X JORNADAS NEUROLOGICAS DEL SUR
CONCEPCION 26 y 27 AGOSTO 2016

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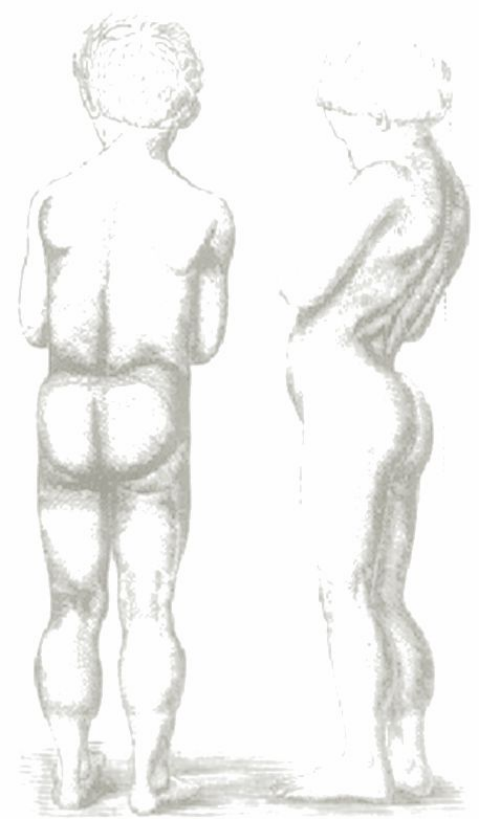

Distrofia Muscular de Duchenne

- Clínica, diagnóstico y diagnóstico diferencial
- **Tratamiento y evolución**

Dra. Karin Kleinsteuber S.

Profesor Asociado, Programa de Formación de Especialistas en Neurología Pediátrica

Universidad de Chile - Clínica Las Condes

Declaración de potenciales conflictos de intereses

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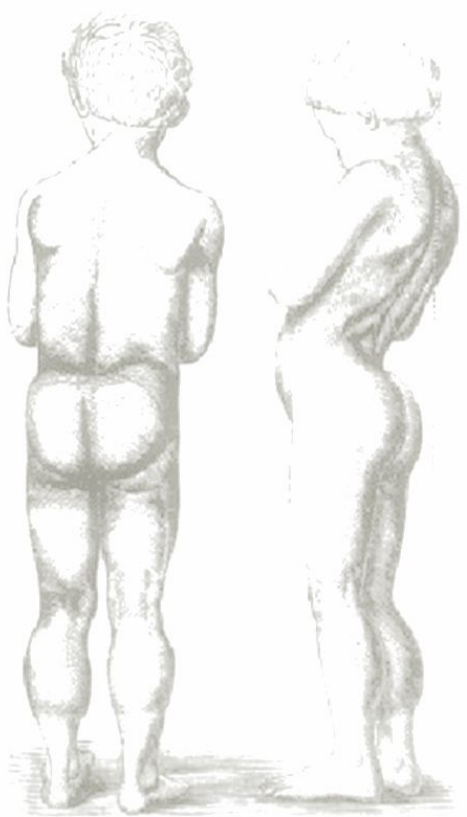
	Declaro no tener ningún potencial conflicto de interés
√	Relativas a esta presentación existen las siguientes relaciones que podrían ser percibidas como potenciales conflictos de intereses:
√	• **Financiada por Laboratorio Genzyme para asistir a: Programa de Entrenamiento en Tratamiento de Enfermedad de Pompe** 2006, Cambridge, Massachusetts y Universidad de Duke , Durham, EEUU: 40 horas/totales. Y **II Simposio Enfermedad de Pompe,** junio 2010, Buenos Aires Argentina: 20 horas/totales
√	• **Principal Investigator :** A Phase III, Randomized, Double Blind, Placebo-controlled Clinical Study to Assess the Efficacy and Safety of GSK2402968 in Subjects With Duchenne Muscular Dystrophy / *A Clinical Study to Assess the Efficacy and Safety of GSK2402968 in Subjects With Duchenne Muscular Dystrophy - ClinicalTrials.gov identifier: NCT01254019* http://www.clinicaltrials.gov/ct2/results?term=+DMD114044 **CONCLUIDO**
√	• **Principal Investigator :** A phase III, multicentre, double-blind, prospective, randomised, placebo-controlled study assessing the efficacy and safety of DYSPORT® used in the treatment of lower limb spasticity in children with dynamic equinus foot deformity due to cerebral palsy. **Y-55-52120-141 Ipsen Clinical Study CONCLUIDO**
√	• **Principal Investigator :** A phase III, prospective, multicentre, open label, extension study assessing the long term safety and efficacy of repeated treatment with DYSPORT® used in the treatment of lower limb spasticity in children with dynamic equinus foot deformity due to **cerebral palsy Y-55-52120-147 Ipsen Clinical Study CONCLUIDO**

Las imágenes mostradas en esta presentación han sido autorizadas por los pacientes y/o sus padres o están disponibles en acceso libre en la red.

Distrofia Muscular de Duchenne.



Evaluaciones necesarias
por etapa de la enfermedad

Seguimiento del paciente con DMD

Neuromuscular

Nutrición

Gastrointestinal

Salud ósea

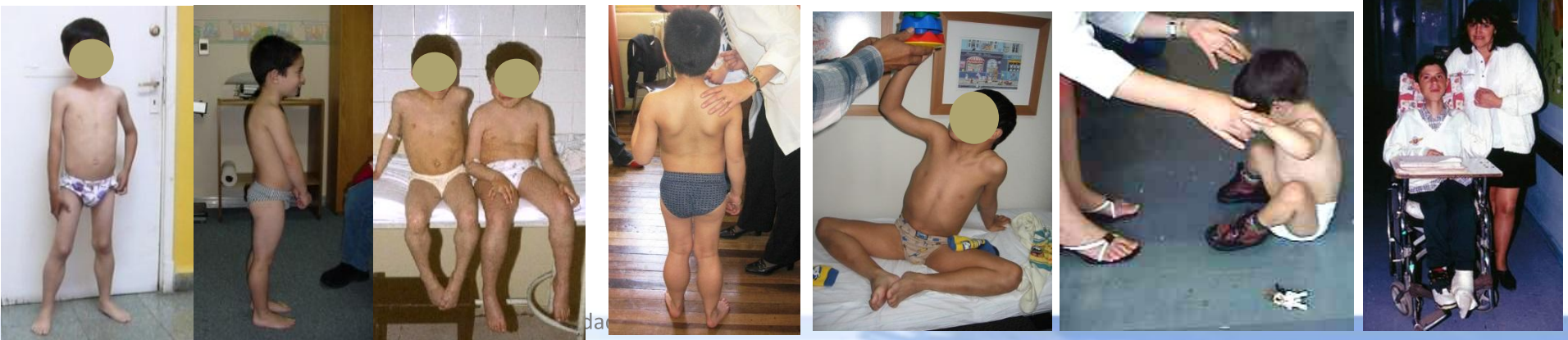
Dra. Karin Kleinsteuber S.

Profesor Asociado, Programa de Formación de Especialistas en Neurología Pediátrica

Universidad de Chile - Clínica Las Condes

Evaluaciones / manejo en DMD en distintas etapas

Etapas	Etapa 1	Etapa 2	Etapa 3	Etapa 4	Etapa 5
	Pre sintomática	Ambulatoria temprana	Ambulatoria tardía	No-ambulatoria temprana	No-ambulatoria tardía
Diagnóstico	Evaluación diagnóstica y consejería genética		Retraso diagnóstico debido a patología concomitante.		
Manejo neuromuscular	"Guía anticipatoria DMD" con plan futuro	Evaluación continua: asegurar que el curso de la enfermedad sea el esperado.			
	Verificar calendario de vacunaciones completo	Evaluación cada 6 meses de función, fuerza y rango de movimiento para definir la fase de la enfermedad, inicio de tratamiento esteroidal y prevención y manejo de efectos adversos.			



MANEJO NEUROMUSCULAR
Qué evaluaciones realizar y porqué ?

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Etapas	Etapa 1	Etapa 2	Etapa 3	Etapa 4	Etapa 5
	Pre sintomática	Ambulatoria temprana	Ambulatoria tardía	No-ambulatoria temprana	No-ambulatoria tardía
Manejo neuromuscular	"Guía anticipatoria DMD" con plan futuro	Evaluación continua para asegurar que el curso de la enfermedad sea el esperado de acuerdo a los estudios diagnósticos.			
	Verificar calendario de vacunaciones completo	Evaluación cada **4- 6 meses** de función, fuerza y rango de movimiento para definir fase de la enfermedad, inicio de tratamiento esteroidal , prevención y manejo de efectos adversos.			

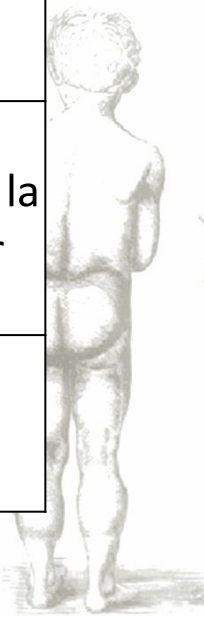
Objetivo:

- **Optimizar manejo, prevenir complicaciones de acuerdo a la evolución y detectar cuando ocurre algo inusual que requiera algún estudio o evaluación adicional.**
- **Indicación de tratamientos en momentos más adecuados.**
- **Protocolos de seguimiento usados en diferentes centros pueden variar.**
- **Control periódico: evaluación clínica: cada 4 a 6 meses.**
- **Lo más importante: control que incluya evaluación clínica + prue… …uan a definir progresión de la enfermedad.**

ADELANTARSE a los problemas

MANEJO NEUROMUSCULAR
Qué evaluaciones realizar?

1	**FUERZA** distintas maneras y secuencias de evaluación / MRC . Importante el registro.
2	**RANGOS ARTICULARES** monitorización de contracturas, definir ejercicios de elongación o intervenciones de mayor ayuda. **EVITAR ASIMETRÍAS.**
3	**VALORACION RUTINARIA CUANTITATIVA** medición de tiempos/ distancias: tiempo para levantarse del suelo, tiempo en recorrer cierta distancia, tiempo para subir varios escalones información de evolución y respuesta a tratamiento.
4	**ESCALAS DE FUNCIÓN MOTORA** gran número de escalas diferentes, IMPORTANTE utilizar de manera rutinaria la misma escala para monitorización sistemática. Diferentes escalas pueden ser necesitadas a diferentes momentos. 6-minute walk test (6MWT)
5	**ACTIVIDADES DE LA VIDA DIARIA** definir necesidad de asistencia en AVD / autonomía



La idea es: mantener fuerza y función mayor tiempo posible

SEGUIMIENTO NEUROMUSCULAR
Qué evaluaciones realizar?

Fuerza muscular

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	Method	Aim of testing	Ambulatory	Non-ambulatory
Strength testing	Manual muscle testing (MRC scale)[58] Quantitative myometry (beneficial if muscle strength 3–5 on MRC scale)*	Serial assessment: to identify outliers from expected clinical course; to monitor disease progression and predict functional losses; to assess response to treatment; and to monitor muscle imbalance	Test lower extremity strength by manual muscle testing every 6 months	Early stages: test upper and lower extremity strength every 6 months Later stages: value of testing is less certain
		integumentary problems To identify need for additional or altered therapeutic/surgical intervention (ie, orthoses, splinting, use of standers, iliotibial band lengthening)		long finger flexors
Timed testing	Standardised use of timed function tests[59,62]	Easy and relevant measure of daily functional status; responsive to change	Timed 10 m walk, timed Gowers' manoeuver, time to climb 4 stairs, time to rise from chair, 6-min walk test Time to put on a shirt might be relevant in late ambulatory stage	Time to put on a shirt might be relevant in early non-ambulatory stage, tim… in late n…
Activities of daily living	A… a… c…		with	Frequency of falls, step activity monitoring, self-care skills, writing, computer use Functioning in school and community settings	Self-care use, cont… wheelcha… Function… commun…
Motor function scales	A… s… s…		n and	Vignos lower extremity scale, North Star Ambulatory Assessment, motor function measure	Brooke u… Egen Kla… assessm… scales, m…

Routine clinic a… st physical and occupational therapy assessments are … 4 months. MR… appropriate assessment tools, they are used more typ… clinical settings.

Table 1: **Suggested neuromuscular assessments for patients with Duchenne muscular dystrophy**

SEGUIMIENTO NEUROMUSCULAR
Qué evaluaciones realizar?

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Fuerza muscular

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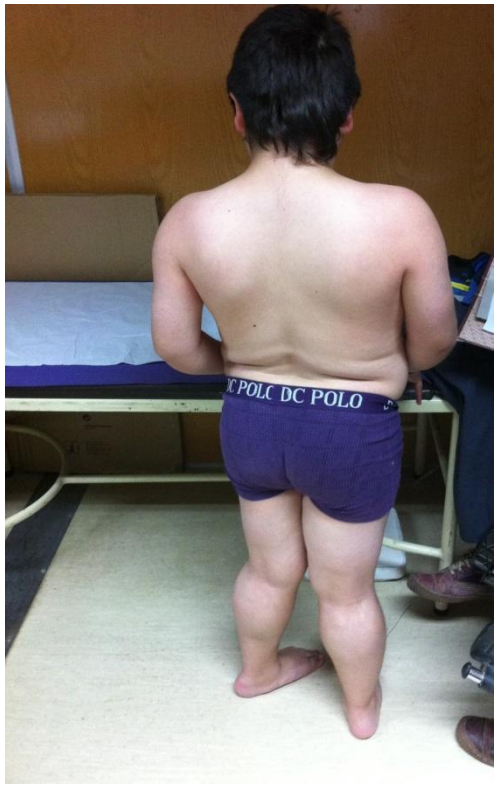

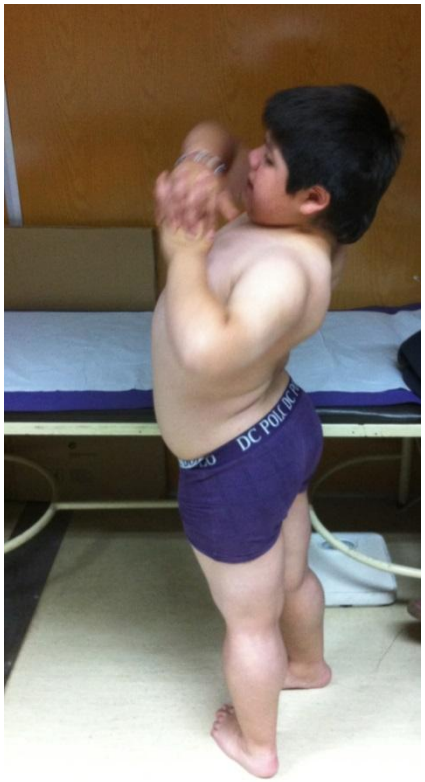

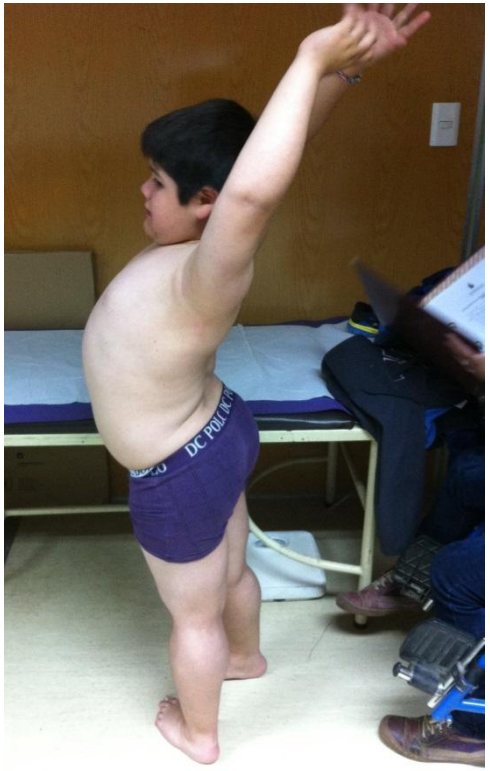

SEGUIMIENTO NEUROMUSCULAR
Qué evaluaciones realizar?

