Bone Health and Beyond:

Comprehensive Endocrine Care in Duchenne Muscular Dystrophy

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Faculty Disclosure

- Nothing to disclose



Educational Need/Practice Gap

Gap = Endocrine complications of Duchenne muscular dystrophy (DMD) are not consistently recognized, screened for, or managed according to established care guidelines.



Need = Healthcare providers need increased awareness of the endocrine complications associated with DMD and evidence-based treatment strategies to improve outcomes.

Objectives

Upon completion of this educational activity, you will be able to:

- **Identify** the major endocrine complications associated with Duchenne muscular dystrophy (DMD) and chronic glucocorticoid therapy.
- **Apply** guideline-based strategies for the screening and management of adrenal insufficiency, osteoporosis, growth failure, and delayed puberty.
- **Evaluate** bone health in individuals with DMD using fracture history, vertebral imaging, DXA, and laboratory assessment to guide osteoporosis treatment.



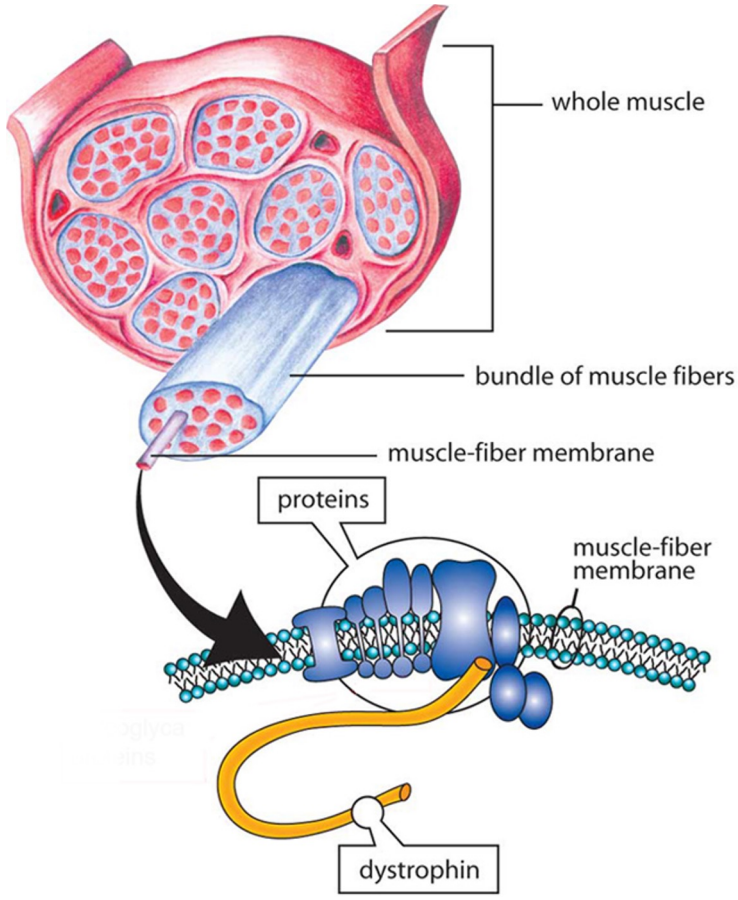
Expected Outcome

- Incorporate guideline-based strategies for the prevention and management of adrenal insufficiency, osteoporosis, growth failure, and delayed puberty into routine clinical care.
    - Improve bone health assessment and treatment decisions using fracture history, vertebral imaging, DXA, and laboratory evaluation.
        - Reduce preventable complications, including fractures and adrenal crises, to improve patient outcomes and quality of life.

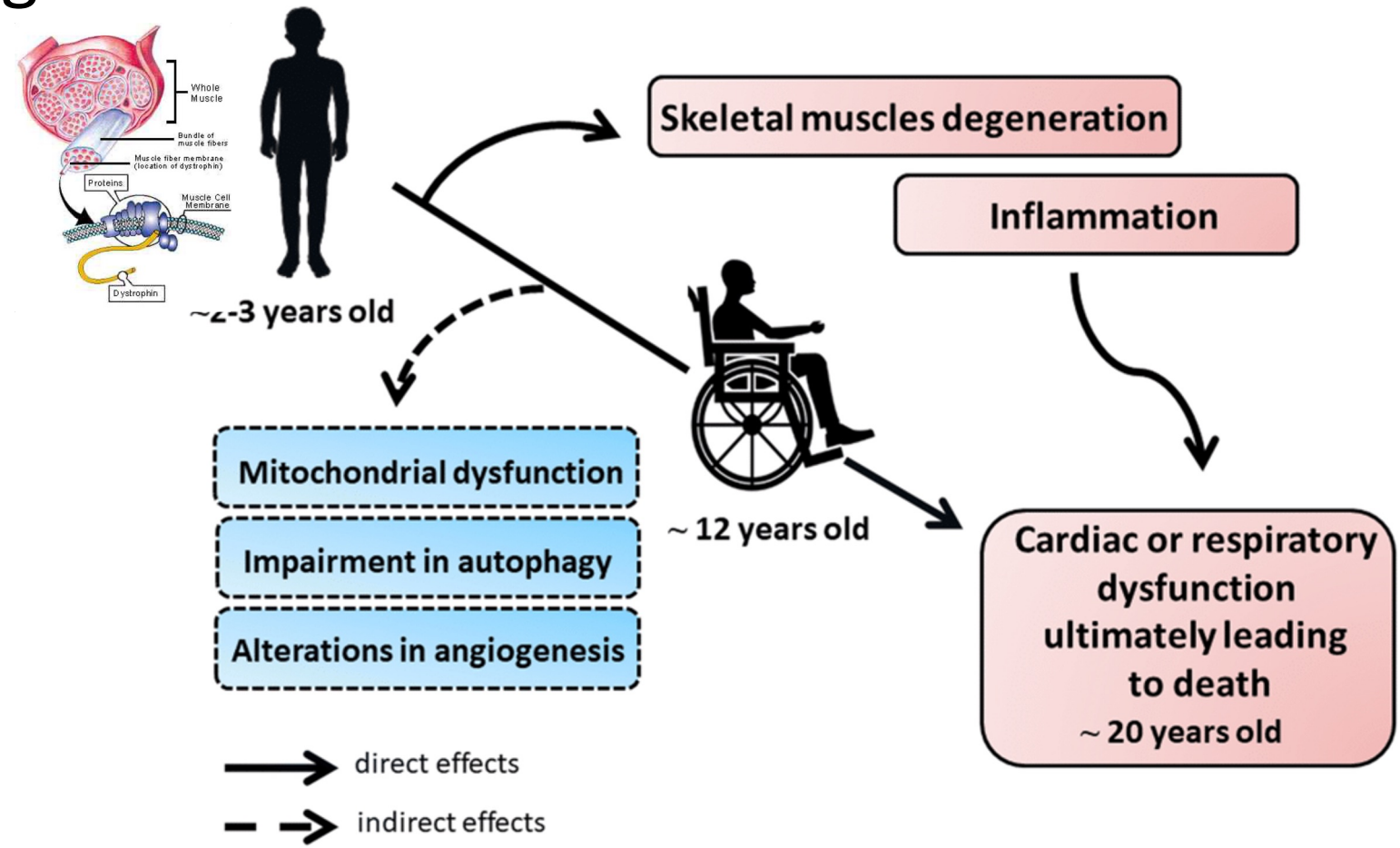


Duchenne Muscular Dystrophy

- X-linked recessive
  - 2/3 receive dystrophin mutation from carrier mother
  - 1/3 of mutations occur de novo
  - **Most of our patients are cis-males**
- Affects 1 in 5,000 male births
- Out-of-frame mutations --> Duchenne
- In-frame mutation --> Becker Muscular Dystrophy



Progression of DMD



Podkalicka et al. 2008.

	Gene Therapy (Elevidys)	Exon Skipping (Exondys 51, Vyondys 53, Viltepso, Amondys 45)
Mechanism	AAV vector delivers **micro-dystrophin gene** → production of truncated but functional dystrophin	Antisense oligo bind pre-mRNA → skip targeted exon → restore reading frame → allow truncated dystrophin
Administration	**One-time IV infusion**	**Weekly IV infusions** (lifelong)
FDA	2023	2016–2021
Durability	Potentially **long-lasting** (uncertain durability)	Requires **lifelong dosing**
Safety	Immune response to AAV; **liver toxicity**; 2 recent fatal cases	Generally mild infusion reactions
Cost	~$3.2M one-time	~$300k–500k annually, lifelong
Steroid Use	**Yes – additional high-dose corticosteroids**	**Yes – patients usually remain on standard chronic steroids**
Key Limitation	Following fatal acute liver failure, the FDA restricted Elevidys to ambulatory patients with DMD aged 4 years and older and added a boxed warning for liver failure.	specific mutations (exon 51, 53, 45) – covers ~30% of patients; modest dystrophin increase

Clinical Case

James is an 4-year-old, 0.7 m^2 boy who presents for evaluation of Duchenne Muscular Dystrophy (DMD), (diag. deletion of exons 45-50 in dystrophin gen

Activity:
Able to walk without assistance.
Walking on his toes intermittently.

Diet:
Picky eater.
Does not like milk or cheese.

Met with Neurology, who started 12 mg prednisolone daily.



