscles

后凸：脊椎旁肌群肌力较弱



Spinal deformities-脊柱畸形

Neck Sagital deformity （颈部矢状位畸形）

Hyperextension （颈部过伸）

Even after spinal fusion ++ DMD （即使脊柱融合术后++ DMD ）

Promote by weakness of hip extensor, neck flexor and extensor

（可通过放松髋伸肌和颈部屈伸肌改善）

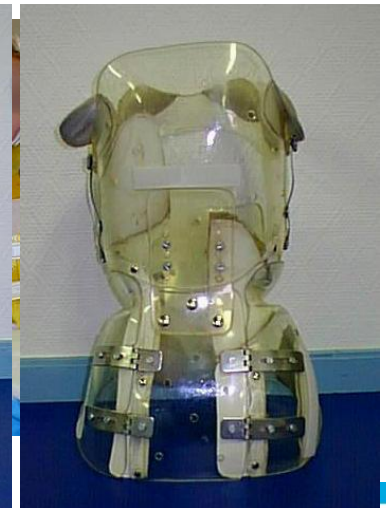
Prevention : promote sitting position against the back support

（预防：通过背部支持改善坐姿）



Trunk Brace (躯干支架)

- Garchois Brace
- For SMA, CMD and CM
(适用于SMA, CMD and CM)
- Prevention and treatment of neurological scoliosis particularly for children with respiratory distress and severe axial hypotonia
- (预防和治理神经性脊柱侧凸, 特别是伴有呼吸窘迫和背部张力过低的儿童)
- Advantages (优点)
 - Improve Function : transfers and installations
(改进功能: 转移和安装)
 - Help maintaining flexibility (帮助维持灵活性)
 - Prevent and improve prognosis of scoliosis
(预防和改善脊柱侧凸预后)
 - No support on the chest [Morillon S 2007]
(胸部无支撑 [Morillon S 2007])
 - Allow a chin and head support
(可支撑下颏和头部)



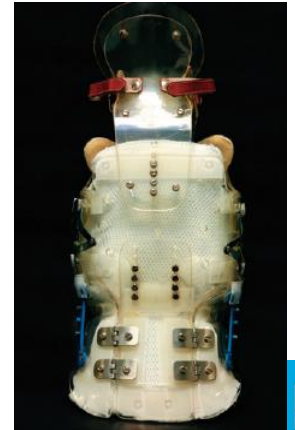
Garchois Brace

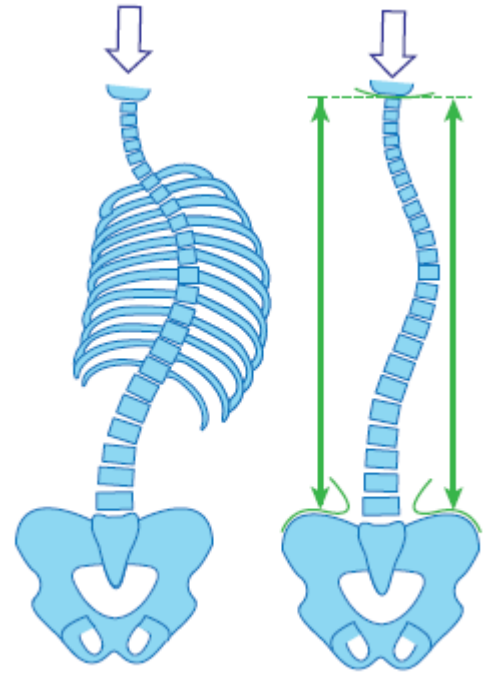
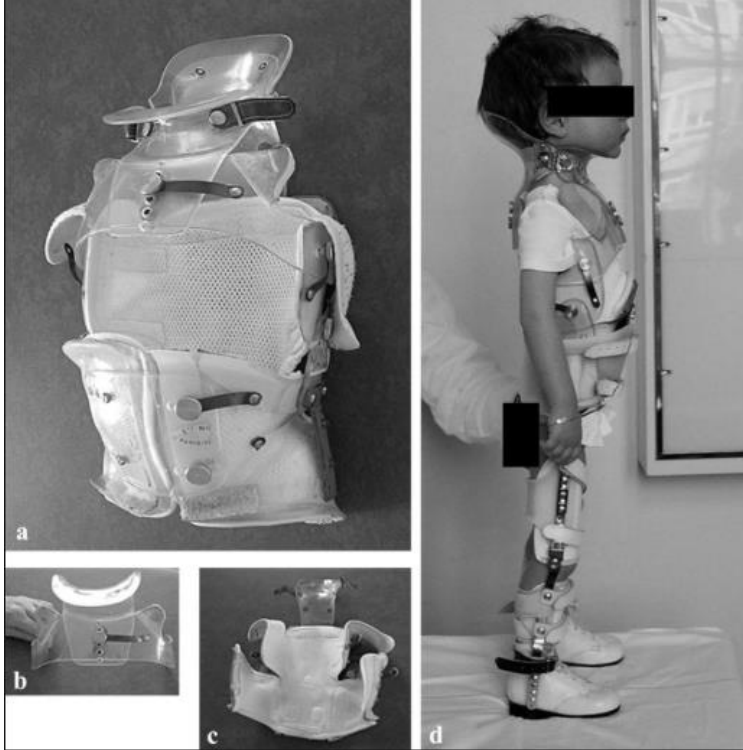
4 modules+ head and chin support (Plexidur®):