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Fuerza muscular

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MANEJO NEUROMUSCULAR
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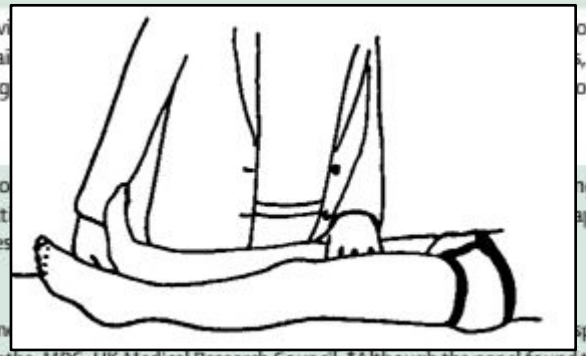
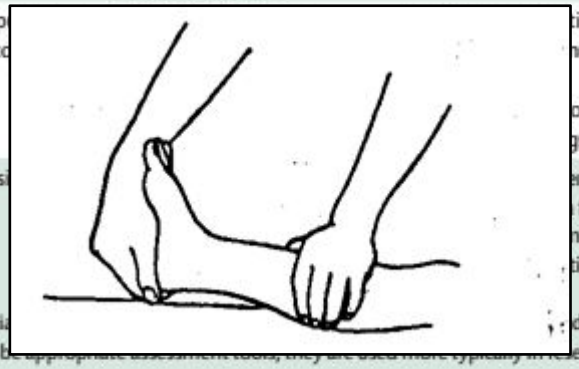
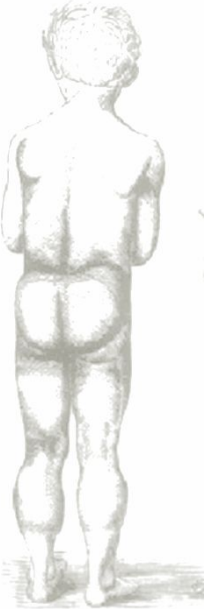
Range of motion	Goniometry[51]	Baseline: to identify emerging muscle hypoextensibility and joint contractures that might contribute/lead to functional deterioration or musculoskeletal or integumentary problems To identify need for additional or altered therapeutic/surgical intervention (ie, orthoses, splinting, use of standers, iliotibial band lengthening)	Lower extremities: hip, knee, ankle joints; iliotibial band; hamstrings, gastrocnemius	Lower extremities: hip, knee, ankle joints; iliotibial band; hamstrings, gastrocnemius Upper extremities: elbow, wrist, long finger flexors
			climb 4 stairs, time to rise from chair, 6-min walk test Time to put on a shirt might be relevant in late ambulatory stage	stage, timed testing not applicable in late non-ambulatory stage

Rangos articulares







4 months. MRC=UK Medical Research Council. *Although the panel found these tests to be clinical settings.

Table 1: **Suggested neuromuscular assessments for patients with Duchenne muscular dystrophy**

MANEJO NEUROMUSCULAR
Qué evaluaciones realizar?

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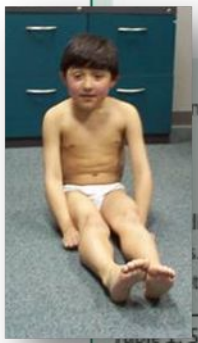
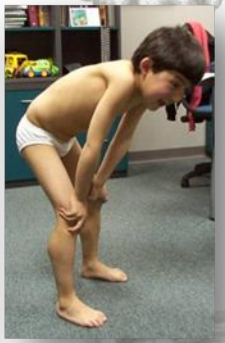
of daily living	Assessment of impairment in daily activities in the home, school, and community settings	Highly relevant to targeted input with aids, adaptations, and access to environmental controls	Frequency of falls, step activity monitoring, self-care skills, writing, computer use	Self-care skills, writing, computer use, control of manual and electric wheelchair

Test cuantitativos





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Routine clinic appointments should be every 6 months, unless otherwise specified. Specialist physical and occupational t
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Table 1: **Suggested neuromuscular assessments for patients with Duchenne muscular dystrophy**

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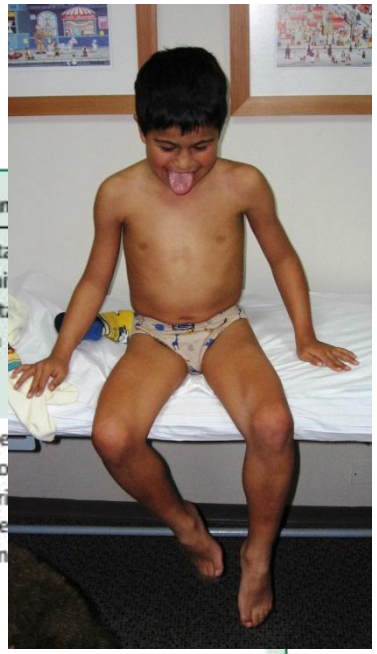
MANEJO NEUROMUSCULAR
Qué evaluaciones realizar?



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of daily living	activities in the home, school, and community settings	aids, adaptations, and access to environmental controls	activity monitoring, self-care skills, writing, computer use Functioning in school and community settings	use, control of manual and electric wheelchair Functioning in school and community settings
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Routine clinic appointments should be every 6 months, unless otherwise specified. Specialist physical and occupational therapy assessments are recommended every 4 months. MRC=UK Medical Research Council. *Although the panel found these tests to be appropriate assessment tools, they are used more typically in research than in clinical settings.

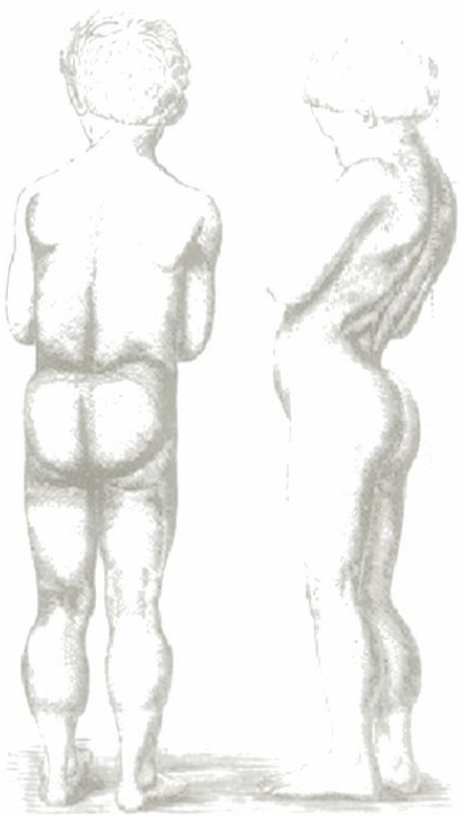
Table 1: **Suggested neuromuscular assessments for patients with Duchenne muscular dystrophy**

Controles neurológicos cada 4- 6 meses.

Evaluaciones kinésicas / T.ocupacionales cada 4 meses.

*Según consenso de expertos: instrumentos de evaluación adecuados, pero habitualmente más usados en investigación que en clínica.

Distrofia Muscular de Duchenne. Presente y Futuro



Evaluaciones necesarias

por etapa de la enfermedad

Seguimiento del paciente con DMD

Neuromuscular

Nutrición

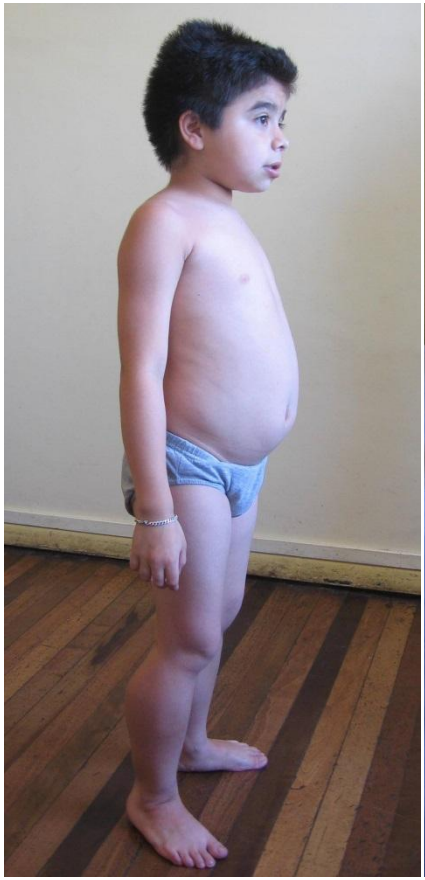
Gastrointestinal

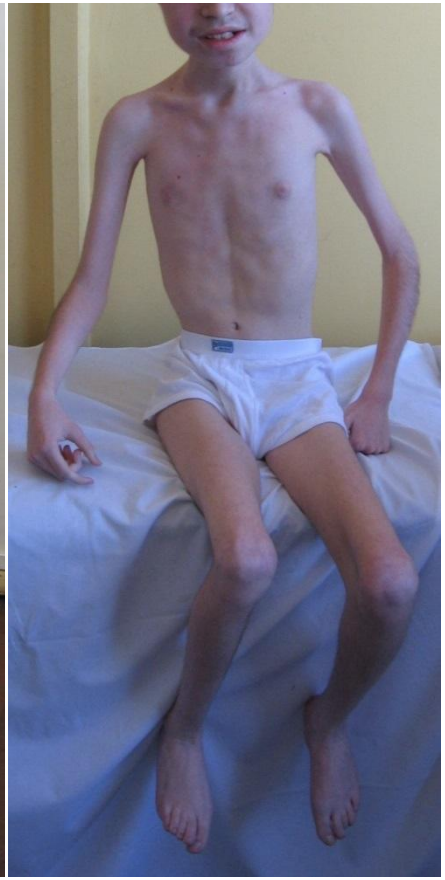
Salud ósea

MANEJO NUTRICIONAL

- IDEA ES:
  mantener un buen estado nutricional, prevenir desnutrición y sobrepeso desde el diagnóstico y por toda la vida.

- **Mantener Peso para la edad o índice de masa corporal para la edad, entre percentil 10 y 85 en curvas nacionales**
- **Dieta balanceada para toda la familia**
- **Control estricto de peso y talla (calculada mediante medición de brazo en no ambulantes).**
- **Manejo nutricional en: sobrepeso, bajo peso, pérdida o aumento de peso involuntaria, si se planea cirugía mayor, estreñimiento crónico y/o si tiene disfagia.**





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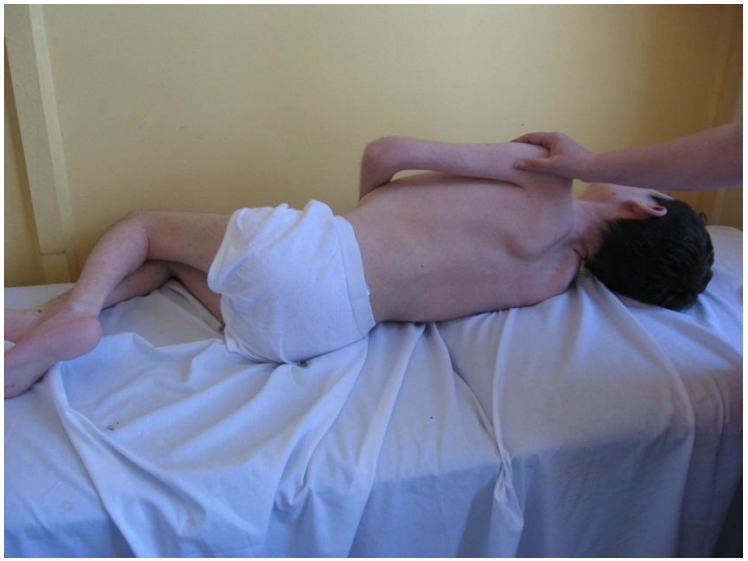
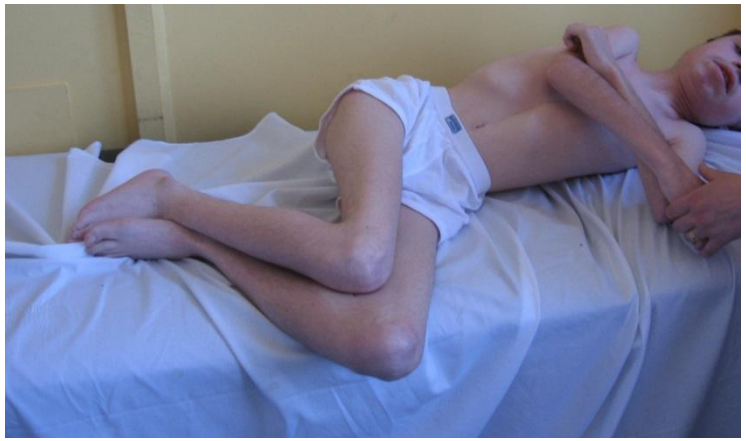
http://www.treat-nmd.eu/diagnosis-and-management-of-DMD

Guglieri M, Bushby K, Recent developments in the management of Duchenne muscular dystrophy, Paediatrics and Child Health (2015), http://dx.doi.org/10.1016/j.paed.2015.07.002

MANEJO NUTRICIONAL

- **Revisión periódica de la Dieta : calorías, proteínas, líquidos, calcio, vitamina D y otros nutrientes.**
  Recomendación: vitamina D y minerales.
  Si hay pérdida de peso, buscar trastorno de deglución.
- **No olvidar posibilidad de complicaciones en otros sistemas, como los sistemas cardíaco o respiratorio, puede contribuir a la pérdida de peso.**
  Si hay pérdida de peso inesperada chequear otras áreas.





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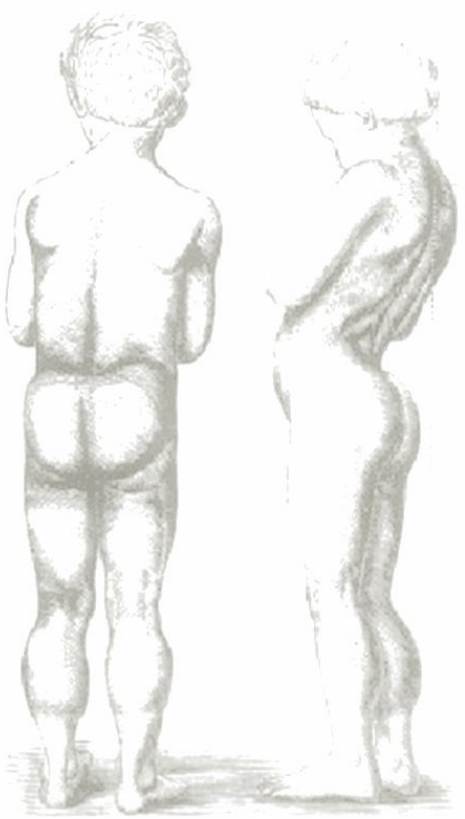
Distrofia Muscular de Duchenne. Presente y Futuro



Evaluaciones necesarias
por etapa de la enfermedad

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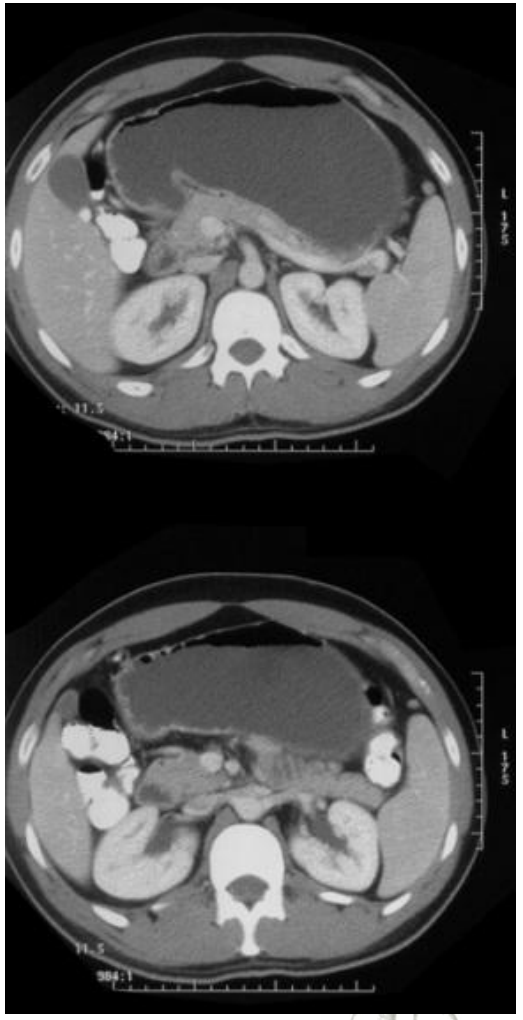
Salud ósea

El estreñimiento y el reflujo gastroesofágico son las dos condiciones gastrointestinales más frecuentes en Duchenne.
Otros: dilatación gástrica aguda y retardo en el vaciamiento gástrico.



IMPORTANTE: dilatación gástrica aguda, suboclusión intestinal.
El estreñimiento ocurre típicamente a una edad mayor y después de la cirugía. Con el aumento de supervivencia, otras complicaciones están siendo reportadas, incluyendo inflamación gástrica e intestinal relacionada con la ingestión de aire debido al uso del ventilador.
•Laxantes y otros medicamentos pueden ser útiles., pero con suficiente ingesta de líquidos.

Aumento de fibra puede empeorar los síntomas, especialmente si los fluidos no se incrementan.

http://www.treat-nmd.eu/diagnosis-and-management-of-DMD

MANEJO GASTROINTESTINAL

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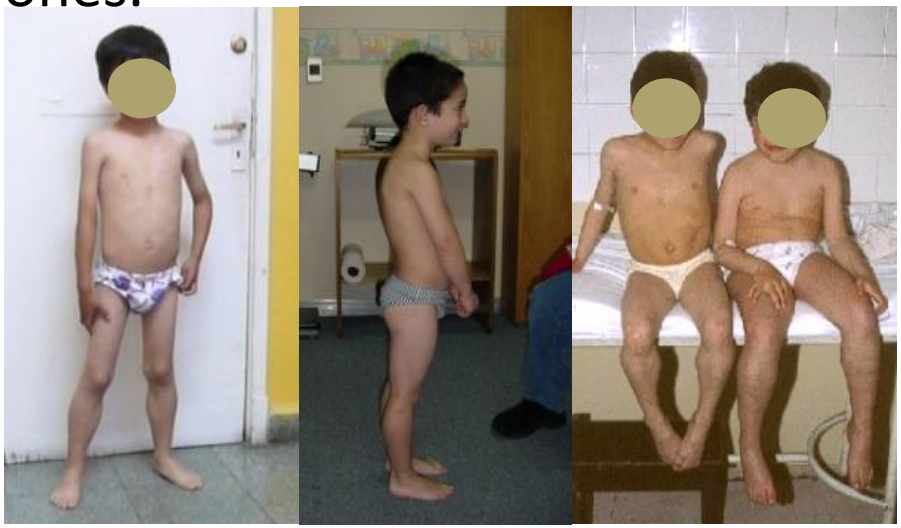
- El reflujo es generalmente tratados con medicamentos apropiados.