<u>Family History:</u>

- Lives with mom, dad, 2-year-old brother
- Maternal and paternal grandmothers: osteoporosis

Referral to Endocrinology





2018 Consensus Guidelines

THE LANCET Neurology

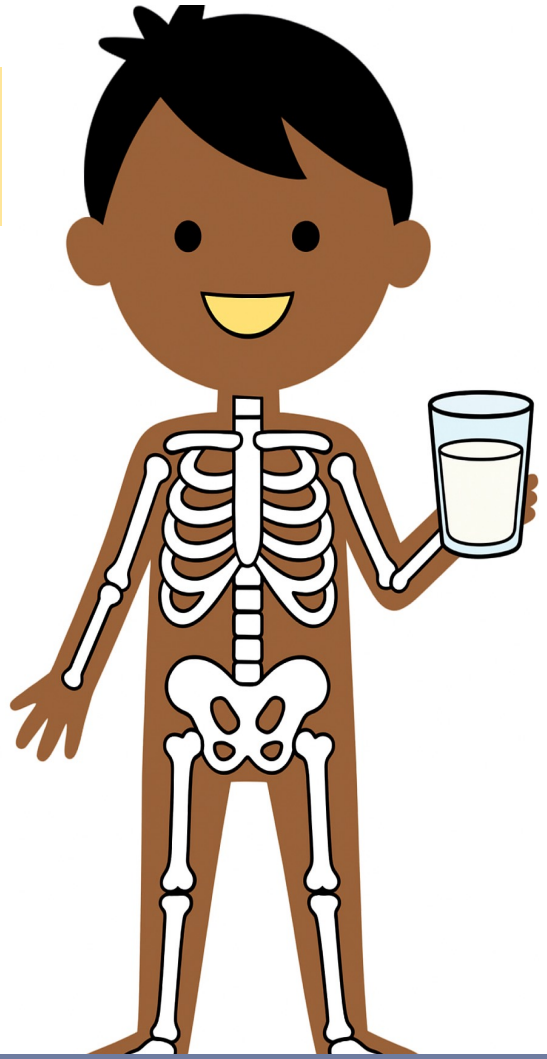
Bone Health Management

- Assess lateral spine x-rays for occult fracture @ diagnosis & @ start of steroids, then Q1 year on steroids, Q2 years if not
- Consider bisphosphonates at first sign of fracture:
  - Genant grade 1 or higher vertebral fracture or first long bone fracture

Endocrine Management

- Measure standing height Q6 months
- Assess non-standing growth every 6 months
- Assess pubertal status every 6 months starting by age 9 years
- Provide family education and stress dose steroid prescription if on glucocorticoids

ENDOCRINE HEALTH PLAN in DMD:



Adrenal
(secondary AI)

Bone
(osteoporosis & fracture)

Carbs
(steroid-induced insulin resistance)

Development
(puberty & growth)

Adrenal
(secondary AI)



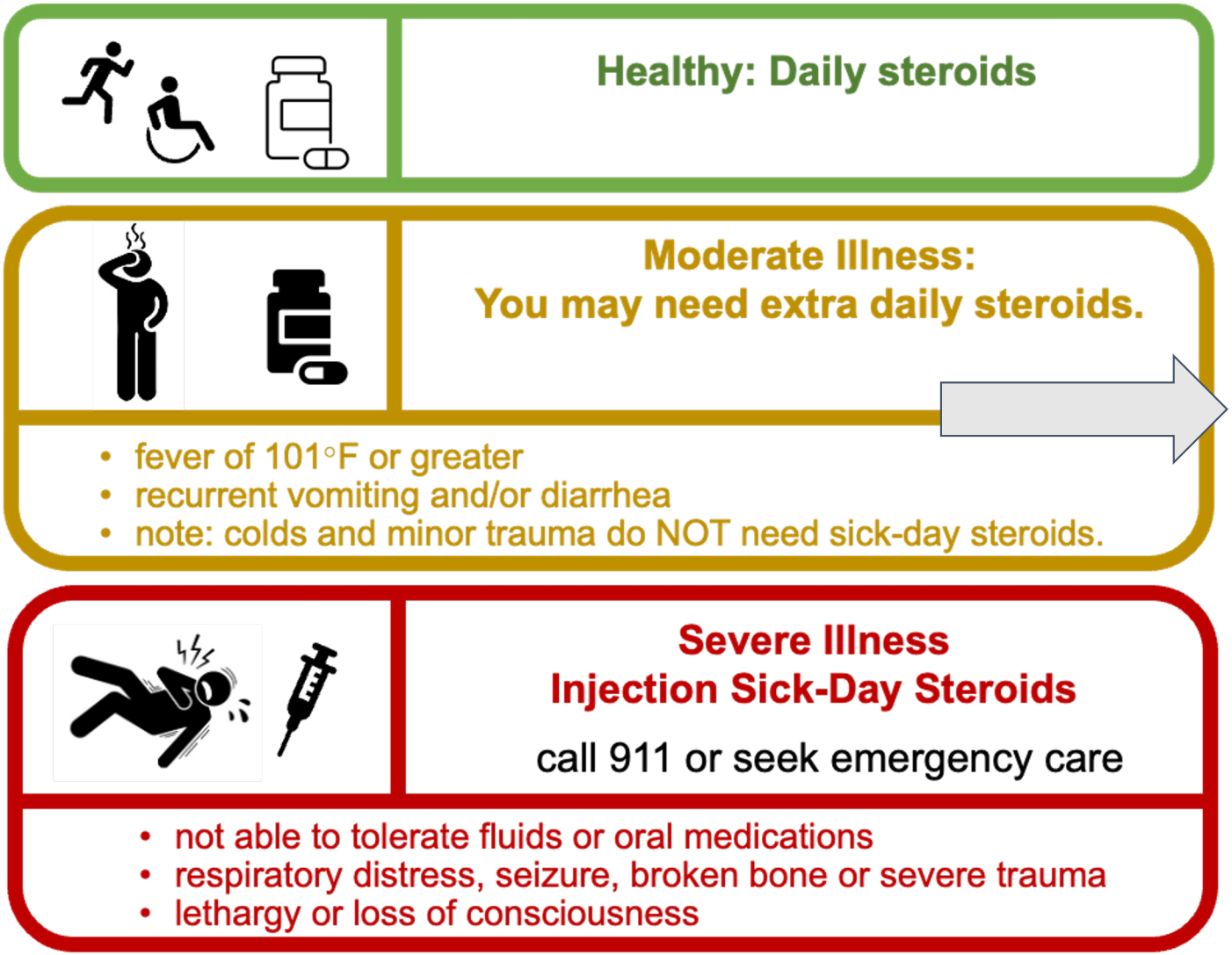
Adrenal insufficiency

- Calculate total daily dose: 12x4 = 48, divide by BSA 0.7m2 = 69 mg/m2/day

- Educate on signs, symptoms, and management of adrenal crisis

- Stress dosing for patients taking suppressive doses of prednisone/deflazacort/vamorolone

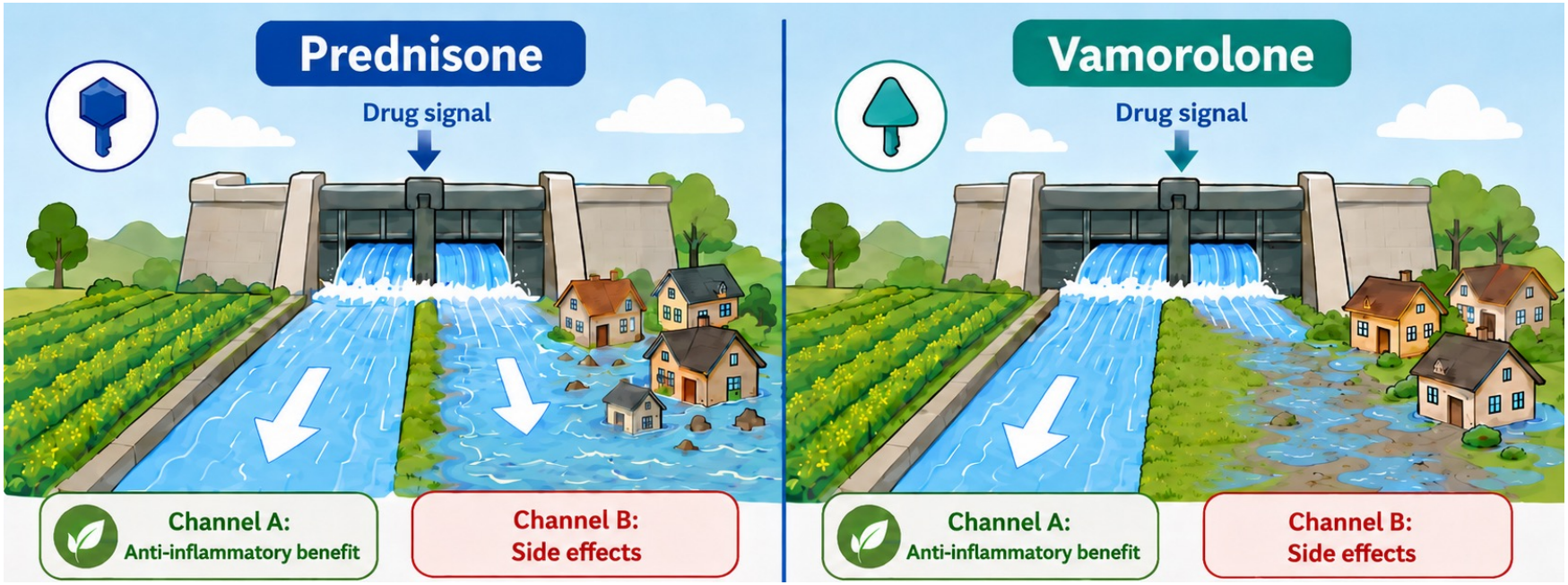
- Prescribe IM hydrocortisone for home
  - 50 mg for children aged <12 years old
  - 100 mg for children aged 12 years +



Who needs an oral sick day plan?