(4个模块+头部和下颌支撑 (Plexidur®))

- **1 lumbo sacral posterior valve** (du coccyx au épines des omoplates)
1 骶骨后阀 (从尾骨到肩胛骨)
- **2 ilio abdominal half-valves** (2.髂腹部半阀)
 - Support on iliac crest (支持髂嵴)
 - Cover the abdominal muscle (覆盖腹肌)
 - Slightly go beyond the lower edge of the last rib
(稍过最下端肋骨下缘)
 - Joined to the posterior valve by hinges (opening phenomenon as a book) (通过铰链连接到后瓣 (像打开书一样))
 - Take the basin and immobilized (取盆固定)
- **1 pre sternal support under** the clavicle (1. 锁骨下胸前支持)
- **No support on the chest allowing free mobility** (胸部无支撑允许自由活动)
- **1 head and chin support** (头部和下颌支撑)

Easy to set up (易于设置)
Changeable during the growing
(生长期可调节)





Garchois Brace

No impact on respiratory function

(Less than 10%)

对呼吸功能无影响 (少于10%)



The Spine Journal 9 (2009) 981–986

THE
SPINE
JOURNAL

Clinical Study

Effects of scoliosis on respiratory muscle strength in patients
with neuromuscular disorders

Deniz Inal-Ince, PhD, PT^{a,*}, Sema Savci, PhD, PT^a, Hulya Arikan, PhD, PT^a,
Melda Saglam, MS, PT^a, Naciye Vardar-Yagli, MS, PT^a, Meral Bosnak-Guclu, PhD, PT^a,
Deniz Dogru, MD^b

^aDepartment of Physical Therapy and Rehabilitation, Faculty of Health Sciences, Hacettepe University, 06100 Sımanpazarı, Ankara, Turkey
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ANNALES
de RÉADAPTATION
et de MÉDECINE PHYSIQUE

<http://france.elsevier.com/direct/ANNRMP/>

Article original

Impact du corset thoracique sur la fonction respiratoire
chez des enfants atteints de maladie neuromusculaire

Effect of thoracic bracing on lung function
in children with neuromuscular disease