Bloqueadores de ácido se recetan comúnmente a los niños en tratamiento con corticoides o bisfosfonatos orales para evitar complicaciones.



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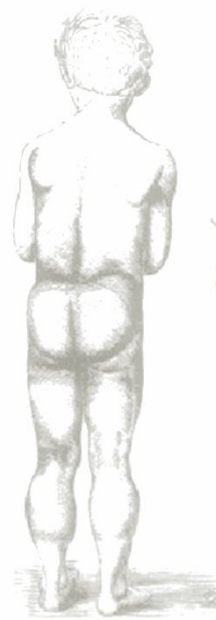

Recomendaciones de Cuidado Bucal

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UNIVERSIDAD DE CHILE

• Control odontológico periódico esencial en Duchenne (DMD) deben ver a un dentista con amplia experiencia y conocimiento detallado de la enfermedad, preferiblemente en una clínica centralizada o especializada. La misión del dentista debe ser procurar un tratamiento, salud oral y bienestar de alta calidad, y funcionar como un recurso para las familias y el propio dentista del niño en su comunidad. Este dentista debe ser consciente de las diferencias
específicas en el desarrollo dental y del esqueleto en los niños con Duchenne, y colaborar con un bien informado y experimentado ortodoncista.
• El cuidado dental y oral se basa en medidas profilácticas con el fin de mantener una buena higiene bucal y dental.
• Dispositivos para asistencia Individual adaptados, y ayudas técnicas para la higiene bucal son de especial importancia cuando la fuerza muscular de las manos, brazos y cuello del niño comienza a disminuir.

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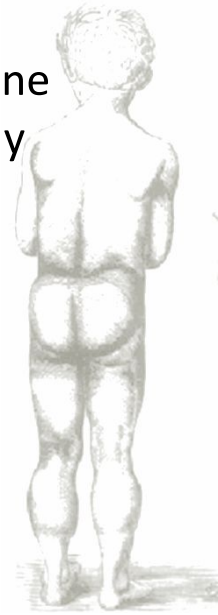
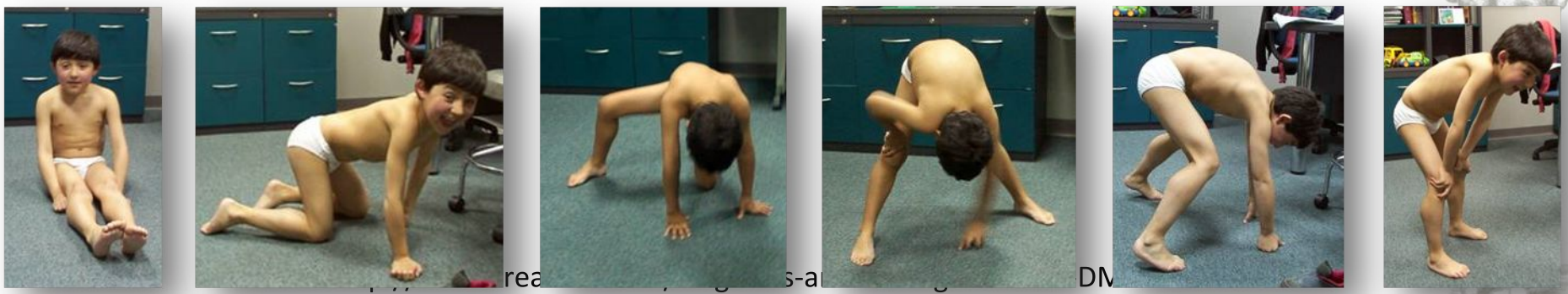


Aumento de fibra puede empeorar los síntomas, especialmente si los fluidos no se incrementan.

• El reflujo es generalmente tratados con medicamentos apropiados.

Bloqueadores de ácido se recetan comúnmente a los niños en tratamiento con corticoides o bisfosfonatos orales para evitar complicaciones.

• El cuidado bucal es un área importante, y aunque esto no fue incluido en la publicación de consenso internacional sobre el cuidado y manejo de Duchenne, TREAT-NMD ha desarrollado recomendaciones de expertos del cuidado bucal.



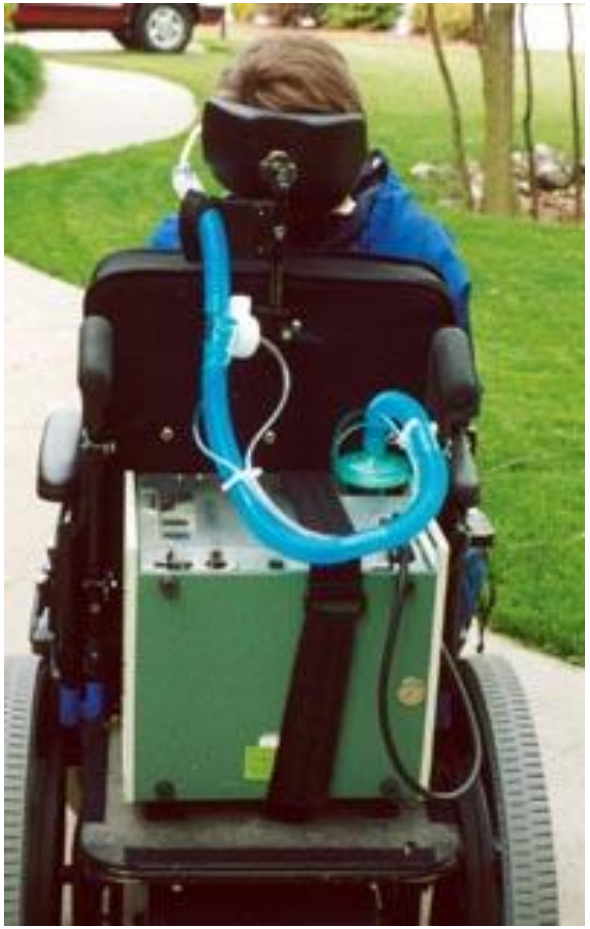
MANEJO DE LA DEGLUCIÓN

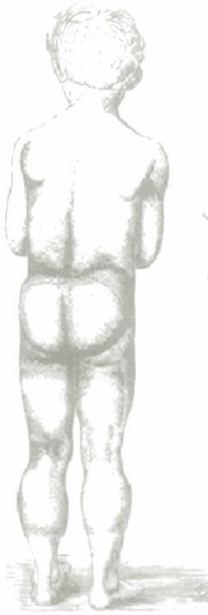
En etapas posteriores, la debilidad de los músculos bulbares puede llevar a disfagia, acentuando aún más los problemas nutricionales.

A menudo, esto puede venir muy gradualmente, lo que significa que puede ser difícil de detectar.

• Pruebas clínicas y de rayos X de la deglución son necesarias cuando existen indicadores clínicos de posible aspiración (entrada de comida en la tráquea) y pobre movimiento de los músculos para tragar (sentir la comida se atora en la garganta).

Estos indicadores incluyen la pérdida de peso involuntaria, de 10% o más, o aumento de peso insuficiente en niños en crecimiento, tiempos prolongados de comida (> 30 minutos) u hora de comida acompañada de fatiga, tos o atragantamiento.





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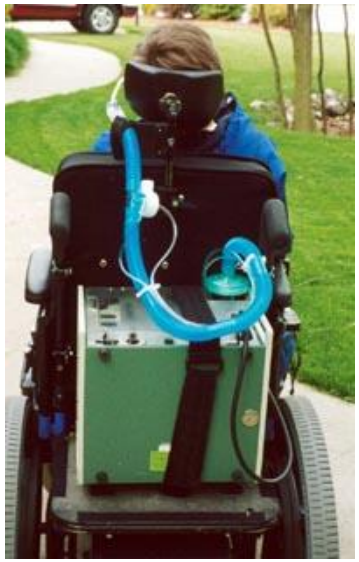
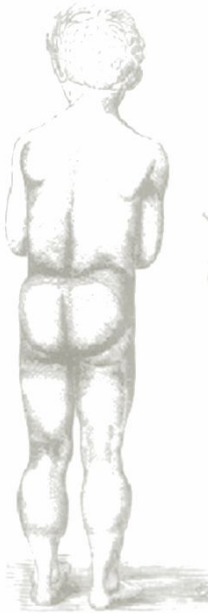
MANEJO DE LA DEGLUCIÓN



- Neumonía causada por líquido que va a los pulmones (neumonía por aspiración), disminución inexplicable de la función pulmonar, o fiebre de origen desconocido pueden ser signos de problemas en la deglución que requiere evaluación.
- En caso de problemas para deglutir, un Terapeuta del Habla y Lenguaje (SLT) debe participar para entregar un plan de tratamiento individualizado. El objetivo es preservar buena función de deglución.
- Colocación del tubo gástrico se debe ofrecer cuando los esfuerzos para mantener el peso y la ingesta de líquidos por vía oral no ayuda lo suficiente. Entre los posibles riesgos y beneficios del procedimiento se deben discutir cuidadosamente. Una gastrostomía puede ser colocada por vía endoscópica o abierta, teniendo en Datos Importantes para

http://mda.org/disease/duchenne-muscular-dystrophy/medical-management#spinal_curvatures



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Recomendaciones de seguimiento

Recordar:

1. Control de talla y peso deben ser controlados en cada visita

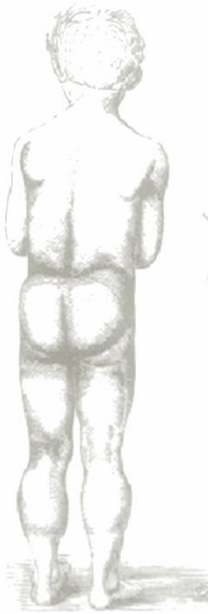
2. Dieta bien balanceada, que incluya la cantidad adecuada de calcio y vitamina D.

3. control nutricional: ayudarle a comer mejor.

4. Evaluación urgente si tiene signos de problemas de deglución.

5. gastrostomía es otra opción después de probar otras maneras de mantener el peso.

Una sonda de alimentación proporcionada en el momento adecuado puede aliviar mucha presión de tratar de comer lo suficiente. Siempre que los músculos de la deglución están bien, tener un tubo de alimentación no significa que no pueda comer los alimentos que desea - sólo que no tiene que depender de las comidas para obtener las calorías y otros nutrientes por lo que puede disfrutar más de la comida.



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Recomendaciones de Cuidado Bucal

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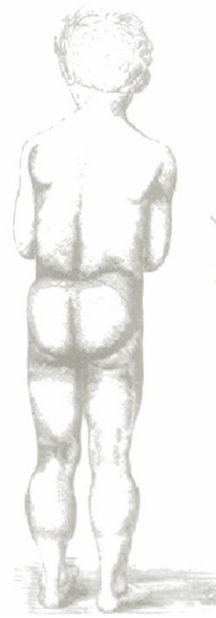

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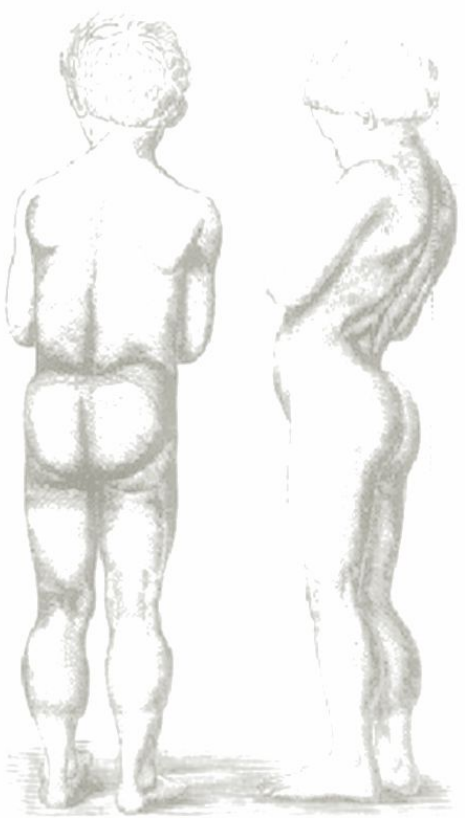
Distrofia Muscular de Duchenne. Presente y Futuro



Evaluaciones necesarias

por etapa de la enfermedad

Seguimiento del paciente con DMD

Neuromuscular

Nutrición

Gastrointestinal

Salud ósea

	Stage 1: Presymptomatic	Stage 2: Early ambulatory	Stage 3: Late ambulatory	Stage 4: Early non-ambulatory	Stage 5: Late non-ambulatory
Diagnostics	Can be diagnosed at this stage if creatine kinase found to be raised or if positive family history Might show developmental delay but no gait disturbance Diagnostic examination and genetic counselling	Gowers' sign Waddling gait Might be toe walking Can climb stairs	Increasingly laboured gait Losing ability to climb stairs and rise from floor Likely to be diagnosed by this stage unless delayed for other reasons (eg, concomitant pathology)	Might be able to self propel for some time Able to maintain posture Might develop scoliosis	Upper limb function and postural maintenance is increasingly limited
Neuromuscular management	Anticipatory planning for future developments Ensure immunisation schedule is complete	Continue assessment to ensure course of disease is as expected in conjunction with interpretation of diagnostic testing At least 6-monthly assessment of function, strength, and range of movement to define phase of disease and determine need for intervention with GCs, ongoing management of GC regimen, and side-effect management			
Orthopaedic management	Orthopaedic surgery rarely necessary		Consider surgical options for TA contractures in certain situations	Monitor for scoliosis: intervention with posterior spinal fusion in defined situations Possible intervention for foot position for wheelchair positioning	
Rehabilitation management	Education and support Preventive measures to maintain muscle extensibility/minimise contracture Encouragement of appropriate exercise/activity Support for function and participation Provision of adaptive devices, as appropriate		Continue previous measures Provision of appropriate wheelchair and seating, and aids and adaptations to allow maximum independence in ADL, function, and participation		
Pulmonary management	Normal respiratory function Ensure usual immunisation schedule includes 23-valent pneumococcal and influenza vaccines	Low risk of respiratory problems Monitor progress		Increasing risk of respiratory impairment Trigger respiratory assessments	High risk of respiratory impairment Trigger respiratory investigations and interventions
Cardiac management	Echocardiagram at diagnosis or by age 6 years	Maximum 24 months between investigations until age 10 years, annually thereafter	Assessment same as in the younger group Increasing risk of cardiac problems with age; requires intervention even if asymptomatic Use of standard heart failure interventions with deterioration of function		
GI, speech/ swallowing, nutrition management	Monitor for normal weight gain for age Nutritional assessment for over/underweight				Attention to possible dysphagia
Psychosocial management	Family support, early assessment/intervention for development, learning, and behaviour	Assessment/intervention for learning, behaviour, and coping Promote independence and social development			Transition planning to adult services

Figure 2: **Stages of disease and care considerations**
ADL=activities of daily living. GCs=glucocorticoids. GI=gastrointestinal. TA=tendo-Achilles.