- Patients on less than 50 mg/m2/day hydrocort equivalents
- Anyone on Vamorolone.

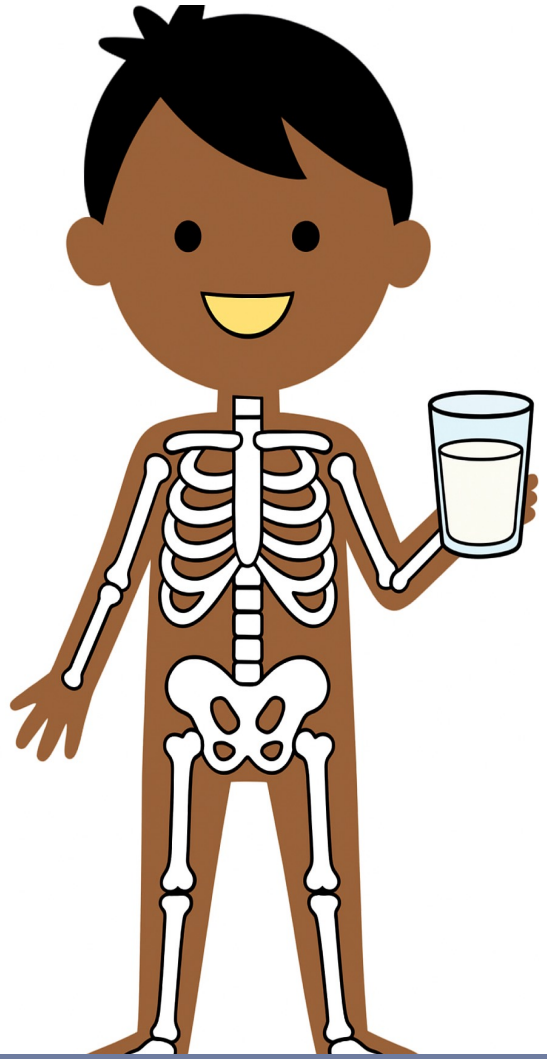
Classic Steroids | **Dissociative Steroid**



CANNOT BE USED FOR STRESS DOSING / SICK DAYS

Feature	Prednisone	Deflazacort (2017)	Vamorolone (2023)
Glucocorticoid Type	Classic Steroid	Classic Steroid	Dissociative Steroid
Anti-inflammatory Effect	High	High	High
Anti-Mineralocorticoid Activity	None	None	**Mineralocorticoid antagonist, SDS with pred/ hydrocort**
Adrenal Suppression	100%	100%	**100% & cannot use for stress dosing**
Growth	Suppression	Suppression	Less growth suppression
Bone Health	High risk of osteoporosis	High risk but improved over Pred	Lowest spine fracture risk
Weight Gain	Significant	Less gain than others	Similar to Prednisone
Puberty Impact	Delayed	Delayed	????

ENDOCRINE HEALTH PLAN in DMD:

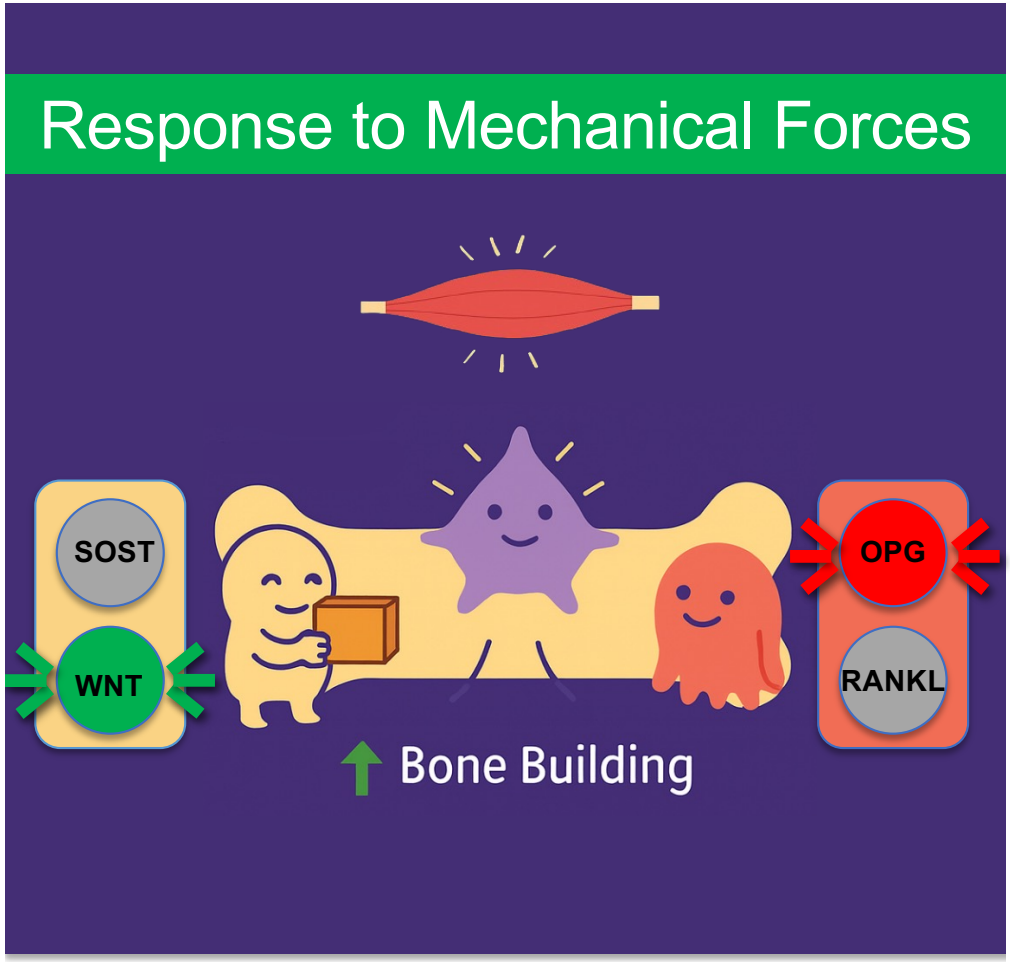


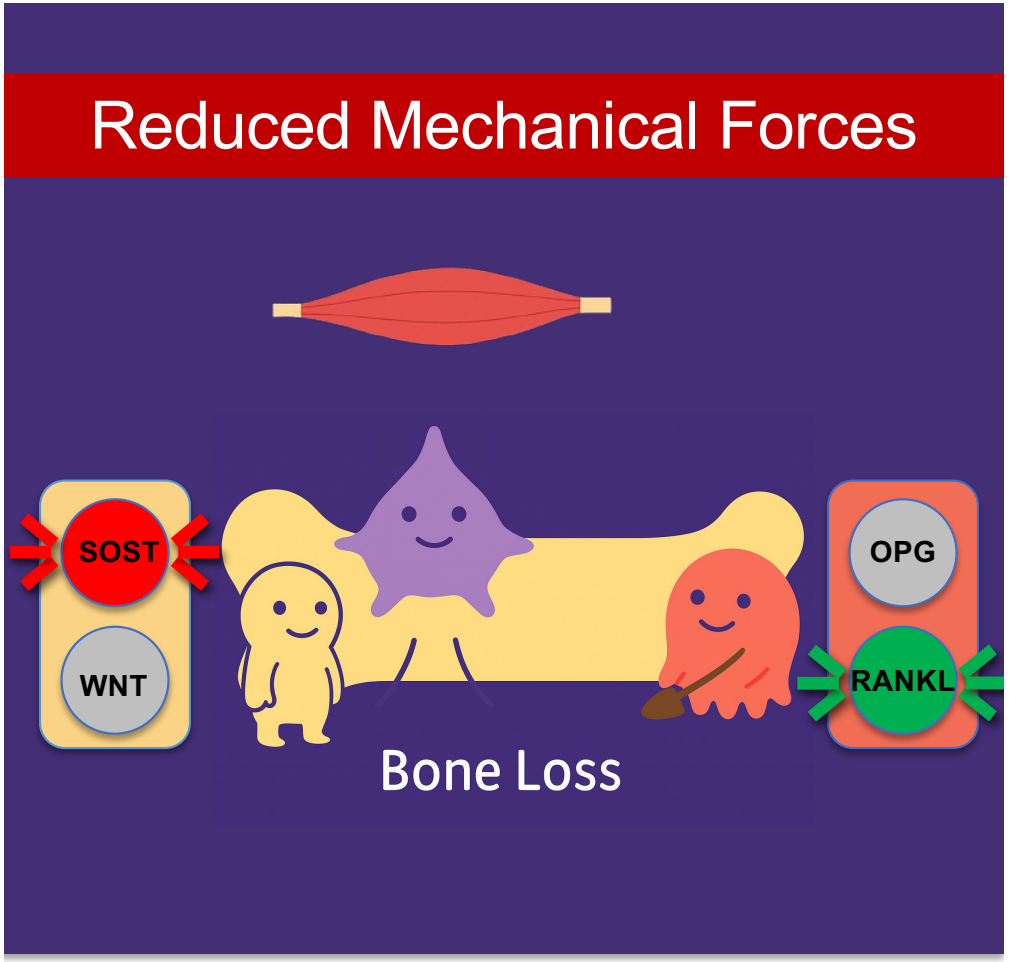
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(steroid-induced insulin resistance)

Development
(puberty & growth)