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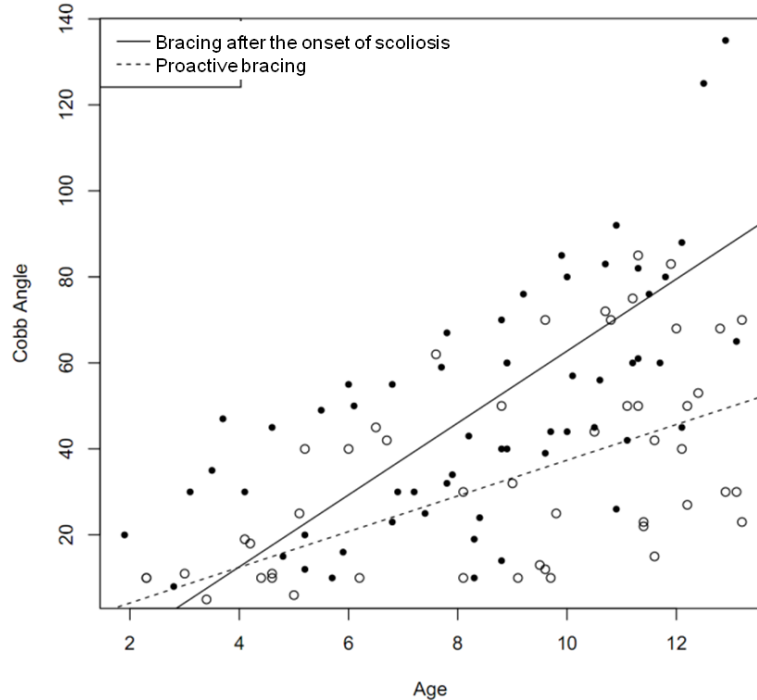
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Reçu le 14 décembre 2006 ; accepté le 21 mars 2007

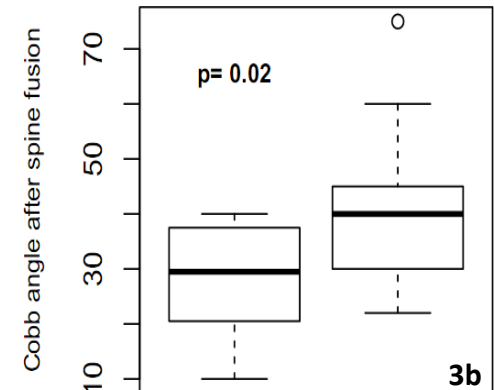
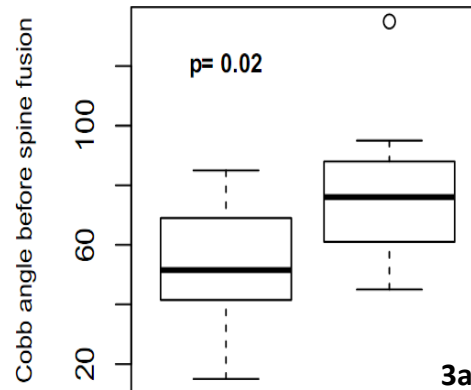


Preventive effect of Garchois Brace on scoliosis and prognosis (Garchois Brace 对脊柱侧弯的预防和预后)



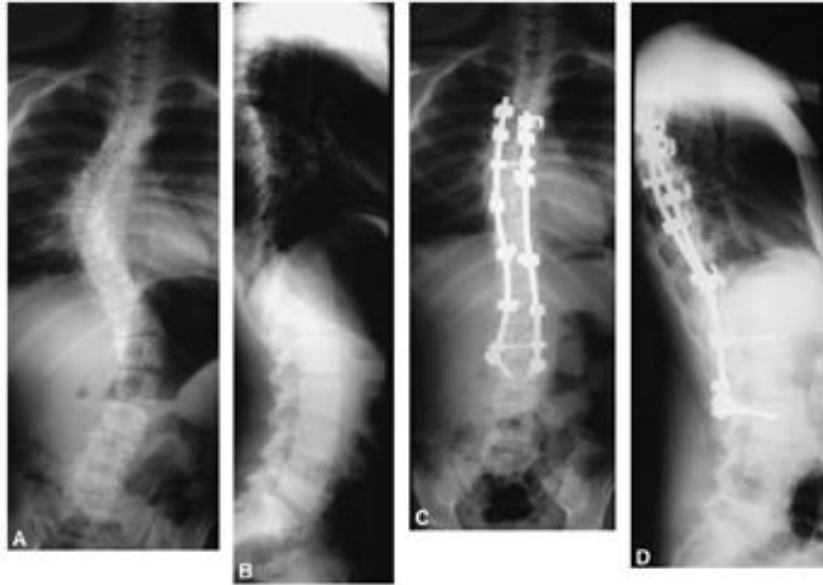
[Catteruccia et al 2016
J Neuromuscul Disease]