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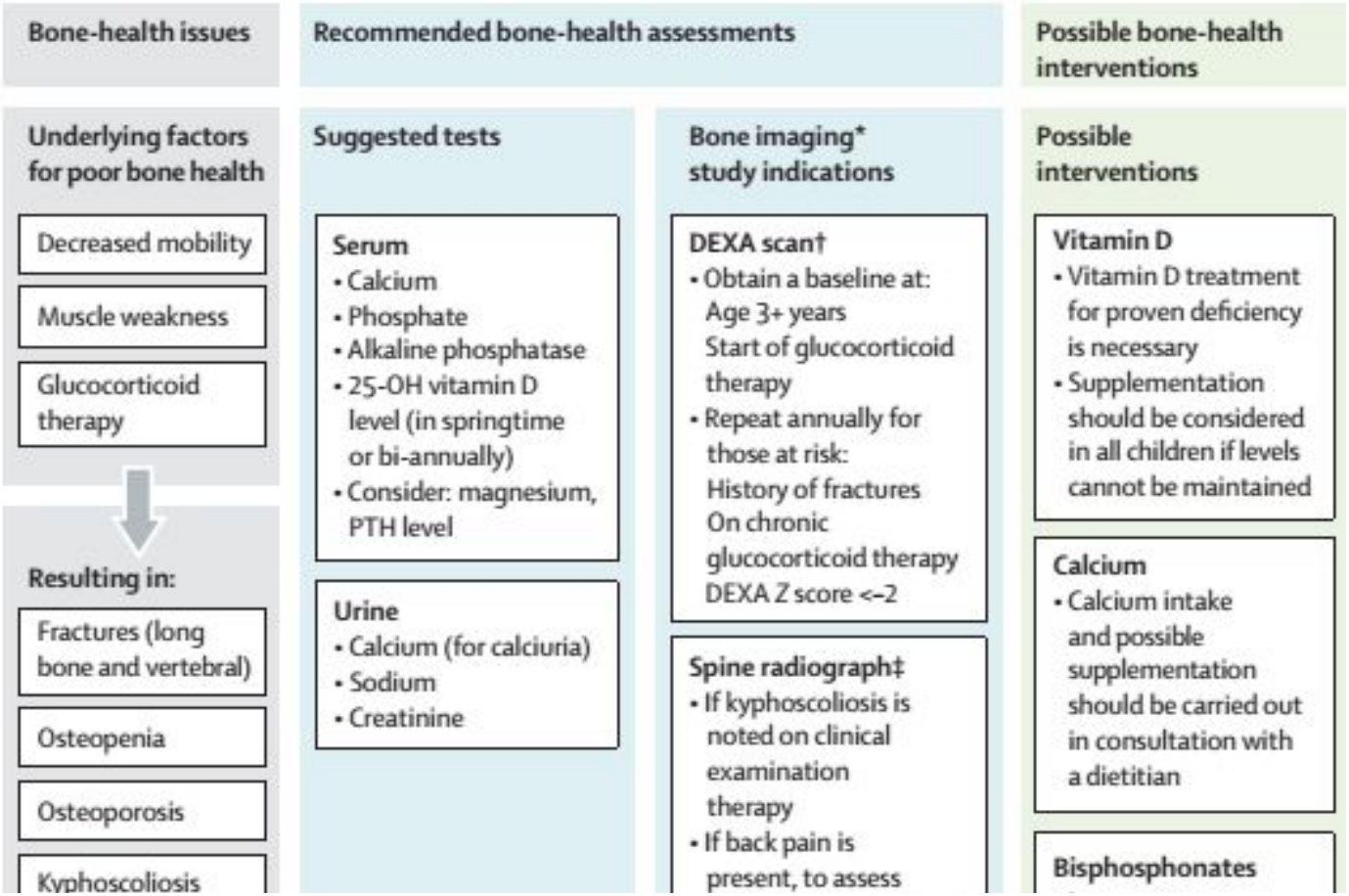


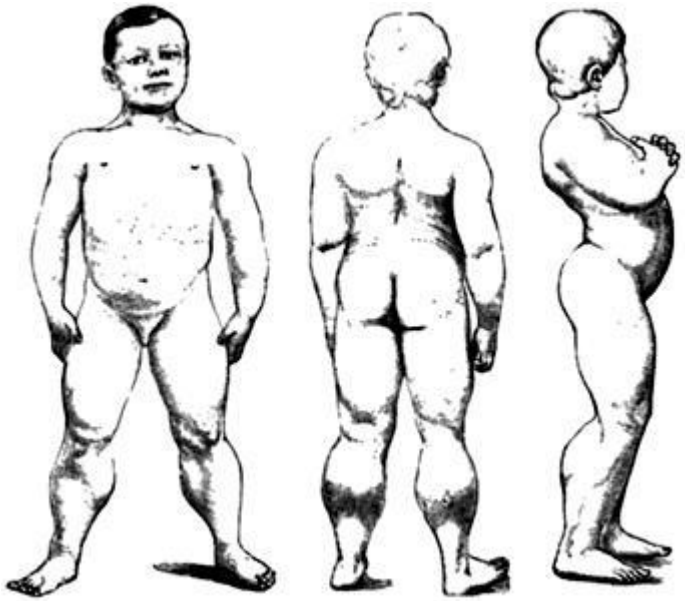
*All imaging assessments should be done at a facility capable of performing and interpreting age-appropriate studies. †A DEXA scan is a better measure than plain fi lm radiographs for detection of osteopenia or osteoporosis. DEXA scans, to assess bone mineral content or body composition, need to be interpreted as a *Z* score for children and a *T* score for adults (compared with age-matched and sex-matched controls).

‡Spine radiographs (posterior/anterior and lateral views) are used for the assessment of scoliosis, bone pain, and compression fractures. It is preferable to obtain them in the standing position, especially if bone pain is the presenting symptom. Useful information can still be obtained in the sitting position for the non-weight-bearing patient. §Bone-age measurements should be done in patients with growth failure (height for age <5% percentile or if linear growth is faltering). If abnormal (>2 SD below the mean), a referral needs to be made to a paediatric endocrinologist.

Lancet Neurol 2010; 9: 77–93

ASPECTOS IMPORTANTES DEL MANEJO

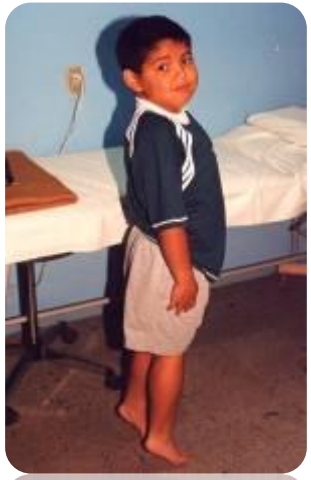
Manejo de complicaciones óseas/esqueléticas.



Bone-health issues	Recommended bone-health assessments		Possible bone-health interventions
Underlying factors for poor bone health Decreased mobility Muscle weakness Glucocorticoid therapy	**Suggested tests** **Serum** • Calcium • Phosphate • Alkaline phosphatase • 25-OH vitamin D level (in springtime or bi-annually) • Consider: magnesium, PTH level **Urine** • Calcium (for calciuria) • Sodium • Creatinine	**Bone imaging* study indications** **DEXA scan†** • Obtain a baseline at: Age 3+ years Start of glucocorticoid therapy • Repeat annually for those at risk: History of fractures On chronic glucocorticoid therapy DEXA *Z* score <–2 **Spine radiograph‡** • If kyphoscoliosis is noted on clinical examination therapy • If back pain is present, to assess vertebral compression fracture **Bone age (left wrist) radiography§** To assess growth failure (on or off glucocorticoid therapy)	**Possible interventions** **Vitamin D** • Vitamin D treatment for proven deficiency is necessary • Supplementation should be considered in all children if levels cannot be maintained **Calcium** • Calcium intake and possible supplementation should be carried out in consultation with a dietitian **Bisphosphonates** • Intravenous bisphosphonates for vertebral fracture are indicated • Oral bisphosphonates as treatment or as a prophylactic measure remain controversial
Resulting in: Fractures (long bone and vertebral) Osteopenia Osteoporosis Kyphoscoliosis Bone pain Reduced quality of life			

Figure 1: **Bone-health management**
Information provided in this figure was not derived from RAND Corporation–University of California Los Angeles Appropriateness Method data and was produced solely using expert discussion. DEXA=dual-energy x-ray absorptiometry. PTH=parathyroid hormone. *All imaging assessments should be done at a facility capable of performing and interpreting age-appropriate studies. †A DEXA scan is a better measure than plain film radiographs for detection of osteopenia or osteoporosis. DEXA scans, to assess bone mineral content or body composition, need to be interpreted as a *Z* score for children and a *T* score for adults (compared with age-matched and sex-matched controls). ‡Spine radiographs (posterior/anterior and lateral views) are used for the assessment of scoliosis, bone pain, and compression fractures. It is preferable to obtain them in the standing position, especially if bone pain is the presenting symptom. Useful information can still be obtained in the sitting position for the non-weight-bearing patient. §Bone-age measurements should be done in patients with growth failure (height for age <5% percentile or if linear growth is faltering). If abnormal (>2 SD below the mean), a referral needs to be made to a paediatric endocrinologist.

Lancet Neurol 2010; 9: 177–89



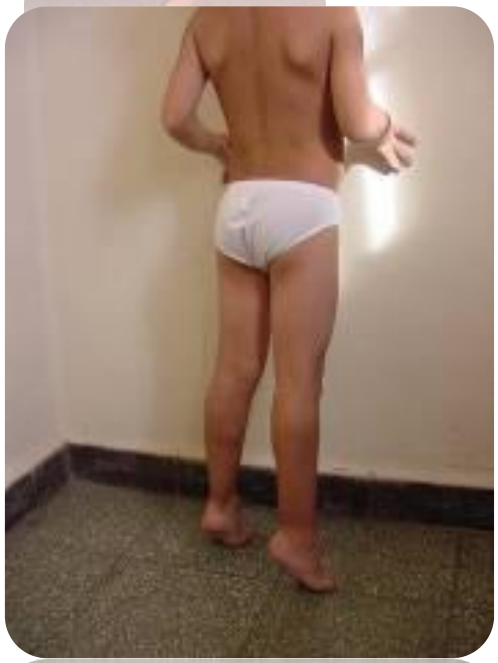
Manejo de la Salud Ósea

Factores subyacentes para pobre salud ósea son:

- Disminución de la movilidad
- Debilidad muscular
- Terapia con corticoides

Intervenciones posibles:

- Vitamina D - necesaria si existe una deficiencia real, suplemento debe ser considerado en los niños.
- Calcio - el consumo es mejor en la dieta, pero la suplementación se debe considerar si la dieta no es adecuada con el asesoramiento de un dietista.
- Bifosfonatos - bifosfonatos IV se recomiendan para fracturas vertebrales.



Importante mantener la bipedestacion.

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EVALUACION DE ASPECTOS PSICOSOCIALES

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Panel 1: **Psychosocial assessments**

Emotional adjustment/coping

- Brief screening of emotional status is strongly recommended at every clinic visit or on an annual basis at a minimum
- Emotional adjustment screening can be informal in nature and does not require a comprehensive assessment
- Use of short standardised rating scales is appropriate and might be helpful
- Could be completed by a social worker or mental health professional or by other clinical staff with sufficient training in this area (eg, attending physician, nurse)

Neurocognitive

- Comprehensive developmental (children ≤4 years old) or neuropsychological (children ≥5 years old) assessment is recommended at or near time of diagnosis and prior to entering formal schooling
- Standardised performance-based tests and parent/patient rating scales should be used
- Should be done by a neuropsychologist or other professional with expertise in brain functioning and development within the context of medical conditions

Speech and language

Assessment for speech and language therapy services is necessary for:

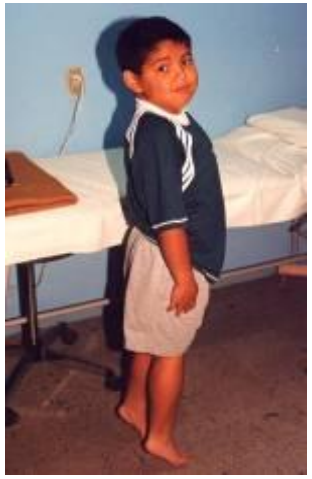
- Younger children who present with suspected delays in speech and/or language development (as identified by caregiver or because of professional concerns)
- Older patients who present with loss or impairment of functional communication ability

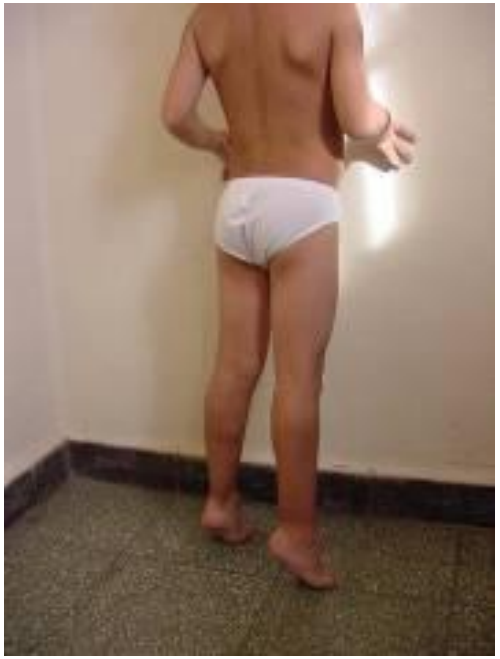
Autism spectrum disorders

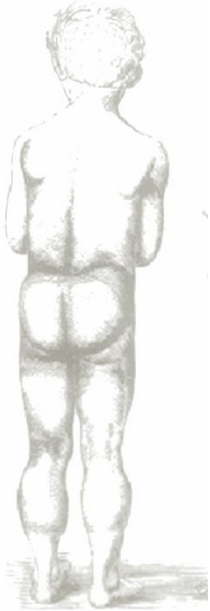
- Screening is necessary in children with DMD who are suspected of having language delays, restricted or repetitive behaviour patterns, or deficits in social functioning (as identified by caregiver or because of professional concerns)
- Necessary to refer to an experienced professional for comprehensive assessment and management of an autism spectrum disorder following positive screening or if ongoing concerns exist

Social work

- Assessment of the caregivers and family by a social-services professional is necessary
- A social services professional is defined as a clinical social worker or other professional who is sufficiently trained and qualified to assess and address emotional adjustment and coping, who has access to financial resources and programmes and social support networks, and who has an understanding/awareness of DMD







Lancet Neurol 2010; 9: 77–93

Otros fármacos y Suplementos Alimenticios

Los expertos concluyeron lo siguiente:

NO RECOMEDABLE: oxandrolona, esteroide anabólico

La seguridad en el uso de Botox no ha sido estudiada para el tratamiento o prevención de contracturas en individuos con Duchenne y no se recomienda.

No existe respaldo para el uso sistemático de creatina.

Un estudio controlado aleatorio de creatina en Duchenne no mostró un beneficio claro. Si el paciente está tomando creatina y tiene evidencia de problemas en riñones, es necesario que suspenda

NO se recomiendan como estandar otros suplementos o fármacos que son utilizados a veces en el tratamiento Duchenne, incluso coenzima Q10, carnitina, aminoácidos (glutamina, arginina), anti-inflamatorios/antioxidantes (aceite de pescado, vitamina E, extracto de té verde, pentoxifilina), y otros incluso extractos herbales o botánicos.

No hay suficiente evidencia en la literatura publicada.

SE requiere investigación adicional y participación activa de familias en actividades que desarrollen mayor conocimiento, como los registros de pacientes y estudios clínicos debe ser ampliamente motivado.

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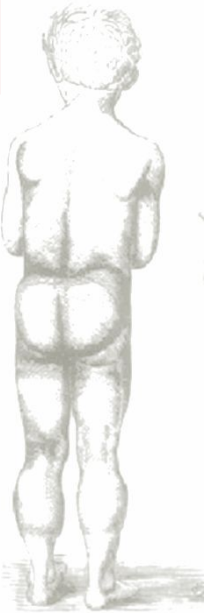
DIFERENTES ESQUEMAS DE TRATAMIENTO CON CORTICOIDES

MANEJO DE CORTICOIDES Y EFECTOS SECUNDARIOS

"Un manejo a conciencia de los efectos secundarios de los corticoides es crucial para una terapia de largo plazo.

Mientras la terapia con corticoides sea la farmacoterapia central en Duchenne, no debe ser tomado a la ligera por doctores y la familia, y debe ser manejado solamente por doctores con experiencia apropiada".

K. Bushby



Bushby K, et al. The Diagnosis and Management of Duchenne Muscular Dystrophy, part 1: diagnosis, and pharmacological and psychosocial management, Lancet Neurology 2010, 9(1) 77-93.

Bushby K, et al. The Diagnosis and Management of Duchenne Muscular Dystrophy, part 2: implementation of multidisciplinary care, Lancet Neurology 2010, 9(2) 177-189.

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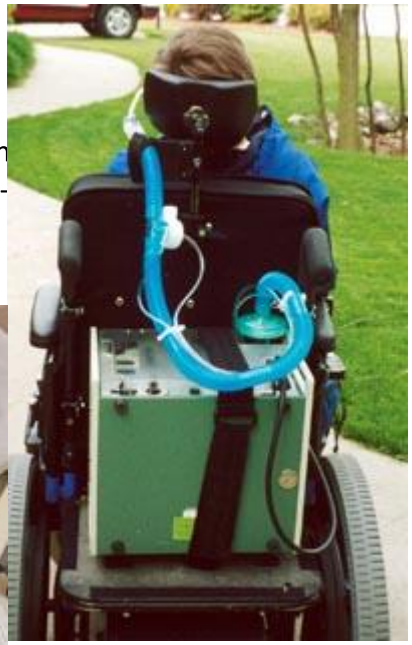
Management of Duchenne muscular dystrophy

Procedure	Frequency	Actions (if required)
Vital signs (HR, BP)	Every 2—4 weeks after initiation of steroids, at least every 6 months thereafter	Referral to a cardiologist for sinus tachycardia, hypertension
Weight and height	3 months after initiation of steroid treatment, at least every 6 months thereafter	Dietary advice Referral to dietician, speech and language therapist Considering steroid treatment modification
Urinalysis (Glycosuria)	Every 2—4 weeks after initiation of steroids, at least every 6 months thereafter	Serum glucose levels Dietary advice Considering steroid treatment modification
Urinalysis (Urine calcium)	Each clinical appointment (if on calcium supplementation)	Modify calcium supplementation
Blood tests (Vitamin D)	Annually	Vitamin D supplementation
Blood tests (Assessment of puberty: Testosterone, LH, FSH)	By the age of 14, if no signs of puberty (if on steroids)	Referral to endocrinologist Treatment (testosterone)
Physical examination	Each clinical appointment (if on steroids)	Gastro-protective treatment (peptic ulcerations) Referral to ophthalmologist (asymptomatic cataracts) Considering steroid treatment modification
Dietary assessment and advice	Each clinical appointment	Referral to dietician, speech and language therapist
Physiotherapy assessment and advice	Every 3—4 months	Stretching exercises Contracture control devices (e.g. AFOs) Wheelchair
Respiratory assessment (FVC)	Each clinical appointment (more frequently if respiratory dysfunction)	Pulse oximetry Referral to respiratory specialist Cough assist machine, non-invasive ventilation
Cardiac assessment (ECG, Echocardiogram)	At diagnosis, once every 2 years until the age of 10 years and annually thereafter (more frequently if cardiac dysfunction)	Referral to a cardiologist Closer follow-up if symptoms/signs of cardiac impairment Treatment (ACE-inhibitors, beta-blockers)

https://sites.google.com/site/cmkoleski18/webtext/poebo070809w_rgbb.jpg

http://mda.org/disease/duchenne-muscular-dystrophy/signs-and-symptoms

http://mda.org/publications/cardiovascular-health-dmd-and-bmd



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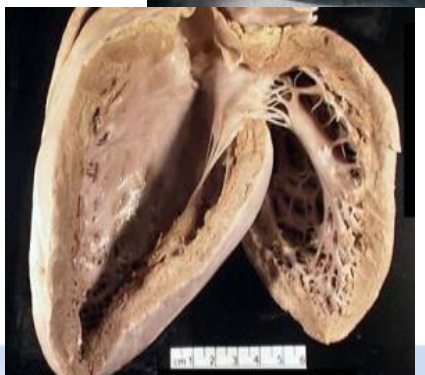