SOST = Sclerostin

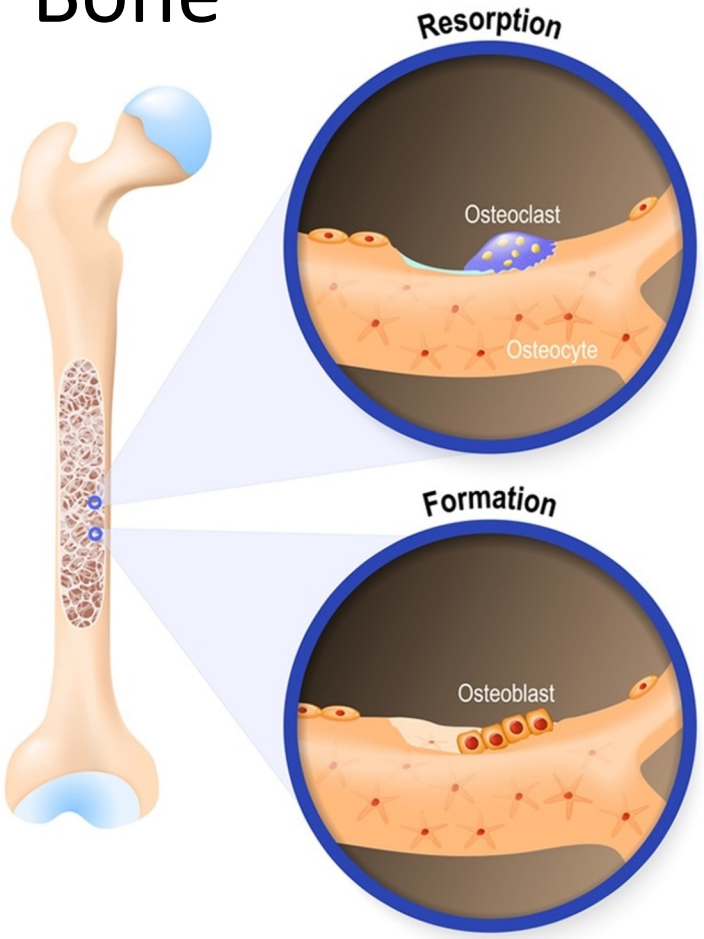
OPG = osteoprotegerin | RANKL = RANK Ligand

Glucocorticoids: **Directly** Bad for Bone

Uncoupled Bone turnover

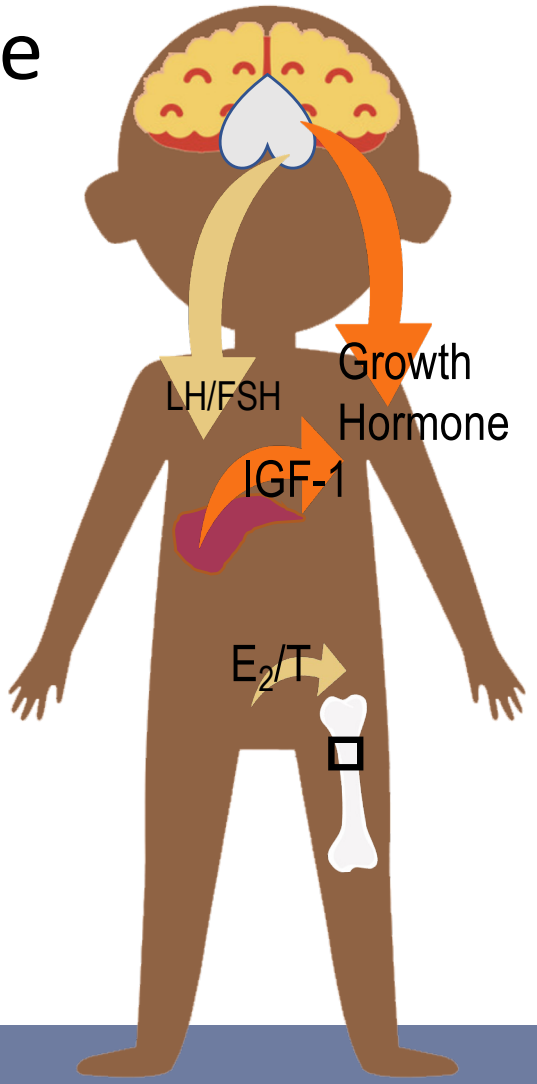
Low formation, high resorption

Cell Type	Glucocorticoids	Inflammation
Osteoblast		
Differentiation	↓	↓
Function	↓	↓
Survival	↓	↓
Osteocyte		
Survival	↓	↓
Osteoclast		
Differentiation	↑	↑
Survival	↑	↑



Glucocorticoids: **Indirectly** Bad for Bone

- Endocrine
  - Reduced GH/IGF-I
  - Reduced gonadal steroids
- Calcium
  - less intestinal absorption
  - more renal loss



What is our Bone Health Workup in DMD?



1 Laboratory Evaluation

- 25(OH)D
- Alkaline phosphatase
  - Provides a measure of osteoblast activation but is often low in patients with reduced mobility
- PTH, Calcium, Phos, and Mag
- Urine Calcium & Creatinine
- Renal function: BUN, Cr→cystatin C (low muscle)

2 Imaging



- DXA Bone Density Scan
- Lateral thoracolumbar spine x-ray
  - low-trauma vertebral compression fractures



- Q 1-2 yr if on steroids
- Q 2-3 yr if not on steroids
- Anytime with back pain or ≥0.5 SD Δ in spine BMDZ

THE LANCET Neurology 2018 Consensus Guidelines

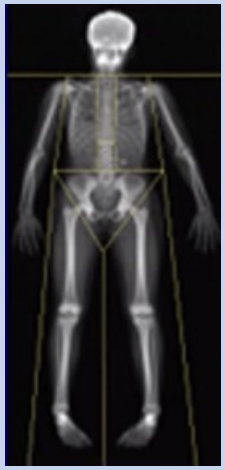
Annual DXA - monitor bone density



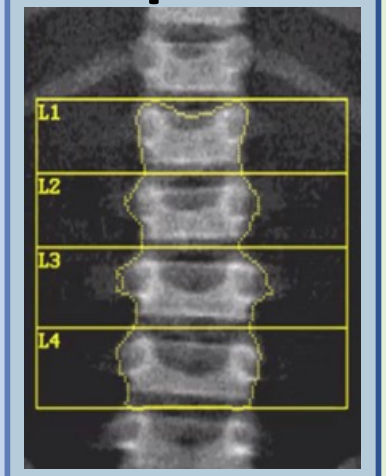
Image from: Hologic, Inc. www.hologic.com

Five Body Sites for DXA

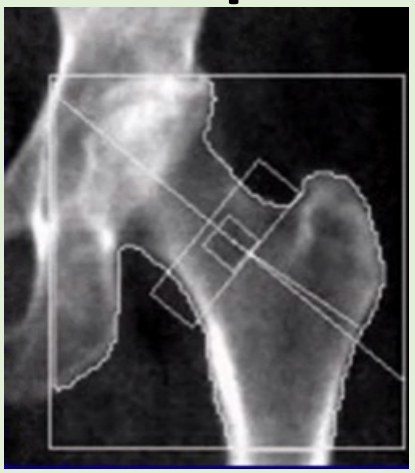
TBLH



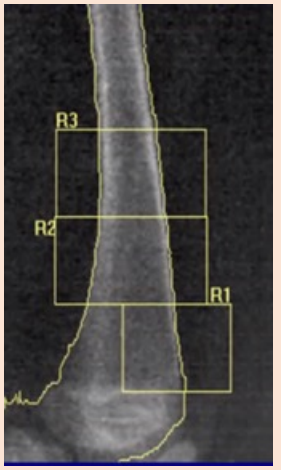
Spine



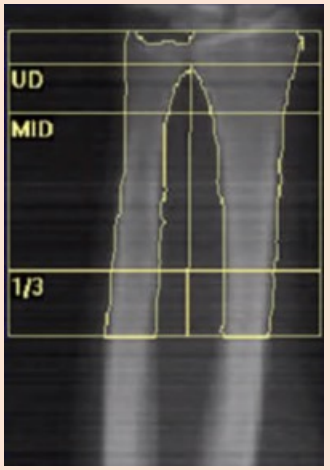
Hip



Distal femur



Forearm



Recommended sites for age 4-15

Recommended sites for age 16+

If hardware or inability to hold still

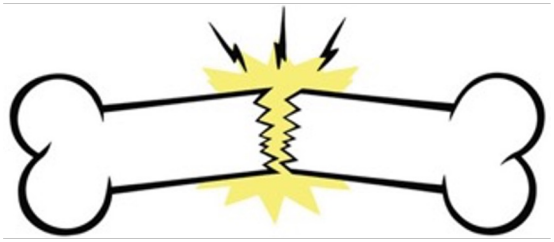
2013 & 2019 ISCD Official Positions

Common questions about DXA

	How much radiation is a DXA scan?	**0.4 microsieverts = ½ day on earth**  **vs. chest x-ray (100 µSv) = ~2 weeks**
	Do I always have to get my DXA on the same machine?	It's best to use the same scanner each year.
	How frequently should a DXA be repeated?	No more than once a year • insurance will not cover more frequently • Bones are slow ☺

Definition of Pediatric Osteoporosis

1



"Clinically Significant Fracture History"