- Mean preoperative and postoperative Cobb angle lower in the group 1 of proactively braced than in group 2 or 3 (Anova $p=0.03$; Kruskal Wallis test $p=0.05$).
(平均术前和术后Cobb角在主动支撑组1中低于组2或3 (Anova $p = 0.03$; Kruskal Wallis 检验 $p = 0.05$))
- Better surgical results were observed in patients with a minor preoperative Cobb angle ($r=0.92$ $p<0.0001$). Moreover, major complications tend to be less in the group 1 proactively braced.
(术前Cobb角较小的患者可观察到更好的手术结果 ($r = 0.92$ $p < 0.0001$)。此外, 主动支撑组1的主要并发症更少。)



Spinal Fusion (脊柱融合)

■ Classical



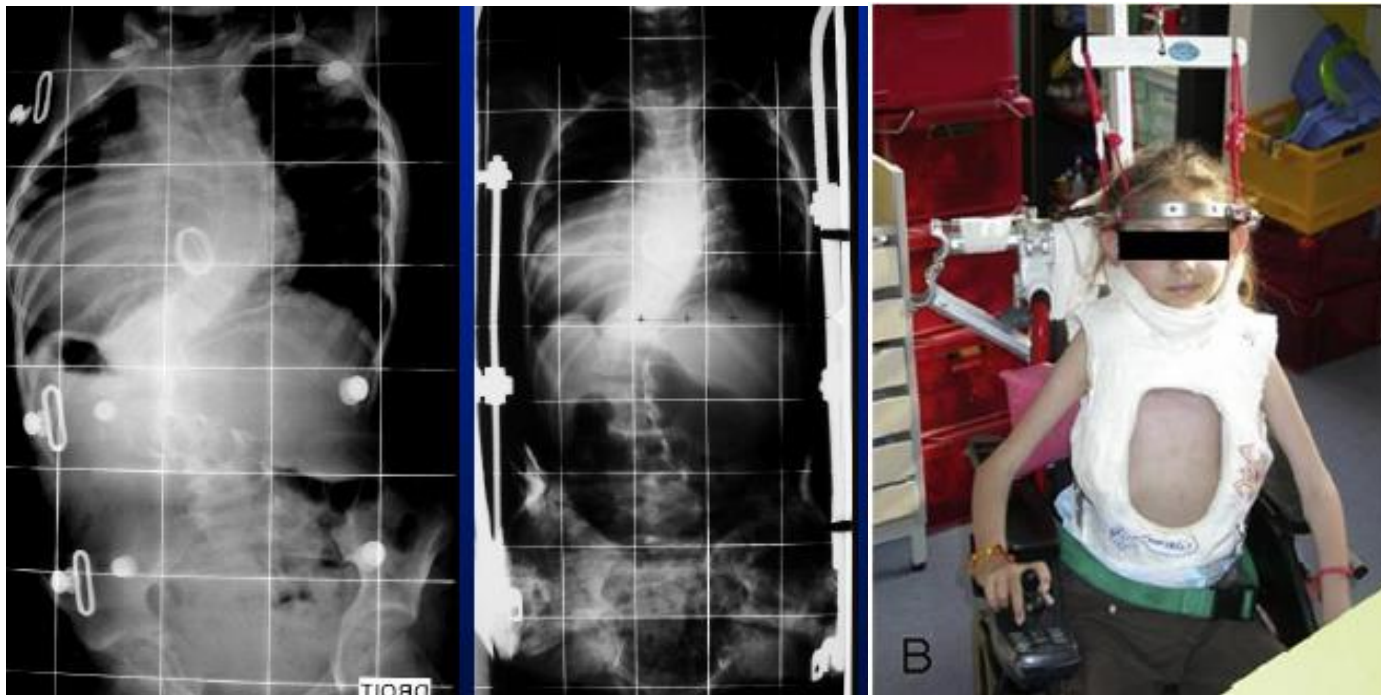
Need to wait until Bone Maturity (需要等到骨成熟)