Tratamiento

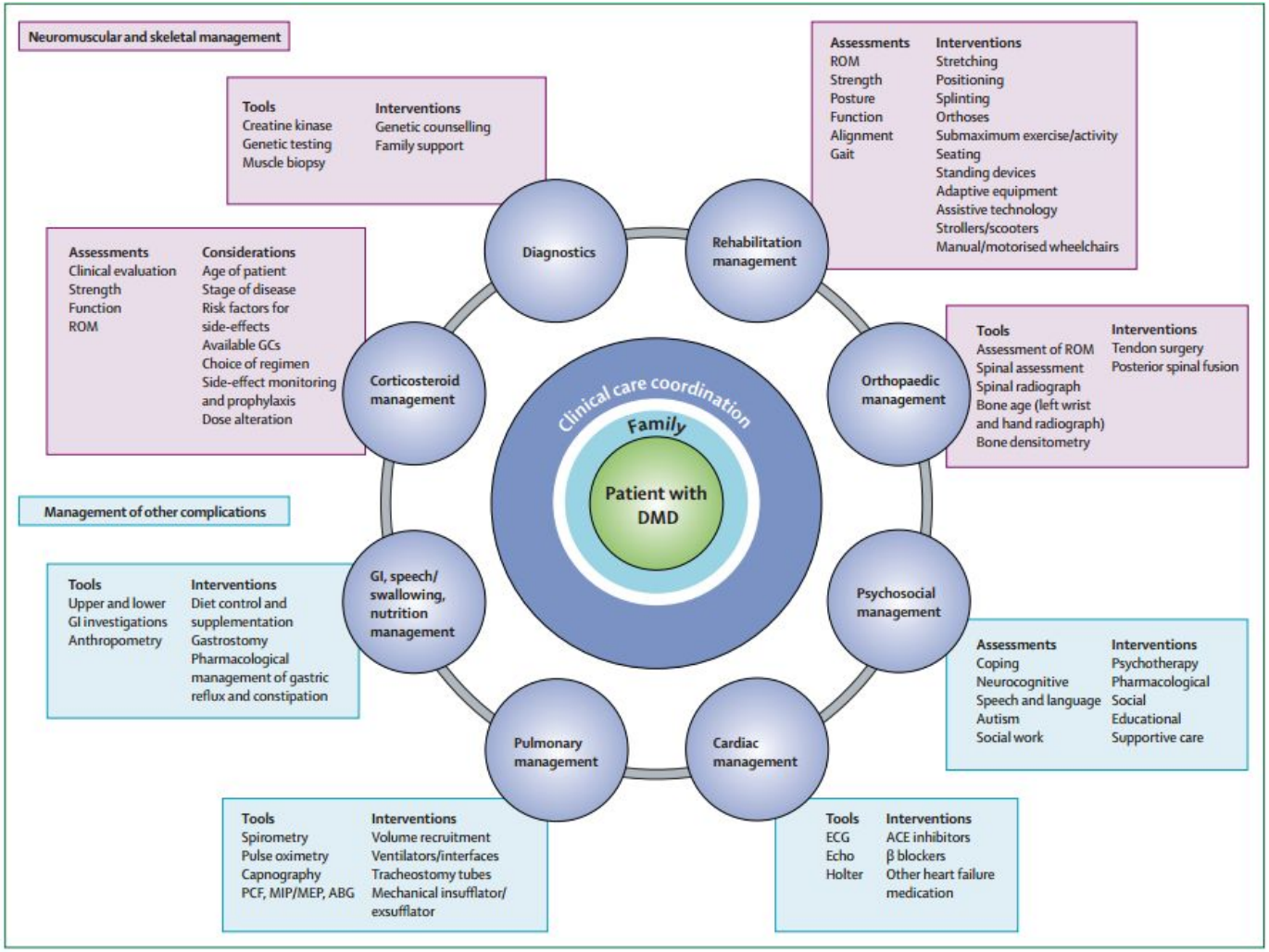

Figure 1: **Interdisciplinary management of DMD**
Coordination of clinical care is a crucial component of the management of DMD. This care is best provided in a multidisciplinary care setting in which the individual and family can access expertise for the required multisystem management of DMD in a collaborative effort. A coordinated clinical care role can be provided by a wide range of health-care professionals depending on local services, including (but not limited to) neurologists or paediatric neurologists, rehabilitation specialists, neurogeneticists, paediatricians, and primary-care physicians. It is crucial that the person responsible for the coordination of clinical care is aware of the available assessments, tools, and interventions to proactively manage all potential issues involving DMD. ABG=arterial blood gas. ACE=angiotensin-converting enzyme. DMD=Duchenne muscular dystrophy. Echo=echocardiogram. ECG=electrocardiogram. GC=glucocorticoids. GI=gastrointestinal. MEP=maximum expiratory pressure. MIP=maximum inspiratory pressure. PCF=peak cough flow. ROM=range of motion.

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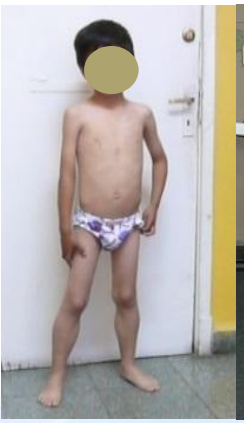
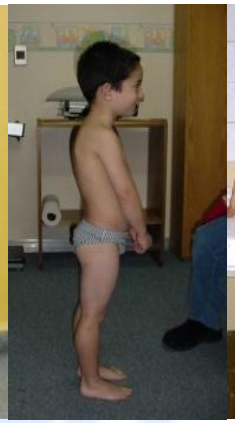
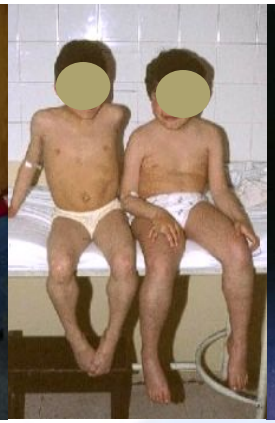
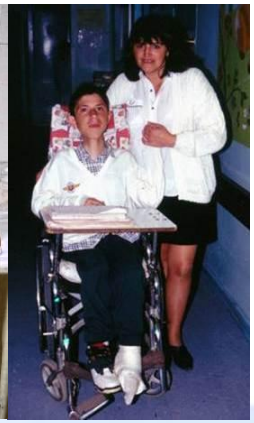
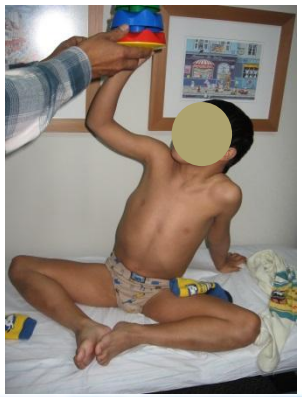
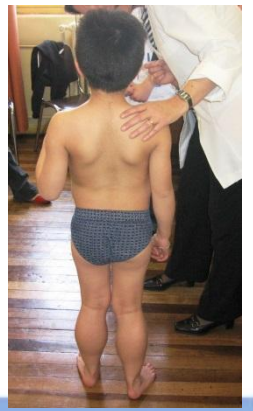
MENSAJES

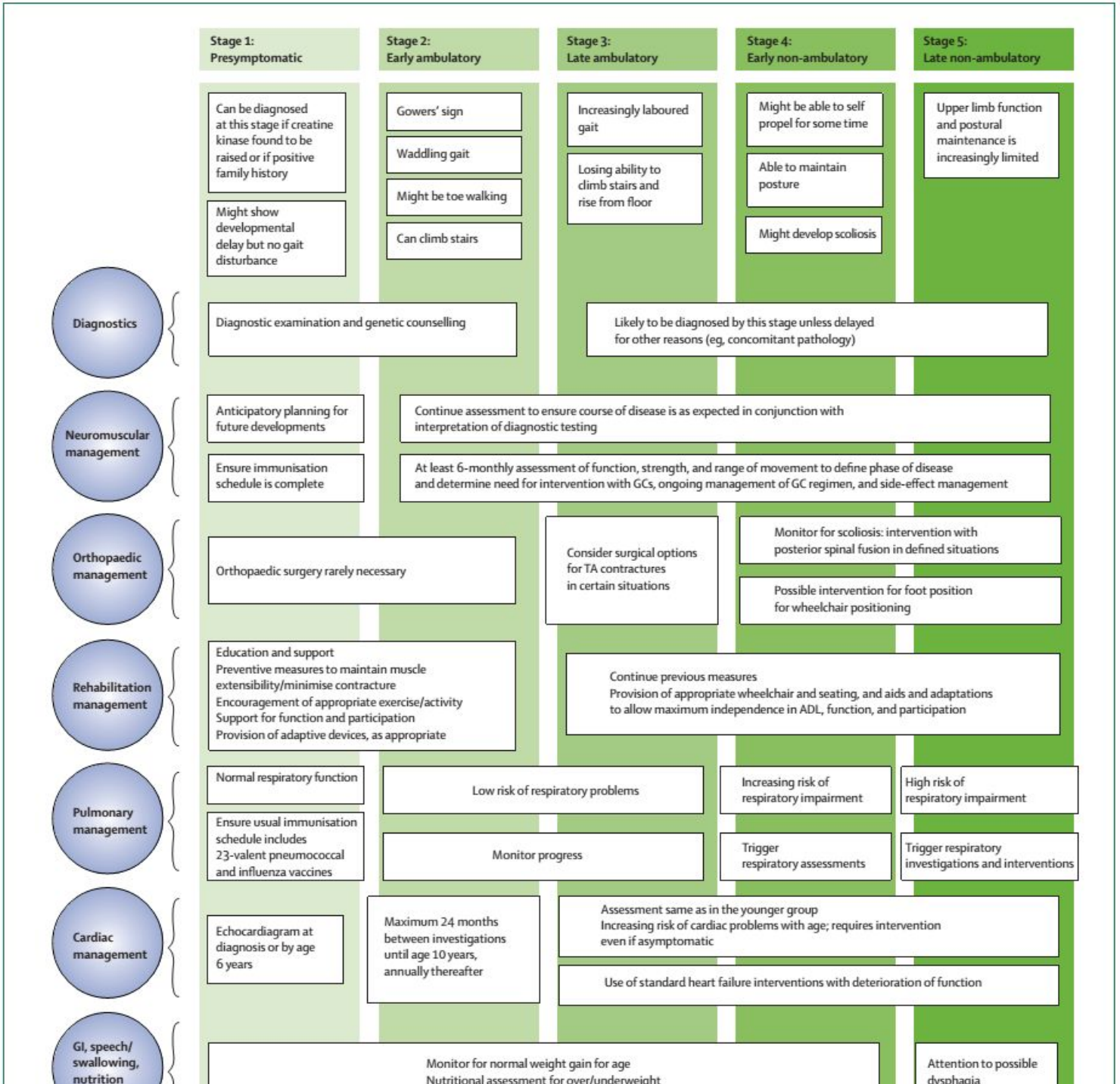
FACULTAD DE MEDICINA
UNIVERSIDAD DE CHILE

- Seguimiento periódico interdisciplinario es fundamental: permite mantener a nuestros pacientes en optimas condiciones a la espera de nuevos tratamientos.

Enfocarse en los objetivos de este proceso: los PACIENTES pensando en el bien superior





Distrofias musculares tratamiento

- Intervención escolar (CI bajo) escolaridad
- peso adecuado: Dieta
- vida activa / adaptaciones mesas de trabajo
- evitar reposo prolongado
- Terapia física: prevenir equino varo del pie y contracturas de flexores de cadera : ejercicios / OTP nocturnas
- manejo farmacológico: Corticoides 2 años más de marcha autónoma
- tenotomías
- instrumentación de columna
- ventilación asistida

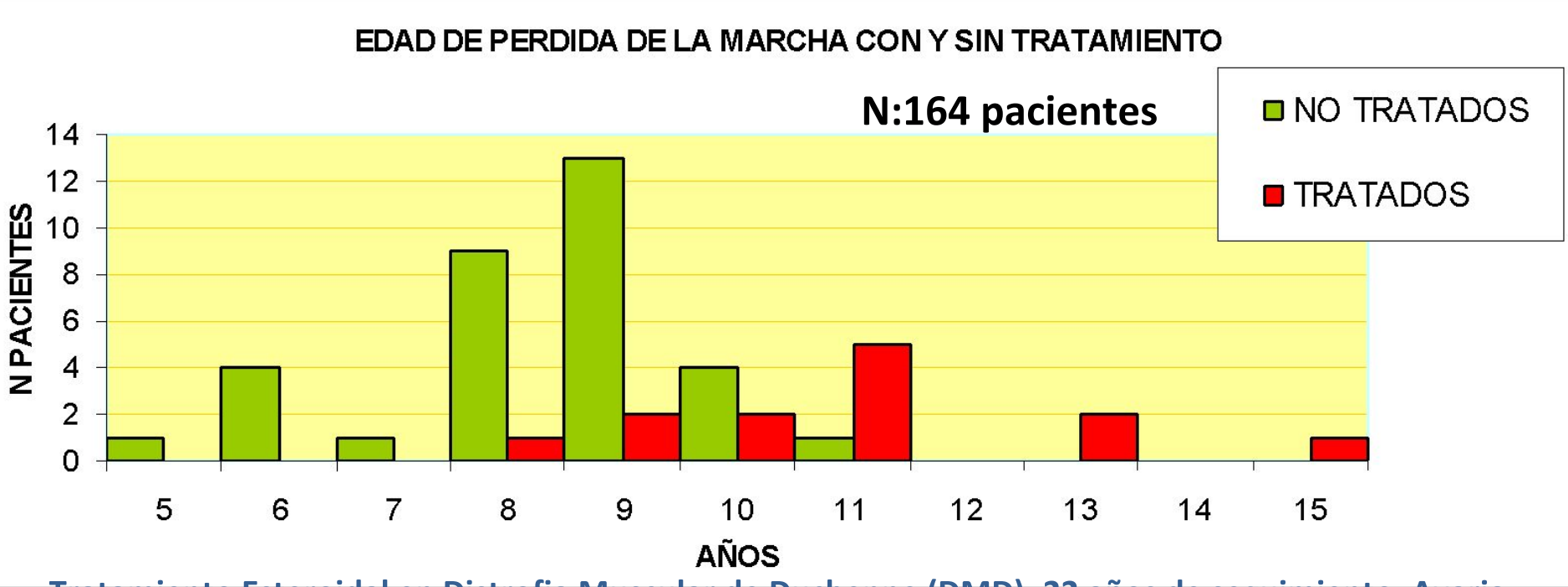
Los corticoides en DMD : un tratamiento de eficacia demostrada
European Journal of Pediatric Neurology 2002

FACULTAD DE MEDICINA

Estudios clínicos en curso (DMD): Muchos…..

Glutamine/Creatine, Coenzyme Q, Pentoxyphillin, Corticosteroides: Weekly treatment; etc
Creatina mejora fuerza muscular en distrofinopatías, FEH y otros. *Neuromuscular DC, UWashington; ENMC*

Remisión de signos clínicos en DMD con tratamiento intermitente de prednisolona bajas dosis. V. Dubowitz European Journal of Pediatric Neurology 2002



Tratamiento Esteroidal en Distrofia Muscular de Duchenne (DMD): 22 años de seguimiento. Avaria, Kleinsteuber, Novoa, et al, 2002

manejo farmacológico: <u>Corticoides</u> 2 años más de marcha autónoma en promedio

La evolución puede ser muy diferente si se deja la enfermedad a su evolución natural o se implementan las medidas de manejo terapéutico validadas actualmente.