Any vertebral compression fractures

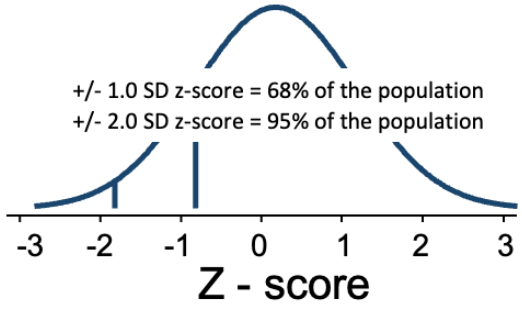


and/or

2+ long-bone fractures by age 10 yr

3+ long-bone fractures by age 19 yr

2



Recall:
Pediatric Osteoporosis = 1+2

NOTE FOR DMD:
Consider treatment in a patient

- **with declining BMD**
- **and/or declining BMD z-scores**

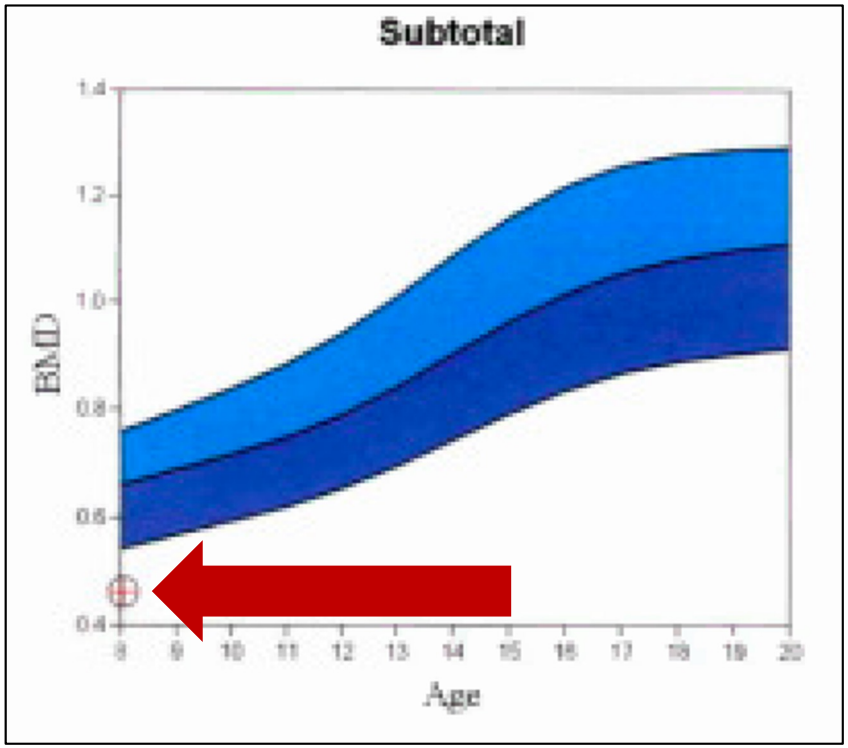
Back To Our Patient:

LABS

- BUN: 12 mg/dL
- Creatinine: 0.2 mg/dL Low
- Calcium: 9.0 mg/dL
- Phosphorus: 4.5 mg/dL
- Magnesium: 2.0 mg/dL
- Alk Phos: 177 unit/L
- PTH Level: 85 pg/mL HIGH
- 25-OH Vit D: 17.2 ng/mL Low
- Urine Calcium Low

Low BMD, Low D and Calcium Deficiency with mild Secondary Hyperpara, but without "Clinically Significant Fracture History" → **not yet Pediatric Osteoporosis**

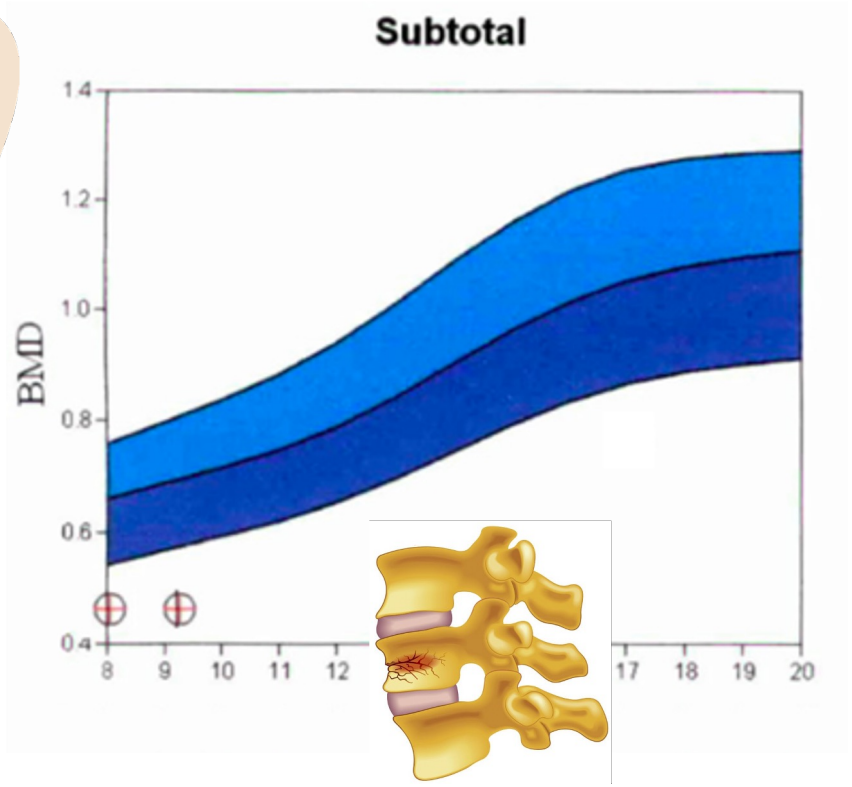
TBLH z-score:-3.1



Back To Our Patient:

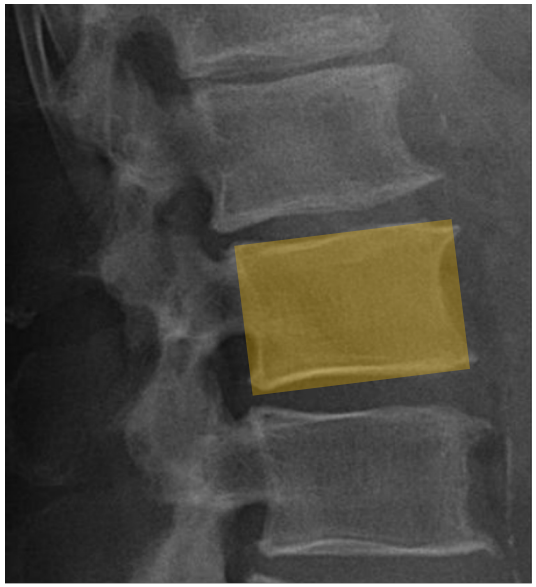
- Calcium
- Vitamin D
- Physical Therapy
- Optimize Chronic Disease

Vitamin D improved to 45 ng/mL,
PTH and Urine Calcium normalized

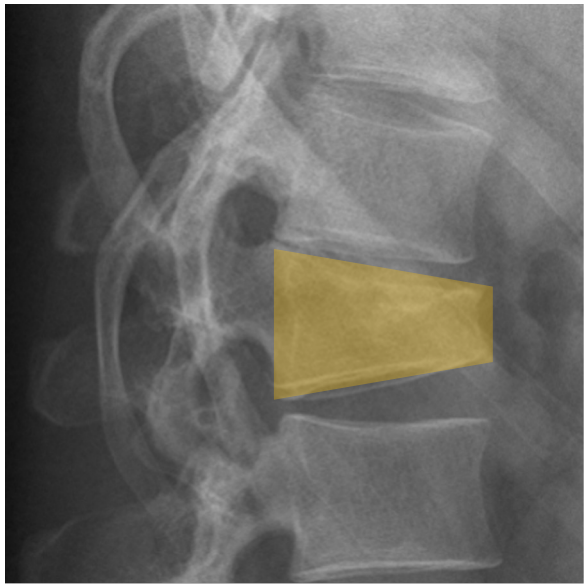


1 year later
spinal compression fracture

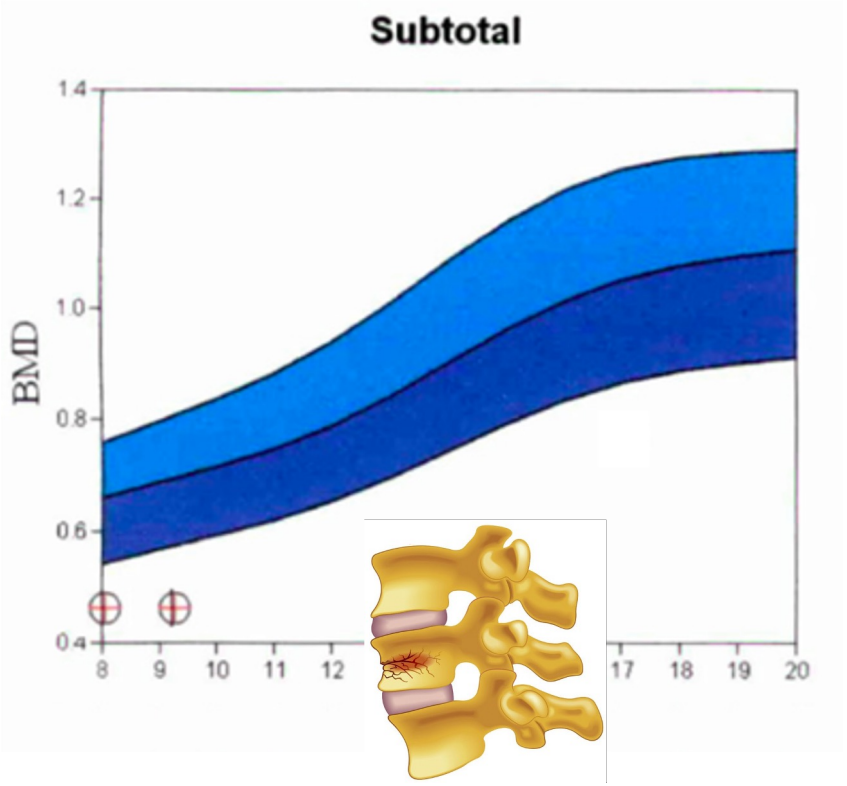
Back To Our Patient:



Healthy



Fracture



1 year later
spinal compression fracture

Offer Bisphosphonates

Mechanism:

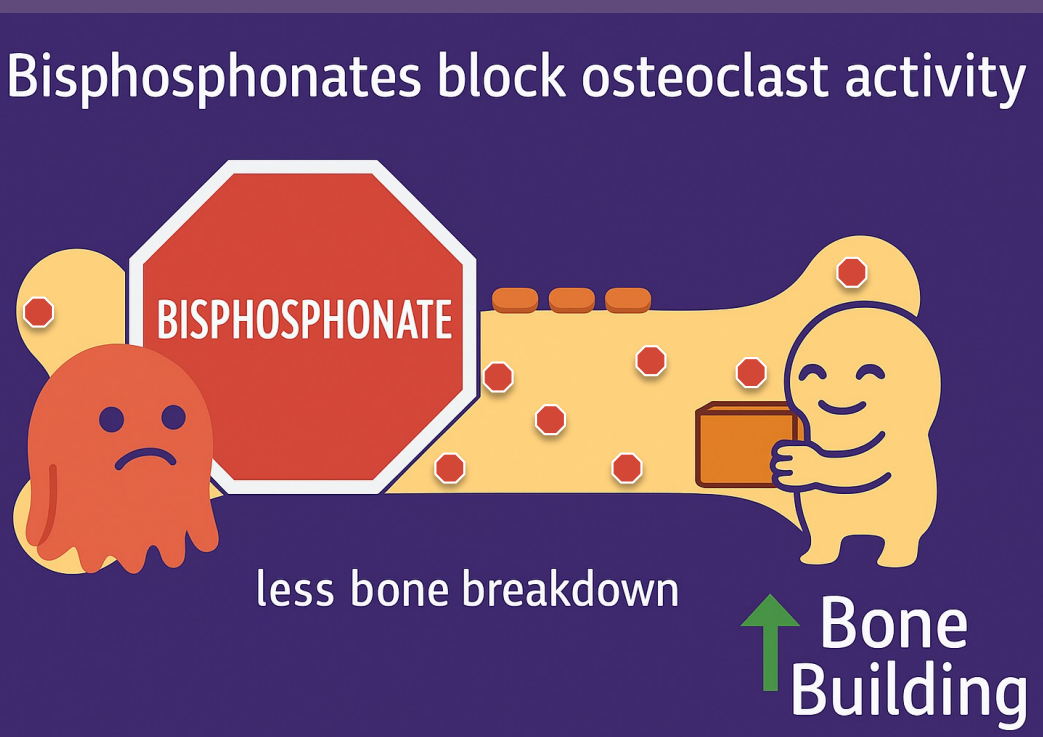
Bisphosphonates get incorporated into bone → osteoclasts ingest them while resorbing bone → toxic to osteoclasts → ↓ activity & survival.

Zoledronic Acid / Zometa / Reclast (IV)

- Risk of low calcium (ensure Vit D >30 ng/mL)
- Risk for renal complications → check kidney function
- Make dentist aware
  (risk of osteonecrosis of the jaw, rare)

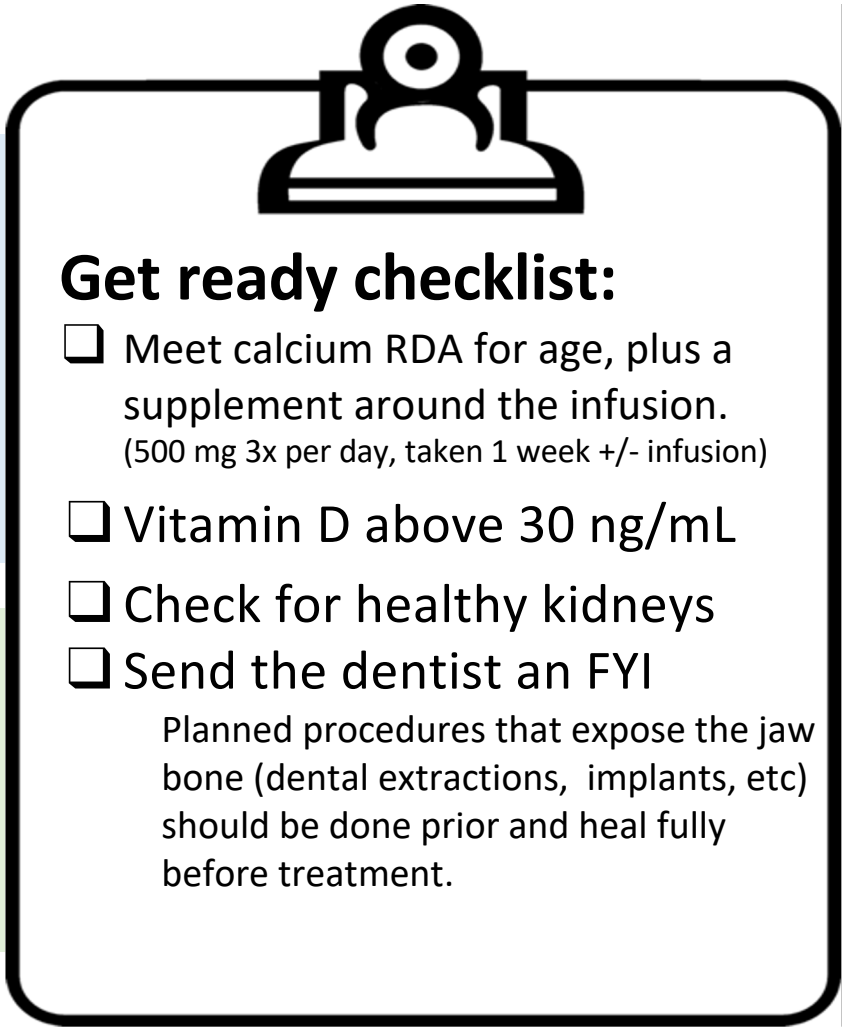
Alendronate / Fosamax (Oral)

- Less potent / Fewer risks
- Taken on an empty stomach with 8 oz plain water
- Sit upright for 30 minutes after taking
  (to avoid esophageal irritation)



Mitigating Risks of Bisphosphonate Infusion

ALWAYS FIRST treat hypocalcemia, vitamin D deficiency, and rickets/osteomalacia



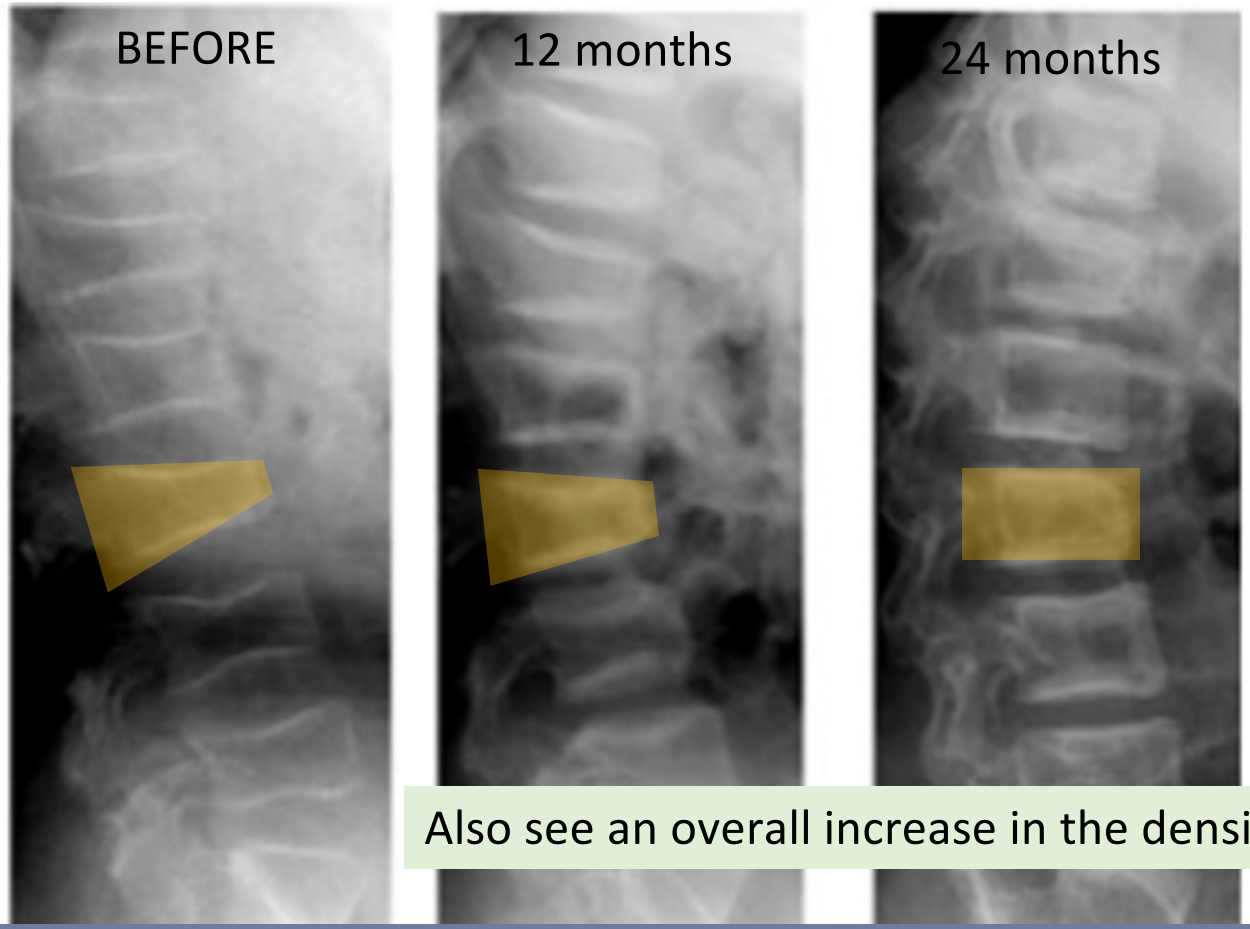
Get ready checklist:

- ❑ Meet calcium RDA for age, plus a supplement around the infusion.
  (500 mg 3x per day, taken 1 week +/- infusion)
- ❑ Vitamin D above 30 ng/mL
- ❑ Check for healthy kidneys
- ❑ Send the dentist an FYI

  Planned procedures that expose the jaw bone (dental extractions, implants, etc) should be done prior and heal fully before treatment.