Post access +/- anterior (后进入 +/- 前)

SMA +++, DMC ++ DMD ++ (less with cortico steroids) and CM

(SMA +++, DMC ++ DMD ++ (少于皮质类固醇) 和CM)

- Pre operative traction by halo (术前牵引) :



Images Pr Griffet – orthopédie Grenoble/ Dr Vialle Paris

- Perspectives : use of growing rods allowing early surgery in case of early severe scoliosis (观点：早期手术使用生长棒以防严重侧弯)



Spine

DEFORMITY

SPINE Volume 36, Number 16, pp 1305-1311
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Growing Rods for Scoliosis in Spinal Muscular Atrophy

Structural Effects, Complications, and Hospital Stays

Mark J. McElroy, MS,* Adam C. Shaner, BS,* Thomas O. Crawford, MD,† George H. Thompson, MD,‡
Rishi V. Kadakia, BS,§ Behrooz A. Akbarnia, MD,§¶ David L. Skaggs, MD,|| John B. Emans, MD,** and
Paul D. Sponseller, MD,*



Improve mobility to improve autonomy (改善活动以提高主动性)

- Technical aids (技术帮助)
- Power wheelchair from 2 yo
(电动轮椅)
- ++



Tricky period:

A change in term of prognosis in many NMD
but still no cure and a lot of hopes (棘手的时期：
许多NMD的预后有所变化，但仍无治愈方法，仅有很多希望)

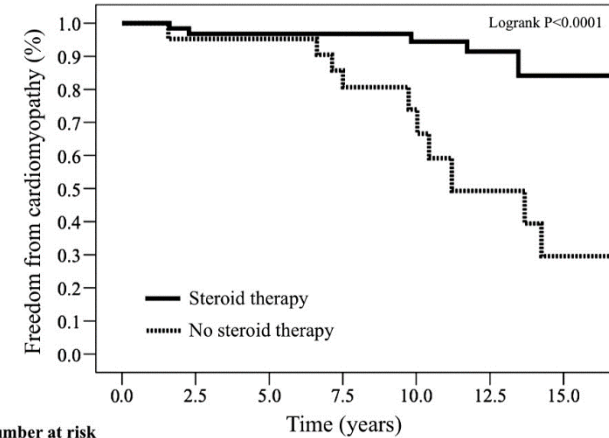


Thanks to orthopedic,
nutritional, cardiologic,
respiratory management

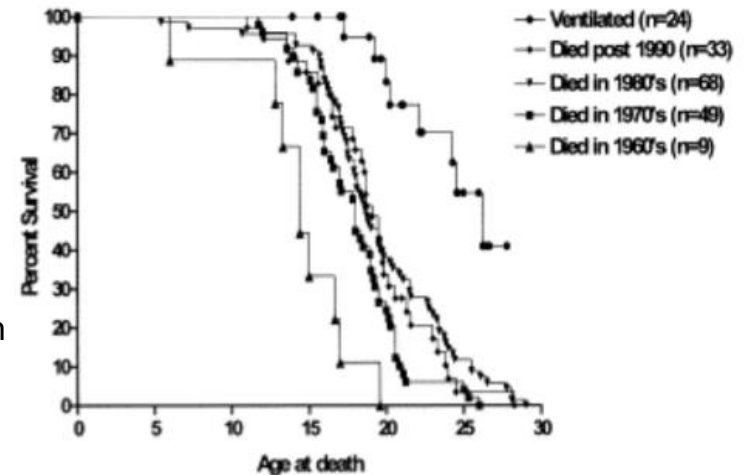
In terms of quantity of life

In terms of quality of life and
participation

(由于有效的骨骼，营养，心血管和呼吸管
理，患者的生活质量和参与方面均有所改变)



	0.0	2.5	5.0	7.5	10.0	12.5	15.0
Steroid therapy	63	59	57	51	39	26	3
No steroid therapy	21	20	20	17	10	5	2



Michelle Eagle : Survival in Duchenne muscular dystrophy: improvements in life expectancy since 1967 and the impact of home nocturnal ventilation



Thank you for your attention
Carole.vuillerot@chu-lyon.fr