PERGAMON

Neuromuscular Disorders 14 (2004) 526–534

www.elsevier.com/locate/nmd

Workshop report

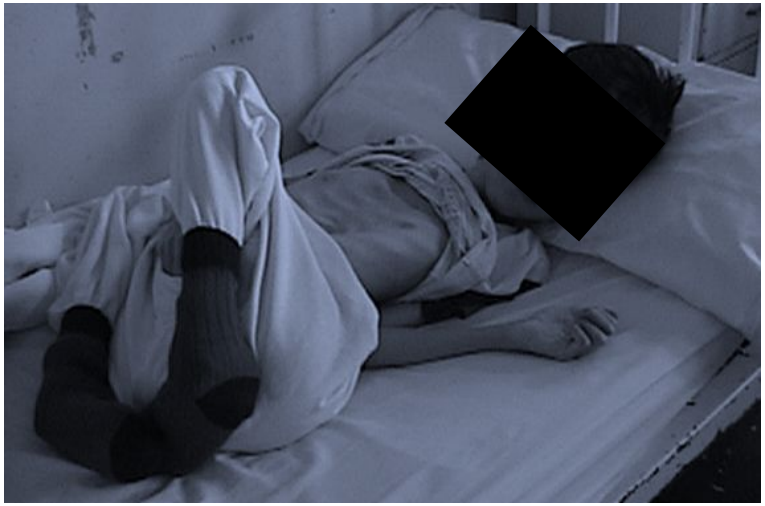
Report on the 124th ENMC International Workshop. Treatment of Duchenne muscular dystrophy; defining the gold standards of management in the use of corticosteroids 2–4 April 2004, Naarden, The Netherlands

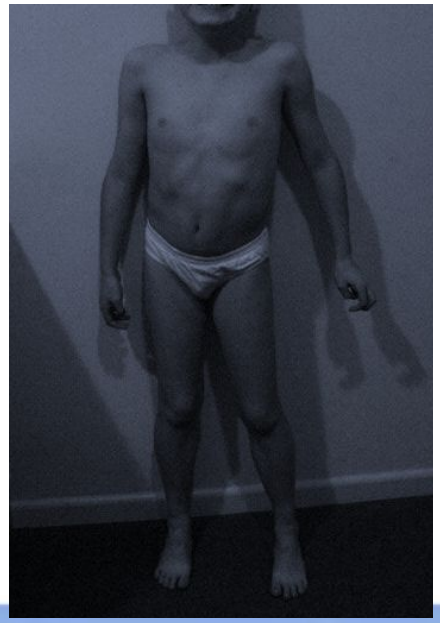
K. Bushby[a,*], F. Muntoni[b], A. Urtizberea[c], R. Hughes[d], R. Griggs[e]

[a]*Institute of Human Genetics, International Centre for Life, Central Parkway, Newcastle upon Tyne NE3 4YQ, UK*
[b]*Hammersmith Hospital, Imperial College, London W12 ONN, UK*
[c]*Institute de Myologie, 75013 Paris, France*
[d]*Department of Clinical Neurosciences, Guys Hospital, London SE1 1UL, UK*
[e]*University of Rochester Medical Centre, New York, NY 14642, USA*

Received 13 May 2004

Ej: 2 pacientes de 15 años con Distrofia de Duchenne 2004





Gentileza Dra. MA Avaria.

Tratamiento

Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and pharmacological and psychosocial management

*Katharine Bushby, Richard Finkel, David J Birnkrant, Laura E Case, Paula R Clemens, Linda Cripe, Ajay Kaul, Kathi Kinnett, Craig McDonald, Shree Pandya, James Poysky, Frederic Shapiro, Jean Tomezsko, Carolyn Constantin, for the DMD Care Considerations Working Group**

Duchenne muscular dystrophy (DMD) is a severe, progressive disease that affects 1 in 3600–6000 live male births. Although guidelines are available for various aspects of DMD, comprehensive clinical care recommendations do not exist. The US Centers for Di... using the RAND Corporati... Considerations Working Gr... gastroenterology and nutri... orthopaedic, and surgical asp... range of practitioners who ca... primary manifestations and ... In part 1 of this Review, we d... on care, pharmacological tre...

Lancet Neurol 2010; 9: 77–93
Published Online
November 30, 2009

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Tratamiento

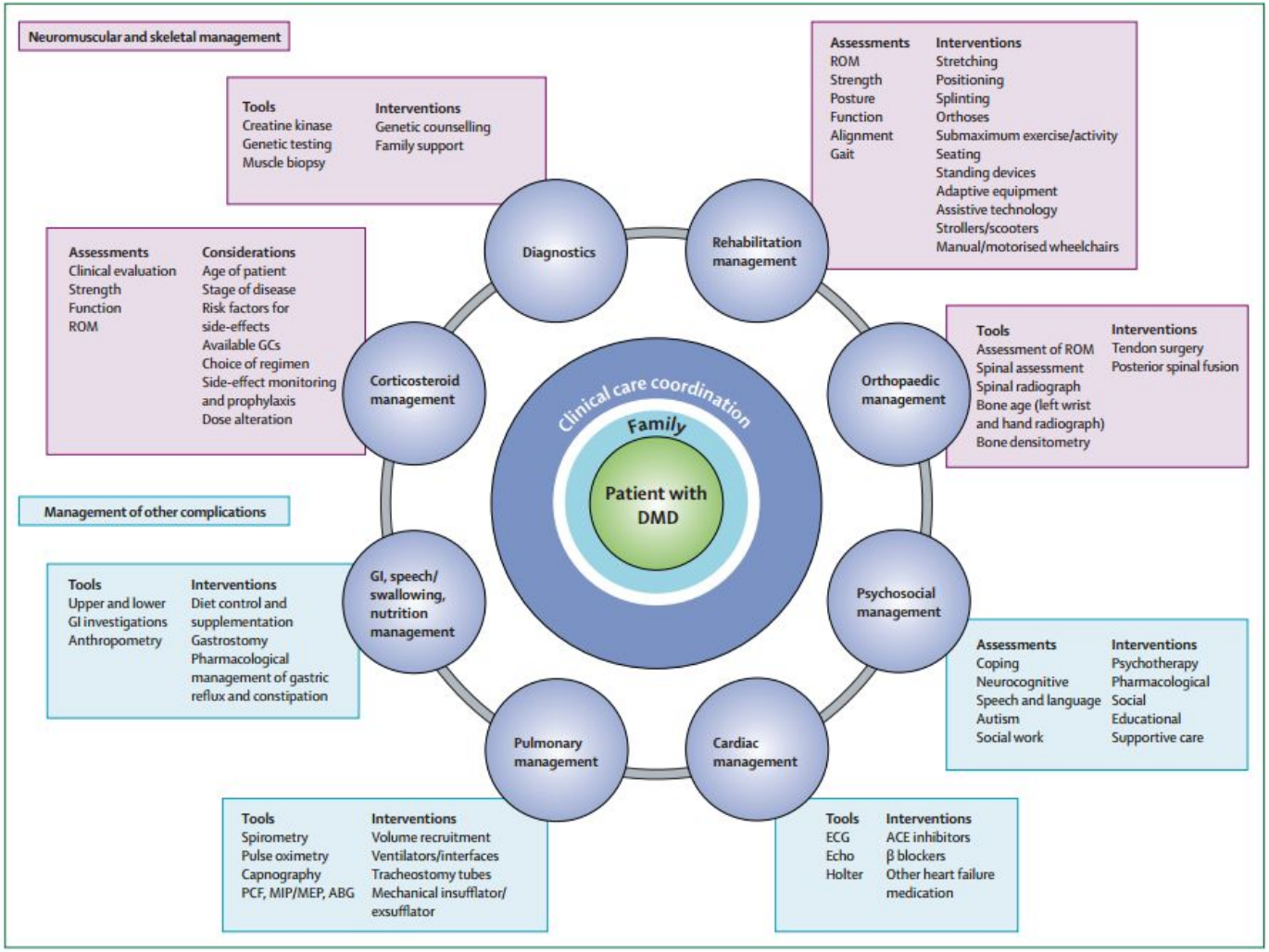

Figure 1: **Interdisciplinary management of DMD**
Coordination of clinical care is a crucial component of the management of DMD. This care is best provided in a multidisciplinary care setting in which the individual and family can access expertise for the required multisystem management of DMD in a collaborative effort. A coordinated clinical care role can be provided by a wide range of health-care professionals depending on local services, including (but not limited to) neurologists or paediatric neurologists, rehabilitation specialists, neurogeneticists, paediatricians, and primary-care physicians. It is crucial that the person responsible for the coordination of clinical care is aware of the available assessments, tools, and interventions to proactively manage all potential issues involving DMD. ABG=arterial blood gas. ACE=angiotensin-converting enzyme. DMD=Duchenne muscular dystrophy. Echo=echocardiogram. ECG=electrocardiogram. GC=glucocorticoids. GI=gastrointestinal. MEP=maximum expiratory pressure. MIP=maximum inspiratory pressure. PCF=peak cough flow. ROM=range of motion.

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Tratamiento

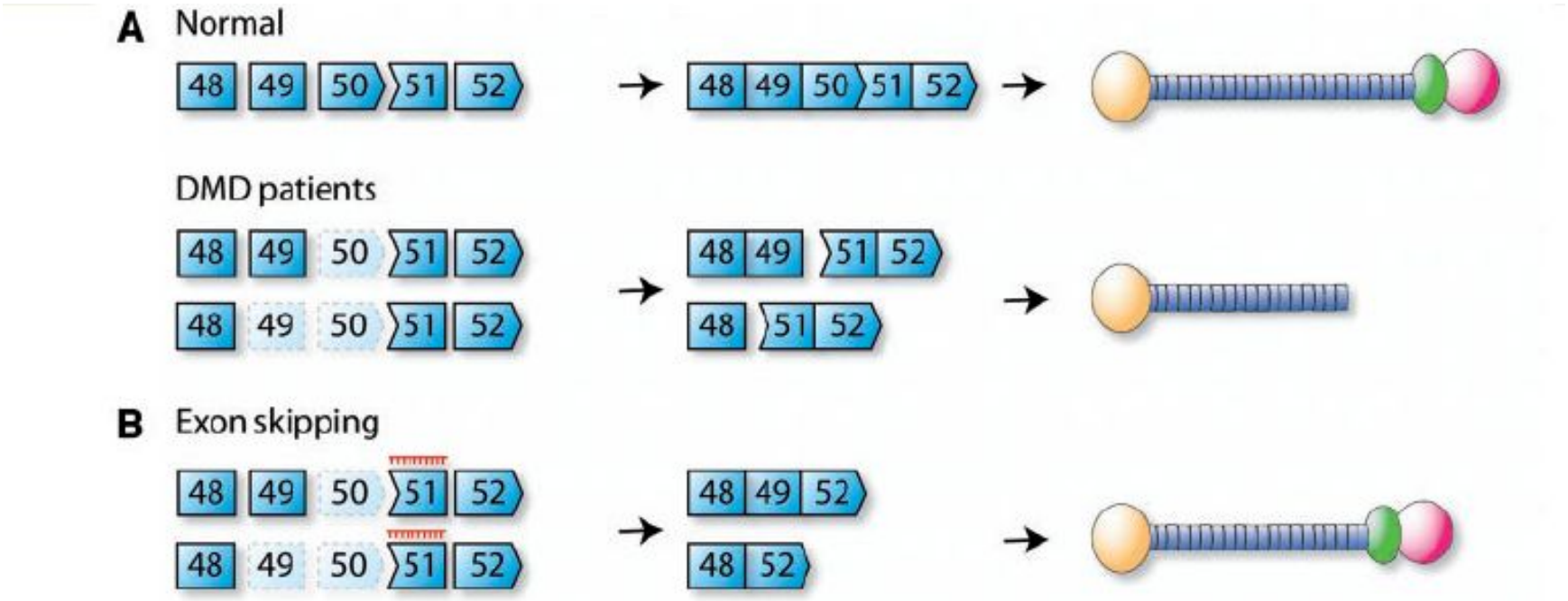
- El tratamiento, común a la mayoría de las distrofias musculares, tiene por objetivo mejorar la calidad de vida, prevenir complicaciones y mantener la deambulación autónoma el mayor tiempo posible.
- Se basa principalmente en mantener vida activa, enfatizando posturas adecuadas y prevención de contracturas, y evitar sobrepeso y reposo prolongado. Es importante mantener escolaridad, con las adaptaciones curriculares necesarias. La *terapia física* se dirige a preservar la fuerza muscular, y prevenir contracturas, capacitando a los padres para la realización de ejercicios de elongación diariamente e indicando uso nocturno de *órtesis tibio-peroneas* para prevenir el equinovaro, y *órtesis isquio-pie* para mantener la marcha según requerimiento. Entrenamiento en fuerza o programas de ejercicio aeróbico pueden mejorar la función muscular y cardiorespiratoria y evitar la atrofia por desuso, sin embargo el ejercicio excesivo puede causar progresión más rápida de la enfermedad.

Tratamiento

1. El tratamiento quirúrgico incluye tenotomías para corrección de contracturas (debe ser evaluado cuidadosamente por riesgo de deterioro asociado a reposo y posibilidad de hipertermia maligna) y corrección quirúrgica de escoliosis. La ventilación asistida no invasiva nocturna o permanente en pacientes con hipoventilación sintomática mejora la calidad de vida y aumenta la supervivencia.
2. Los corticoesteroides son hasta el momento el único tratamiento que modifica la evolución natural de la enfermedad aumentando el tiempo de marcha autónoma dos años en promedio y con efecto positivo en función pulmonar y cardiaca. Los corticoesteroides más utilizados son la prednisona y el deflazacort. No existe consenso sobre la edad de inicio y o de suspensión del tratamiento o el esquema a emplear.
3. La Academia Americana de Neurología, basada en estudios controlados randomizados, recomienda 0,75 mg/kg/día de prednisona o 0,9 mg/kg/ día de deflazacort, similares en su efectividad.
4. El rol de la suplementación de calcio y vitamina D es aún controversial. Se recomienda adecuar la dieta y exposición al sol para mejor aporte de Calcio y vitamina D y suplementar esta última si la concentración sérica cae bajo 20 ng/lt. No hay evidencia que apoye el uso profiláctico de bifosfonatos, pero si están indicados en presencia de fractura vertebral.

Tratamientos en Investigación

Actualmente existen protocolos de investigación que apuntan a la modificación de la expresión génica pre-ARNm, en los transcritos anormales producto de la mutación en el gen *DMD*. Las dos estrategias representativas de este enfoque son la "omisión o salto del exón" (*exon skipping*) y la "omisión o translectura del codón de parada (*stop codon read through*) o la modulación farmacológica de otros genes que influyen en el fenotipo distrófico como la utrofina o la miostatina.



EXON SKIPPING:

DMD: es causada por alteraciones en un gen: el gen de una proteína muy importante para el músculo: la distrofina. Deleciones de uno o más exones (partes de este gen)

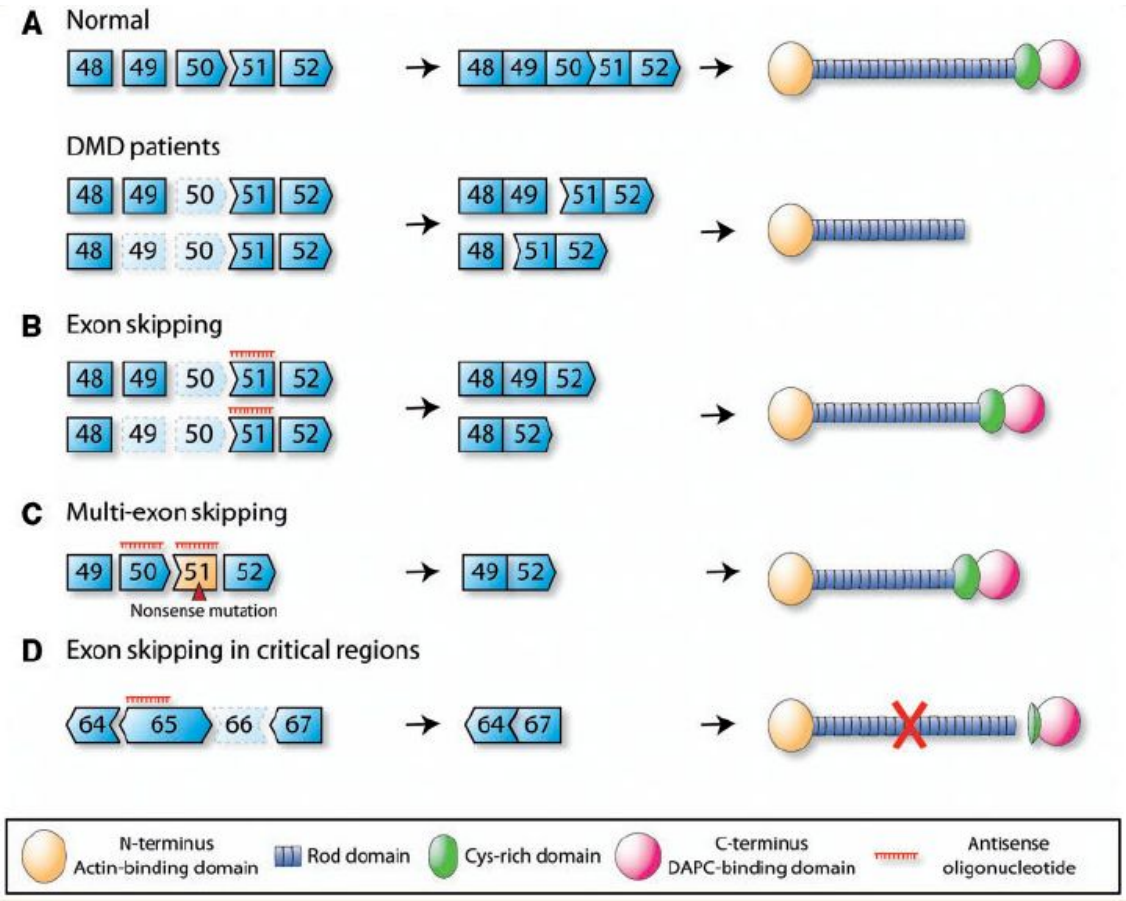
Cuando falta un exón ej: Exón 50 se produce una alteración en el empalme de los exones que quedan.

Se produce lo que se llama un codón de terminación prematura, no es posible leer la información genética necesaria para producir la proteína distrofina y no se produce síntesis de esta proteína

Si una molécula externa: oligodeoxynucleotide antisentido (ODN) dirigida a exón 51 se une a él como un "parche" se permite que el mecanismo de empalme pueda saltar sobre esta parte , pudiendo unirse los exones 49 y 52.

Esto restaura el marco de lectura correcta de modo que sólo una versión ligeramente alterada de la distrofina se produce, es decir, una distrofina BMD-tipo.

27 diciembre de 2007 en el New England Journal of Medicine.