Vertebral compression fractures before and after IV bisphosphonates

15-year-old patient with DMD



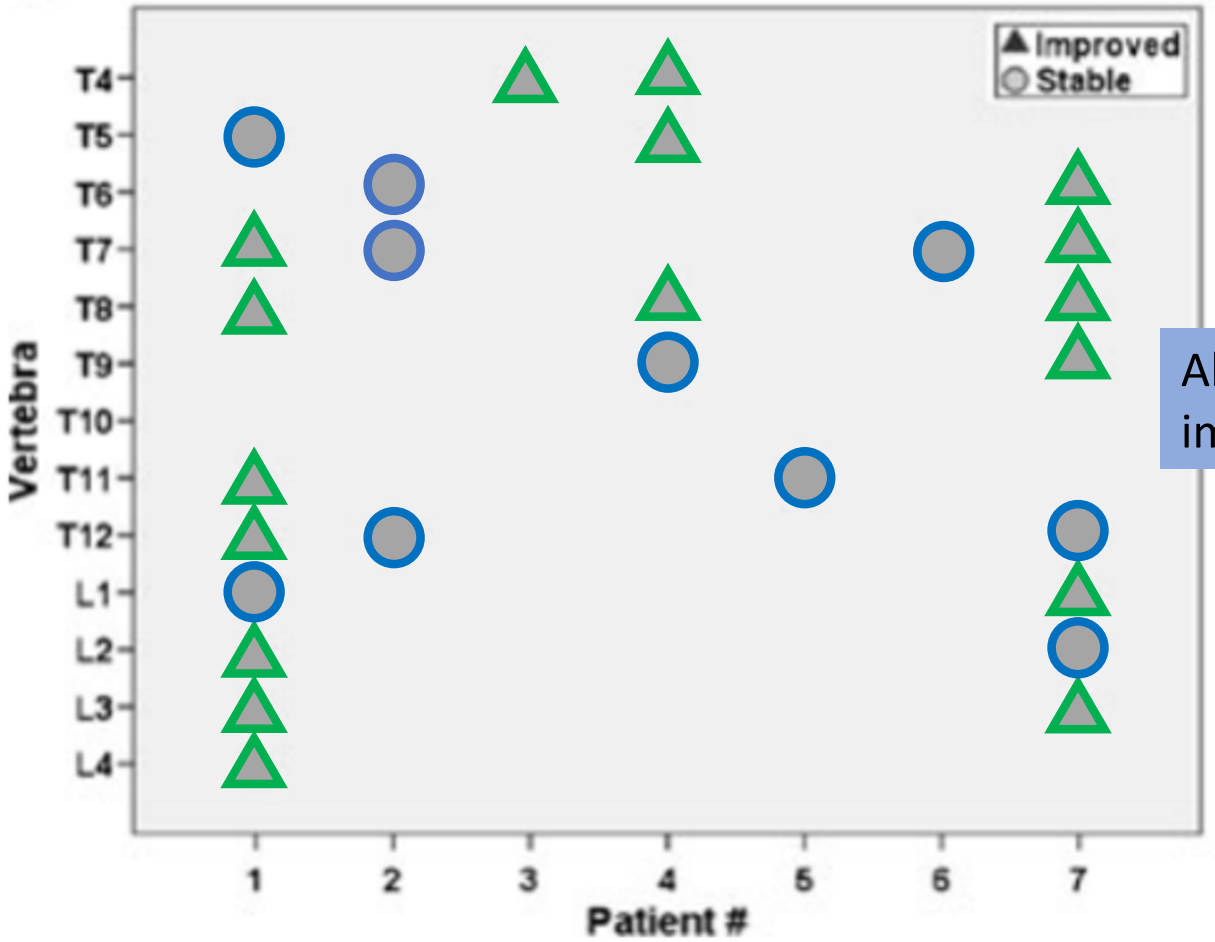


Dr. Leanne Ward, Professor of Pediatrics at the University of Ottawa

Misof et al. J Bone Miner Res. 2016.

Also see an overall increase in the density of the vertebral endplates

Change in Vertebral Height after 12 months of IV Bisphosphonate



All 7 patients were stable or improved

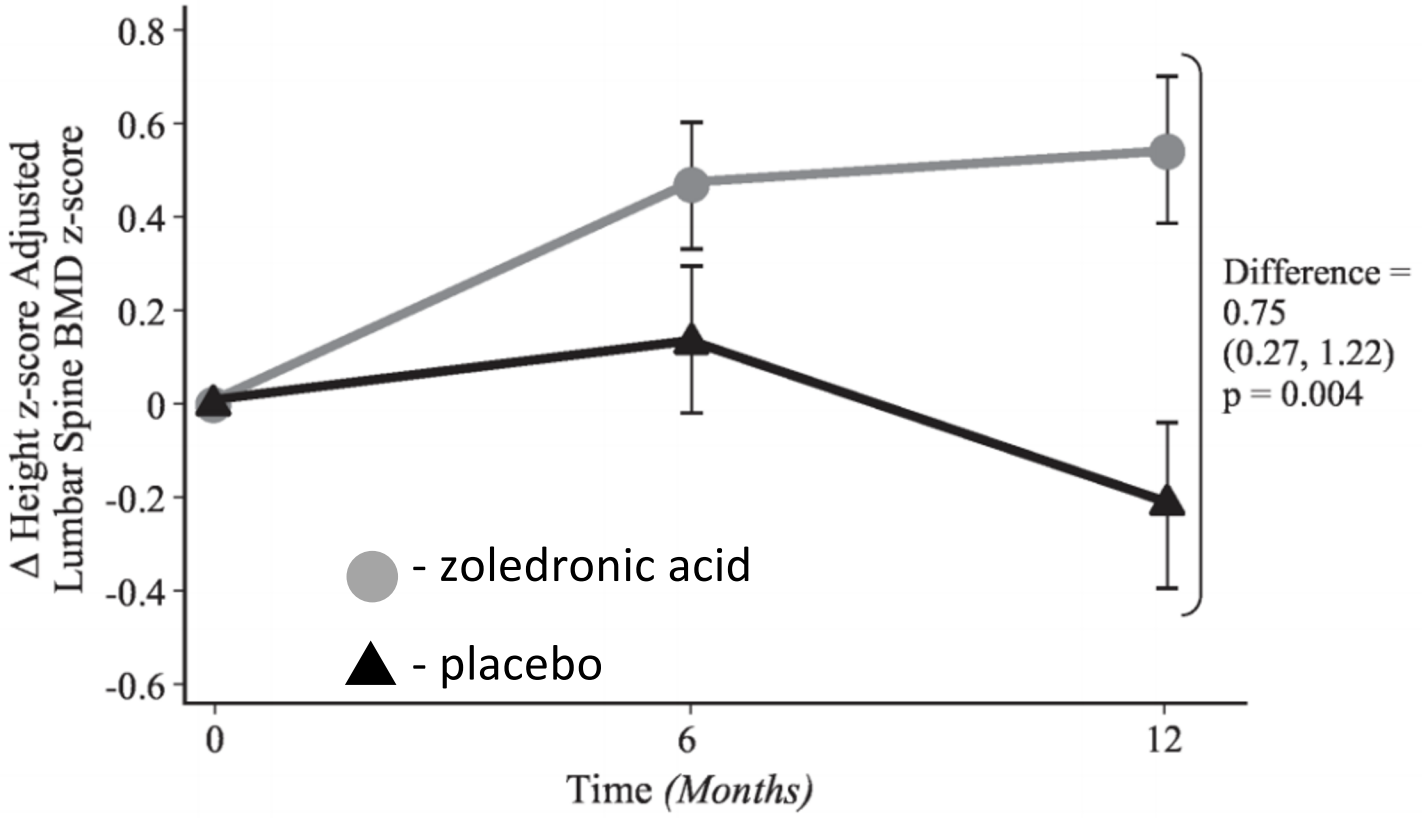
- 3 stable

- 4 improved

Misof et al. J Bone Miner Res. 2016.