Neuromuscular Disorders 20 (2010) 355–362

Contents lists available at ScienceDirect

Neuromuscular Disorders

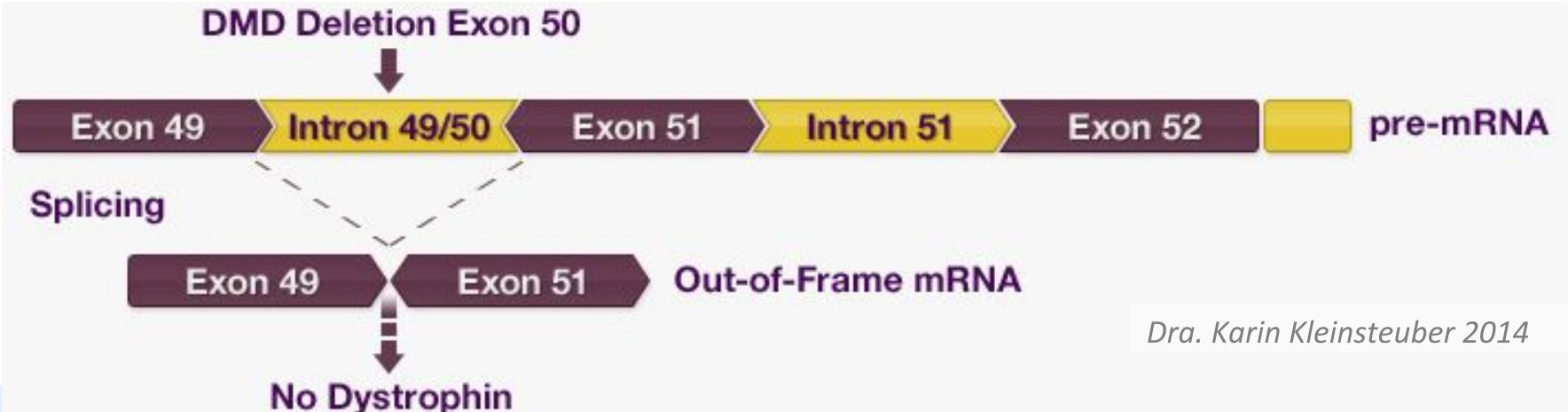
journal homepage: www.elsevier.com/locate/nmd

Workshop report

The development of antisense oligonucleotide therapies for Duchenne muscular dystrophy: Report on a TREAT-NMD workshop hosted by the European Medicines Agency (EMA), on September 25th 2009

F. Muntoni *, on behalf of the meeting steering committee, and of the TREAT-NMD Network

The Dubowitz Neuromuscular Centre, University College London, Institute of Child Health, 30 Guildford Street, London, WC1N 1EH, UK



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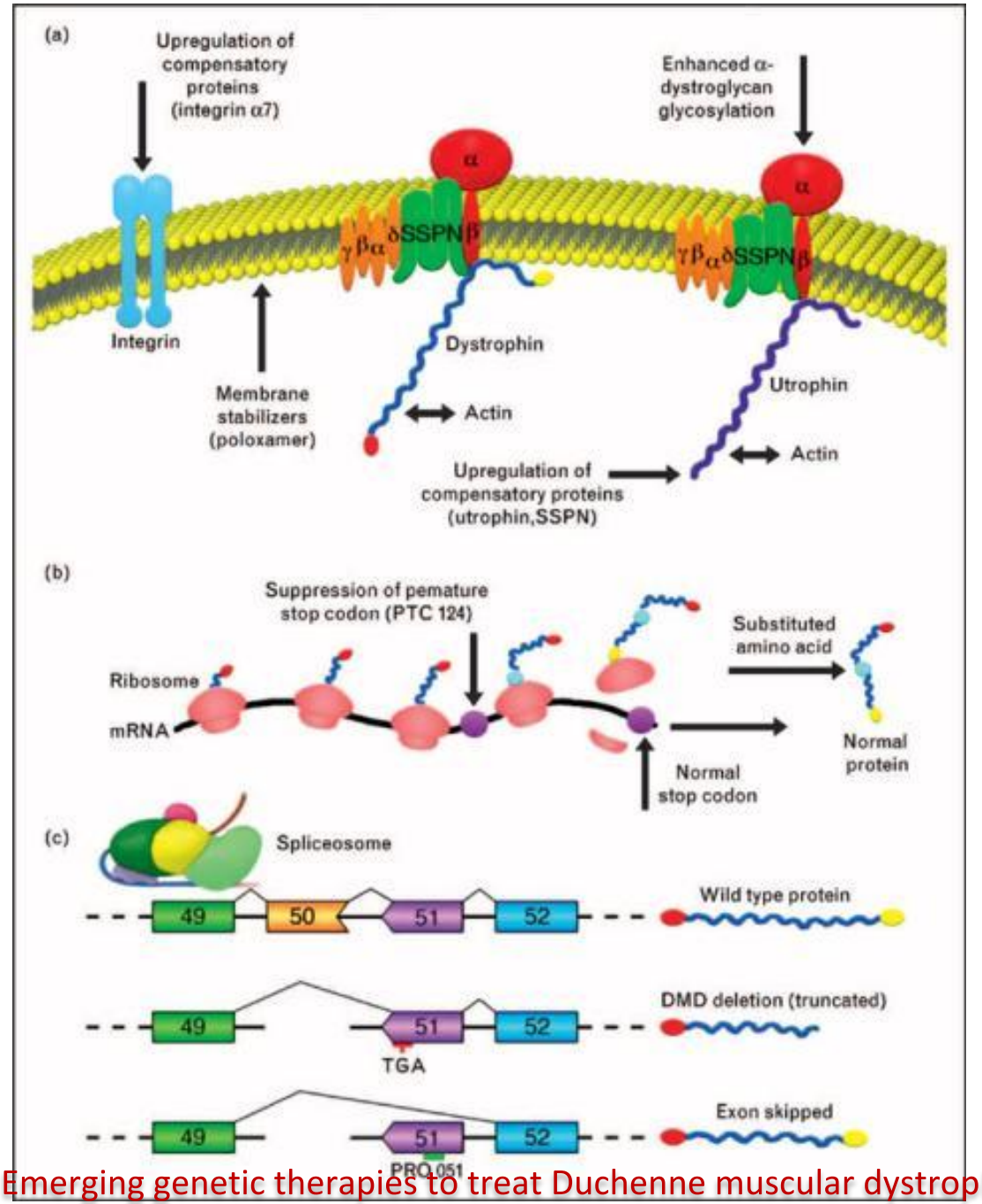

Nelson SF, et als. Emerging genetic therapies to treat Duchenne muscular dystrophy. Curr Opin Neurol. 2009

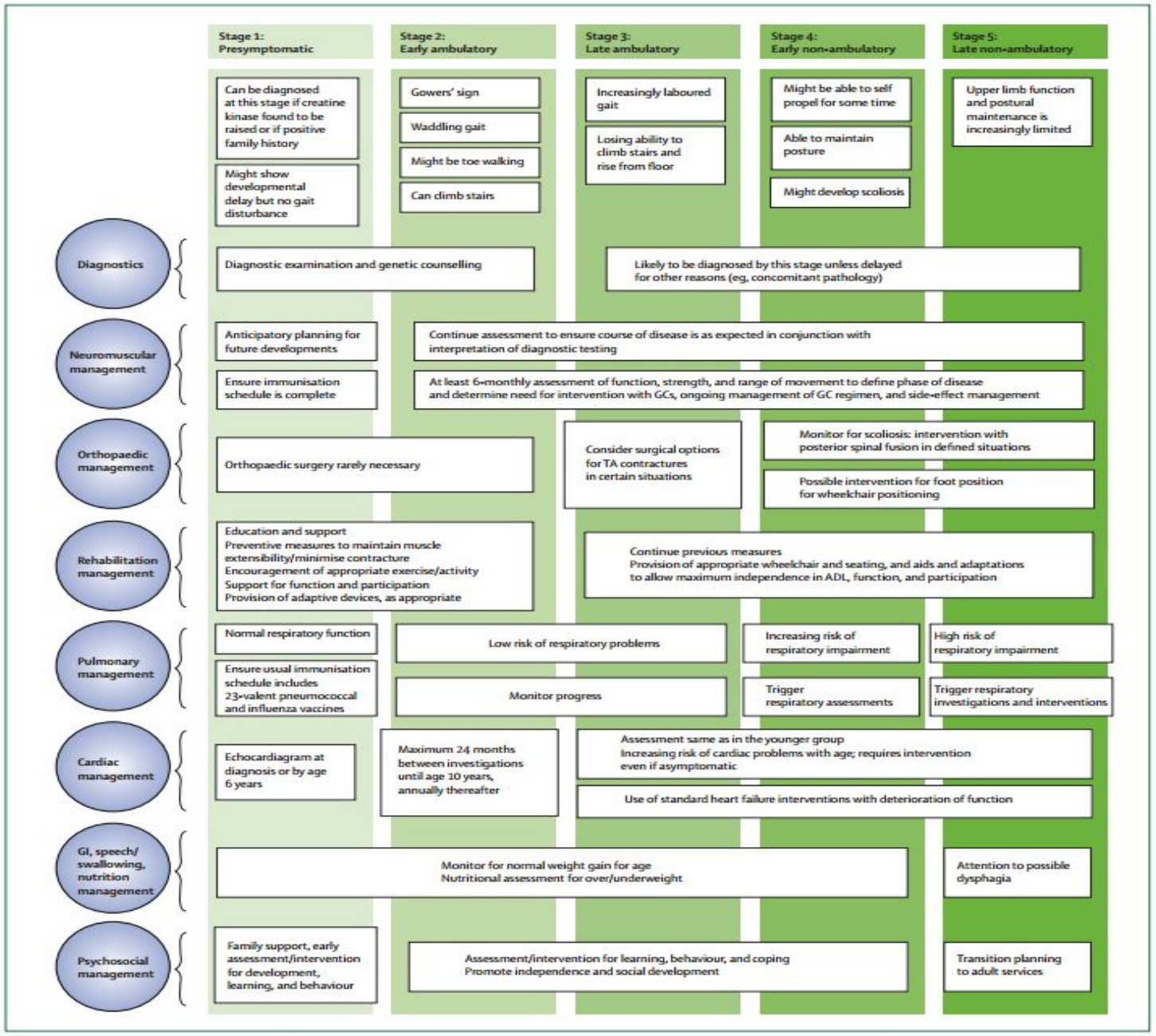

Figure 2: Stages of disease and care considerations
ADL=activities of daily living. GCs=glucocorticoids. GI=gastrointestinal. TA=tendo-Achilles.

Corticoides en Duchenne

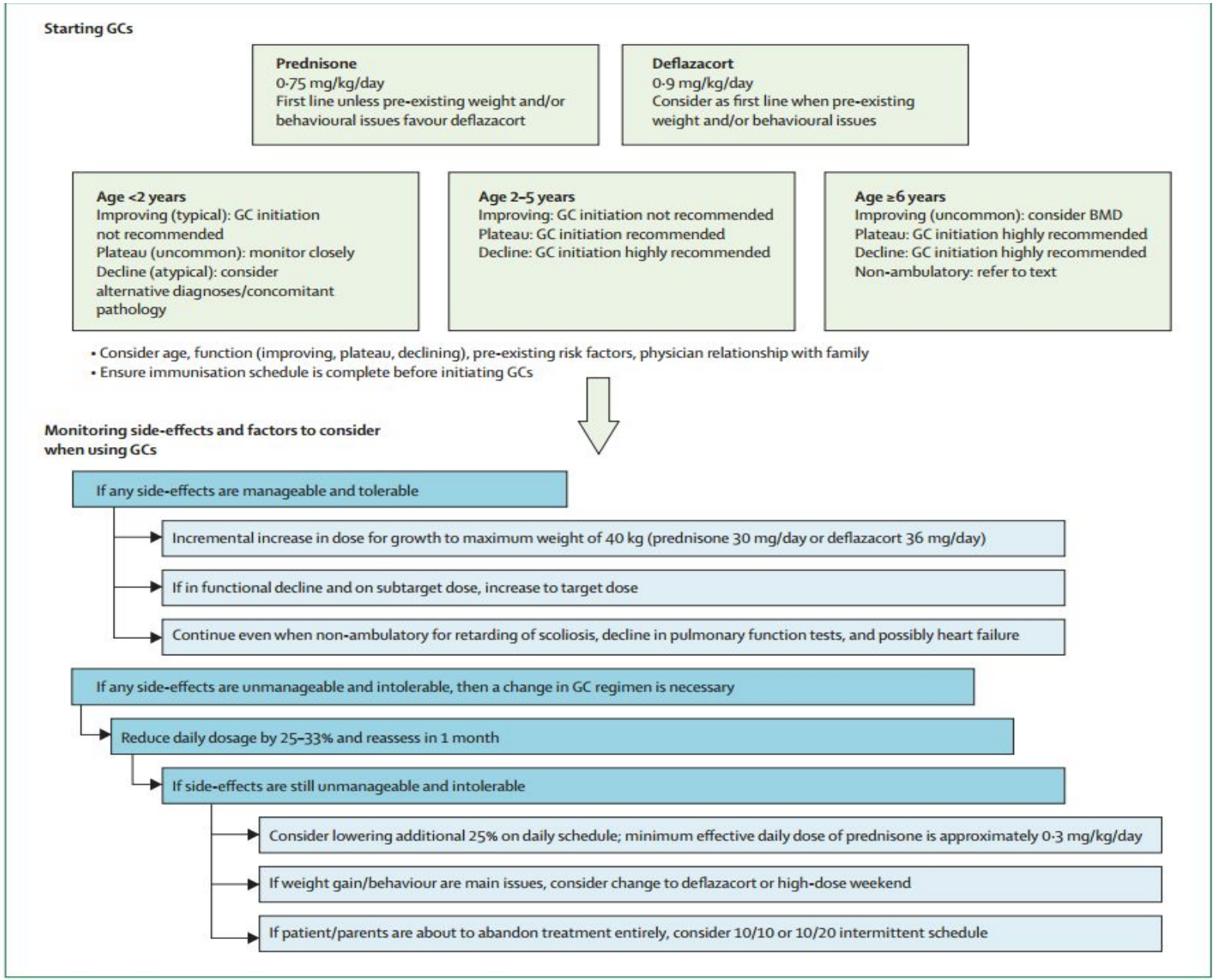

***Figure 4*: Schema for initiation and management of GC medication in Duchenne muscular dystrophy**[59,68,80]
See table 2 for more on monitoring side-effects. BMD=Becker muscular dystrophy. GC=glucocorticoid.

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Corticoides en Duchenne

Los distintos esquemas de tratamiento

	Prednisone dose*	Deflazacort dose*	Comments	In case of side-effects
Alternate day	0·75–1·25 mg/kg every other day	2 mg/kg every other day	Less effective but consider when a daily schedule has side-effects that are not effectively managed or tolerated	Must reduce dose if side-effects are not manageable or tolerable
High-dose weekend	5 mg/kg given each Friday and Saturday	Not yet tested	Less data on effectiveness as compared to a daily schedule Consider as an alternative to daily treatment, especially if weight gain and behavioural issues are problematic	Must reduce dose if side-effects are not manageable or tolerable
Intermittent	0·75 mg/kg for 10 days alternating with 10–20 days off medication	0·6 mg/kg on days 1–20 and none for the remainder of the month	Less effective but has fewer side-effects Consider as the least effective but possibly best tolerated regimen before abandoning steroid treatment altogether	Must reduce dose if side-effects are not manageable or tolerable

GC=glucocorticoid. *No set dose ranges have been clearly accepted as optimum.

Table 3: **Alternative GC dosing strategies**

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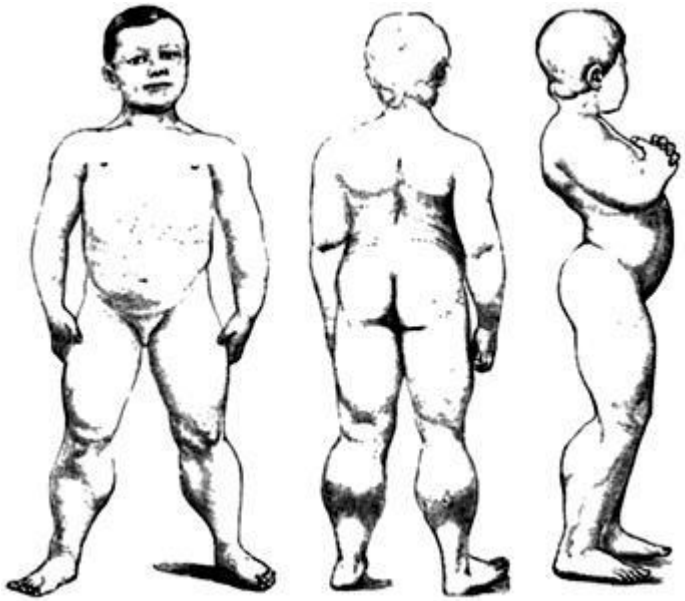
ASPECTOS IMPORTANTES DEL MANEJO

Manejo de complicaciones óseas/esqueléticas.

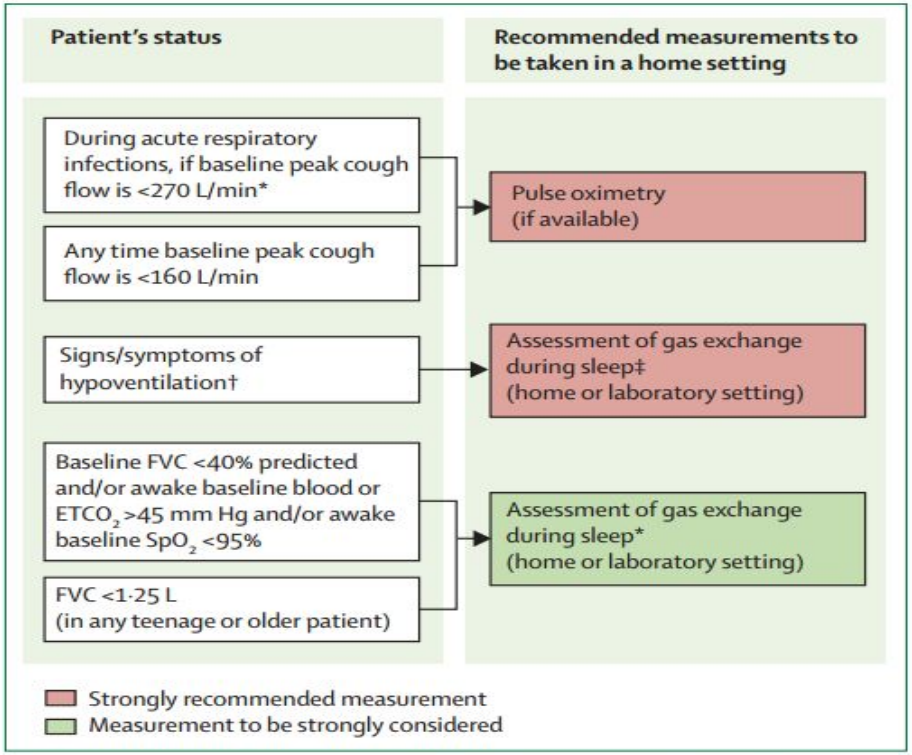


Bone-health issues	Recommended bone-health assessments		Possible bone-health interventions
Underlying factors for poor bone health Decreased mobility Muscle weakness Glucocorticoid therapy	**Suggested tests** **Serum** • Calcium • Phosphate • Alkaline phosphatase • 25-OH vitamin D level (in springtime or bi-annually) • Consider: magnesium, PTH level **Urine** • Calcium (for calciuria) • Sodium • Creatinine	**Bone imaging* study indications** **DEXA scan†** • Obtain a baseline at: Age 3+ years Start of glucocorticoid therapy • Repeat annually for those at risk: History of fractures On chronic glucocorticoid therapy DEXA *Z* score <–2 **Spine radiograph‡** • If kyphoscoliosis is noted on clinical examination therapy • If back pain is present, to assess vertebral compression fracture **Bone age (left wrist) radiography§** To assess growth failure (on or off glucocorticoid therapy)	**Possible interventions** **Vitamin D** • Vitamin D treatment for proven deficiency is necessary • Supplementation should be considered in all children if levels cannot be maintained **Calcium** • Calcium intake and possible supplementation should be carried out in consultation with a dietitian **Bisphosphonates** • Intravenous bisphosphonates for vertebral fracture are indicated • Oral bisphosphonates as treatment or as a prophylactic measure remain controversial
Resulting in: Fractures (long bone and vertebral) Osteopenia Osteoporosis Kyphoscoliosis Bone pain Reduced quality of life			

Figure 1: **Bone-health management**
Information provided in this figure was not derived from RAND Corporation–University of California Los Angeles Appropriateness Method data and was produced solely using expert discussion. DEXA=dual-energy x-ray absorptiometry. PTH=parathyroid hormone. *All imaging assessments should be done at a facility capable of performing and interpreting age-appropriate studies. †A DEXA scan is a better measure than plain film radiographs for detection of osteopenia or osteoporosis. DEXA scans, to assess bone mineral content or body composition, need to be interpreted as a *Z* score for children and a *T* score for adults (compared with age-matched and sex-matched controls). ‡Spine radiographs (posterior/anterior and lateral views) are used for the assessment of scoliosis, bone pain, and compression fractures. It is preferable to obtain them in the standing position, especially if bone pain is the presenting symptom. Useful information can still be obtained in the sitting position for the non-weight-bearing patient. §Bone-age measurements should be done in patients with growth failure (height for age <5% percentile or if linear growth is faltering). If abnormal (>2 SD below the mean), a referral needs to be made to a paediatric endocrinologist.

Lancet Neurol 2010; 9: 177–89

ASPECTOS IMPORTANTES DEL MANEJO
Evaluación de función ventilatoria (ambulatoria)

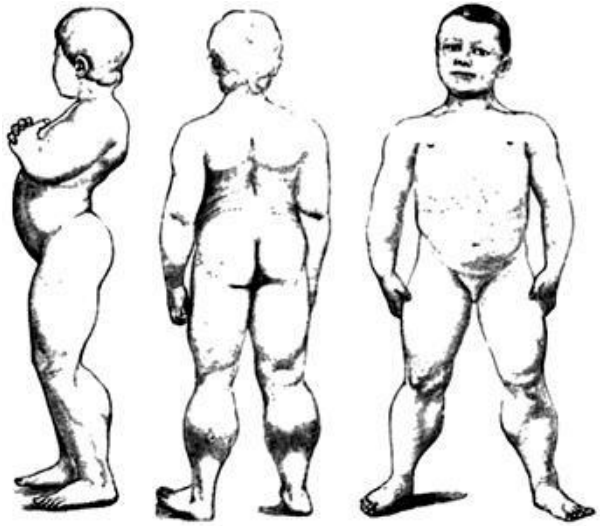


For the **American Academy of Pediatrics** see http://aapredbook.aappublications.org/

For the **CDC's information on influenza** see http://www.cdc.gov/flu/

Figure 3: **Respiratory assessment (at home) of patients with Duchenne muscular dystrophy**

$ETCO_2$=end-tidal CO_2. FVC=forced vital capacity. SpO_2=pulse oximetry. *All specified threshold values of peak cough flow and maximum expiratory pressure apply to older teenage and adult patients. †Signs/symptoms of hypoventilation include fatigue, dyspnoea, morning or continuous headaches, sleep dysfunction (frequent nocturnal awakenings [>3], difficult arousal), hypersomnolence, awakenings with dyspnoea and tachycardia, difficulty with concentration, frequent nightmares. ‡Dual-channel oximetry-capnography in the home is strongly recommended, but other recommended methods include home oximetry during sleep and polysomnography, the method of choice being determined by local availability, expertise, and clinician preference.



Lancet Neurol 2010; 9: 177–89

ASPECTOS IMPORTANTES DEL MANEJO
Evaluación de función ventilatoria (hospitalizado)

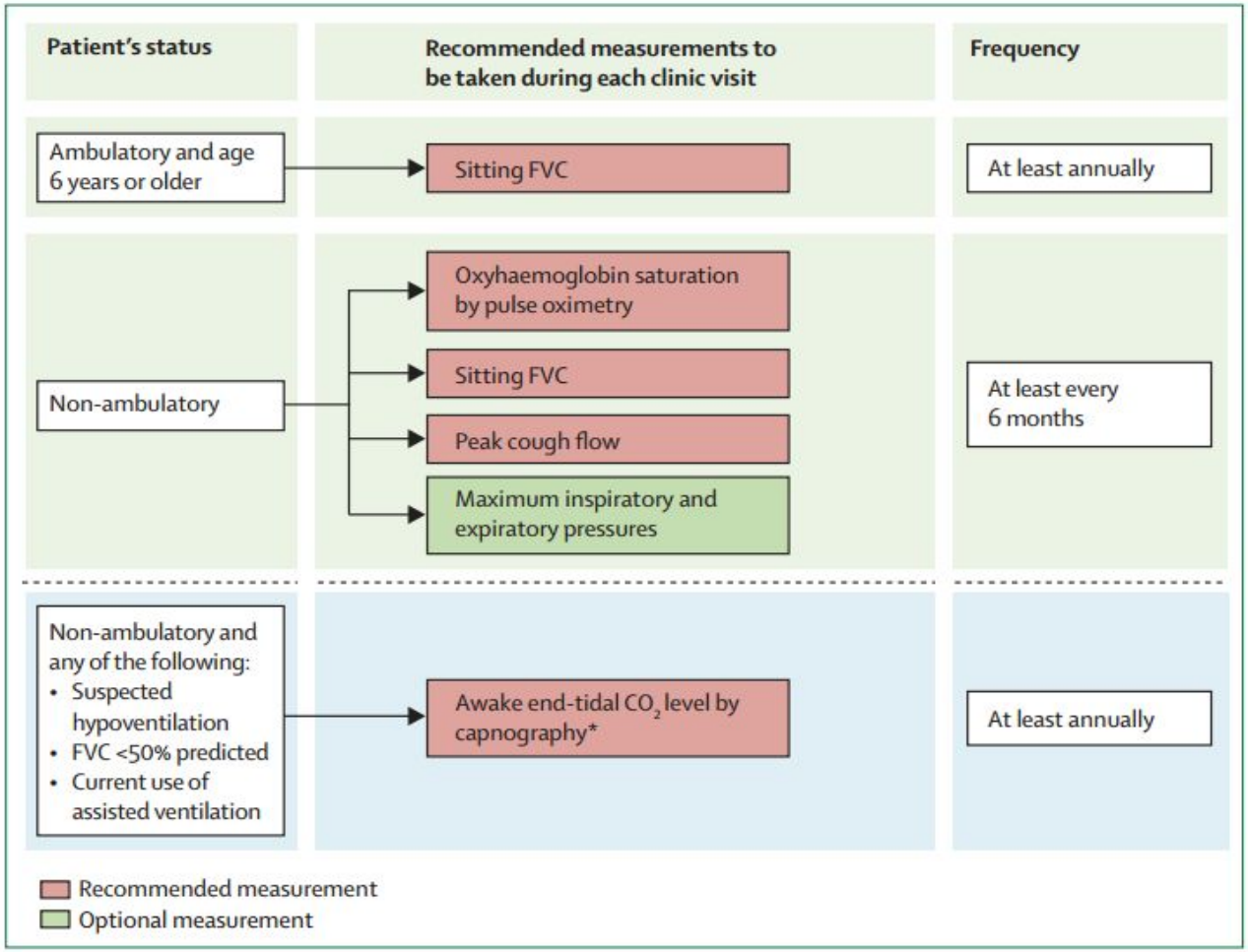


Figure 2: **Respiratory assessment (in the clinic) of patients with Duchenne muscular dystrophy**
FVC=forced vital capacity. *Also measure end-tidal CO_2 any time that a patient with an FVC of <50% predicted has a respiratory infection.

Lancet Neurol 2010; 9: 177–89

ASPECTOS IMPORTANTES DEL MANEJO

Manejo respiratorio

Bryson Foster, de 11 años, Embajador Nacional de Buena Voluntad 2012 de la ADM. Foster fue diagnosticado con Distrofia Muscular de Duchenne a los cinco años de edad. *Fotografía cortesía de la Asociación de Distrofia Muscular.*http://www.oandp.com/articles/2012-04_01.asp

Panel 1: **Respiratory interventions indicated in patients with Duchenne muscular dystrophy**

Step 1: volume recruitment/deep lung inflation technique

Volume recruitment/deep lung inflation technique (by self-inflating manual ventilation bag or mechanical insufflation–exsufflation) when FVC <40% predicted

Step 2: manual and mechanically assisted cough techniques

Necessary when:

- Respiratory infection present and baseline peak cough flow <270 L/min*
- Baseline peak cough flow <160 L/min or maximum expiratory pressure <40 cm water
- Baseline FVC <40% predicted or <1·25 L in older teenager/adult

Step 3: nocturnal ventilation

Nocturnal ventilation† is indicated in patients who have any of the following:

- Signs or symptoms of hypoventilation (patients with FVC <30% predicted are at especially high risk)
- A baseline SpO_2 <95% and/or blood or end-tidal CO_2 >45 mm Hg while awake
- An apnoea–hypopnoea index >10 per hour on polysomnography or four or more episodes of SpO_2 <92% or drops in SpO_2 of at least 4% per hour of sleep

Optimally, use of lung volume recruitment and assisted cough techniques should always precede initiation of non-invasive ventilation

Step 4: daytime ventilation

In patients already using nocturnally assisted ventilation, daytime ventilation‡ is indicated for:

- Self extension of nocturnal ventilation into waking hours
- Abnormal deglutition due to dyspnoea, which is relieved by ventilatory assistance
- Inability to speak a full sentence without breathlessness, and/or
- Symptoms of hypoventilation with baseline SpO_2 <95% and/or blood or end-tidal CO_2 >45 mm Hg while awake

Continuous non-invasive assisted ventilation (with mechanically assisted cough) can facilitate endotracheal extubation for patients who were intubated during acute illness or during anaesthesia, followed by weaning to nocturnal non-invasive assisted ventilation, if applicable

Step 5: tracheostomy

Indications for tracheostomy include:

- Patient and clinician preference§
- Patient cannot successfully use non-invasive ventilation
- Inability of the local medical infrastructure to support non-invasive ventilation
- Three failures to achieve extubation during critical illness despite optimum use of non-invasive ventilation and mechanically assisted cough
- The failure of non-invasive methods of cough assistance to prevent aspiration of secretions into the lung and drops in oxygen saturation below 95% or the patient's baseline, necessitating frequent direct tracheal suctioning via tracheostomy

FVC=forced vital capacity. SpO_2=pulse oximetry. *All specified threshold values of peak cough flow and maximum expiratory pressure apply to older teenage and adult patients. †Recommended for nocturnal use: non-invasive ventilation with pressure cycled bi-level devices or volume cycled ventilators or combination volume-pressure ventilators. In bi-level or pressure support modes of ventilation, add a back-up rate of breathing. Recommended interfaces include a nasal mask or a nasal pillow. Other

Lancet Neurol 2010; 9: 177–89

ASPECTOS IMPORTANTES DEL MANEJO

Detección de problemas asociados:
Complicaciones inherentes a cada miopatía en particular
Ej Cardíacas

Sensoriales

Nutricionales

Educación:
Desarrollo cognitivo acorde a edad

Integrar a sistema escolar regular con las intervenciones necesarias.

Asesoría pedagógica

Control pediátrico periódico, vacunas,
Información acerca de derechos sociales, legales, beneficios, SENADIS, Chile inclusivo.

ESCOLARIDAD
Un aspecto frecuentemente considerado de baja prioridad es la inserción escolar de los niños.
Es importante mantener escolaridad, con las adaptaciones curriculares y apoyo psicológico necesarios de acuerdo al perfil cognitivo y síntomas asociados y tratamiento de trastornos del aprendizaje.
El Trastorno por Déficit de Atención con o sin hiperactividad es muy frecuente y requiere tratamiento ambiental y farmacológico apropiado.

101 Consejos de "ayuda fácil" para pacientes con padecimiento neuromuscular

Irwin M. Siegel, M.D.
Patricia Casey, M.S., OTR/L

Introducción

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Para Vestirse

20-25:
La Comunicació

26-36:
Para Sentarse, l
Movilidad en Ge

37-50:
Para su Entrete

51-52:
Los Quehaceres

Dispositivos adicionales

53-61:
Dormir y Descansar

62-68:
El Arreglo Personal

69-73:
Uso del W.C.

74-90:
Las Comidas

91-101:
Ejercicios y Superación de Contracturas

Impacto de la ventilación nocturna a domicilio

Distrofia Muscular de Duchenne: Sobrevida promedio 1960: 14.4 años. Sobrevida prom. 1990 post ventilación nocturna: 25,3 años *"Survival in Duchenne muscular dystrophy: improvements in life expectancy since 1967 and the impact of home nocturnal ventilation" Eaglea et al. Neuromuscular Disorders 12 (2002) 926–929*

Sd. Hipoventilación central congénita y las ENM: causas más frecuentes en Suiza de ventilación a domicilio *Rev Pediatrica Vol 13 N°2 2002*

http://annamaryas.com/2013/03/20/eteplirsen-the-end-to-duchenne-muscular-dystrophy/

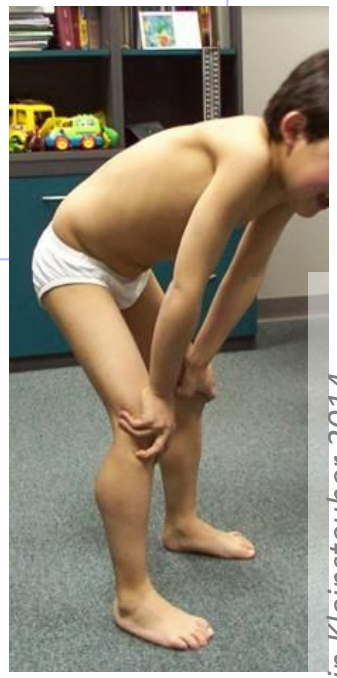
MENSAJES

La clínica sigue siendo esencial.
diagnóstico y consejo genético oportunos
diagnóstico es clave para un tratamiento RACIONAL

Basado en evidencia
Actualizado

PENSAR en las recomendaciones de estudio y tratamiento a la luz de la evidencia actual

TRATAMIENTO



Dra. Karin Kleinsteuber 2014

No perder de vista los objetivos de este proceso: los PACIENTES

Las fotografías y videos mostrados han sido autorizadas por los pacientes y/o sus padres; o en las que se señala están disponibles en la red.

World Duchenne Awareness Day Celebration 2015

Gracias

World Duchenne Awareness Day Celebration 2015

Dra. Karin Kleinsteuber S.

Profesor Asociado, Programa de Formación de Especialistas en Neurología Pediátrica

Universidad de Chile - Clínica Las Condes