Zoledronic acid versus placebo in pediatric glucocorticoid-induced osteoporosis: A randomized double-blind phase 3 trial

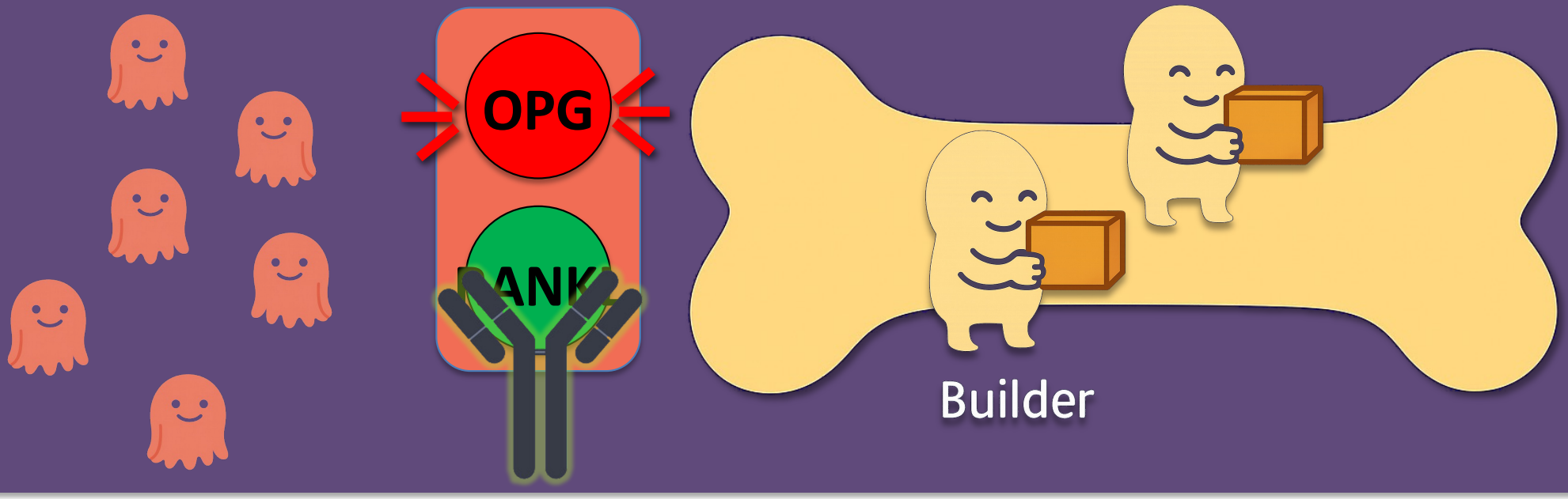




Ward et al. J Clin Endocrinol Metab. July 2021.

What about Denosumab?

Denosumab also targets the **osteoclast**, but in a **different way than bisphosphonates**:
Denosumab is a monoclonal antibody that **binds to RANKL and shuts off** the "go" signal for osteoclasts.



Bone Building Options

PTH Analogues

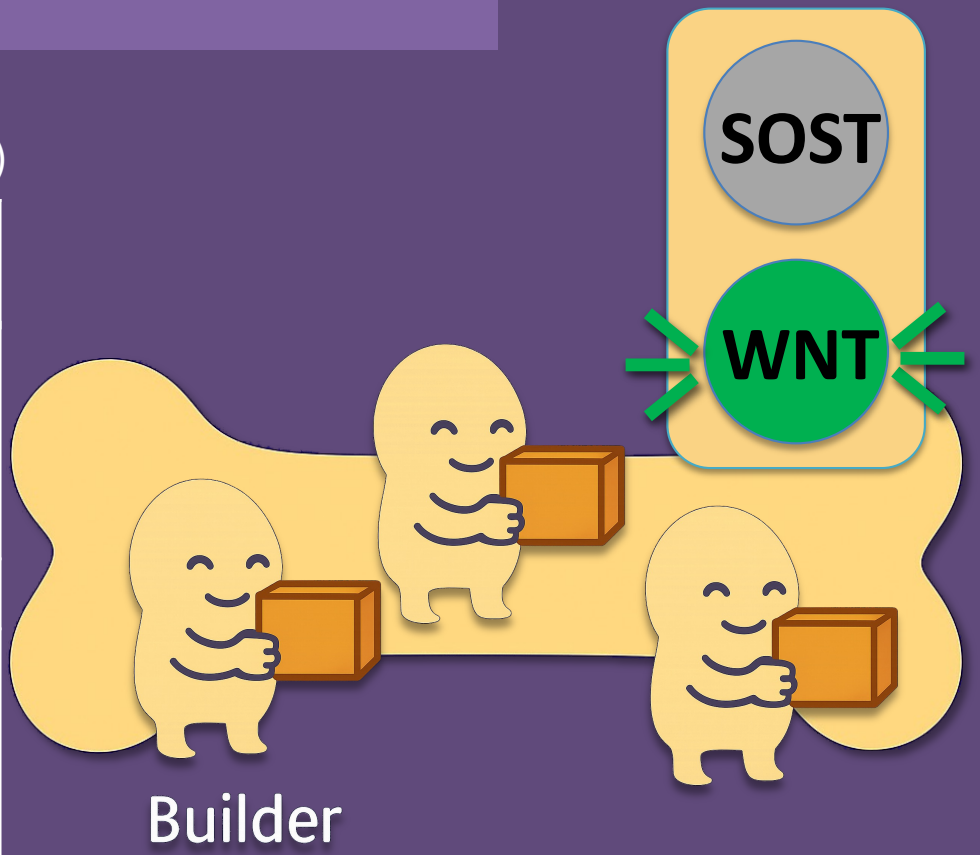
(Teriparatide (Forteo, Bonsity) and Abaloparatide (Tymlos))

What is it?
Recombinant human parathyroid hormone

- **Intermittent** (daily injection) stimulates **osteoblast activity more than osteoclast activity**.
- This leads to **net bone formation** (unlike continuous high PTH, which would cause bone loss).

Mechanism:

- Binds to **PTH receptors** on osteoblasts → increases their number and activity.
- Stimulates **WNT signaling** and decreases **SOST**
- Use for 2 years, then lock in gains with bisphosphonates.



Bone Building Options

Romosozumab

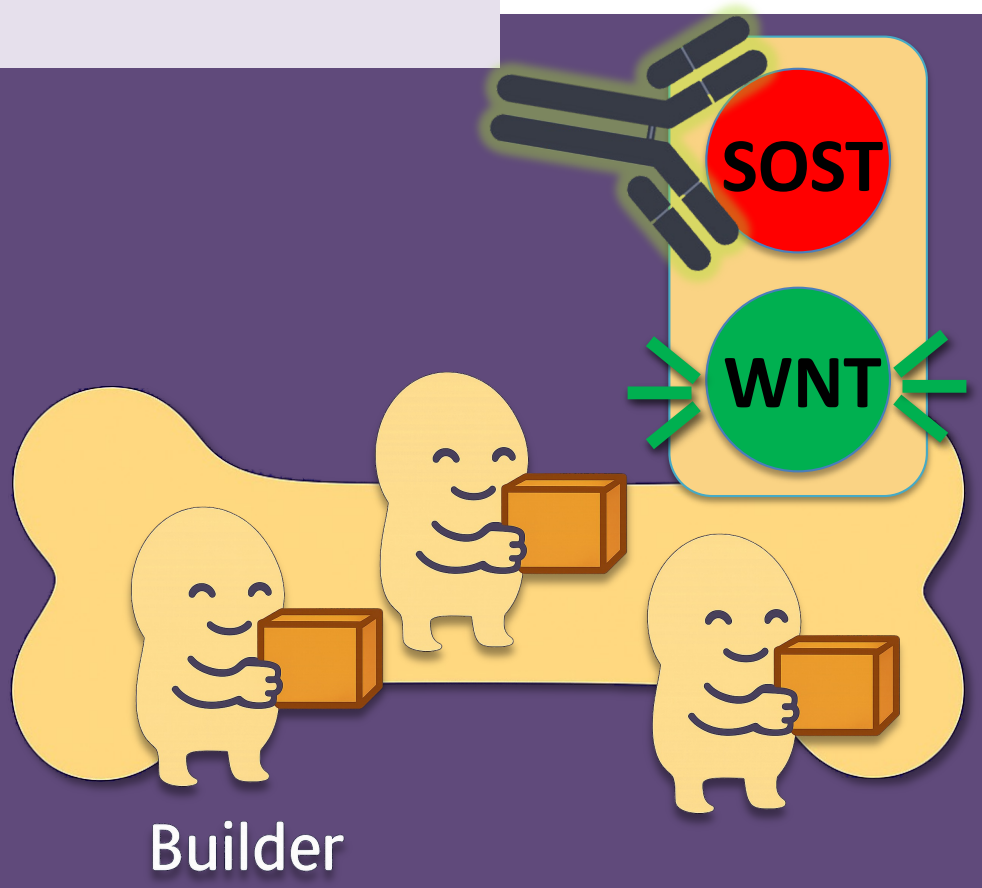
(Evenity)

What is it?
Humanized monoclonal antibody against sclerostin

- Monthly subcutaneous injection
- **Increases bone formation**

Mechanism:

- **Binds the osteoblast stop signal, SOST, to ↑ WNT**
- Not approved in kids. Clinical trials underway.
- Avoid in patients with heart attack / stroke in the past year (small increase in one study: 2.5% vs 1.9%).
- Promotes bone building for 1 year, then lock in gains with bisphosphonates.



Summary of Bone Medication Options

Medication	Dosing	Considerations
Oral bisphosphonate [1] (Alendronate)	Weekly Pill	- Must take on empty stomach, with 8 oz water, and sit upright for 30 minutes - Dosing: 35 mg weekly (child), 70 mg weekly (adult)
IV bisphosphonate [2] (Zoledronic Acid)	Infusion every 6 months (child) every 12 months (adult)	- First dose can cause fatigue/fever - Vitamin D level > 30 ng/dL and recommend taking calcium with meals 1-week prior and 1-week after the infusion - Check for healthy kidneys - Dosing: 0.0125 mg/kg (initial), followed by 0.025 mg/kg every 6 months or 0.05 mg/kg annually (max 5 mg/year)
Denosumab [3] (anti-RANKL antibody)	Injection every 6 months	- Can have rebound BMD loss & severe rebound hypercalcemia
Romosozumab (anti-sclerostin antibody)	Monthly Injection	- 1 year then plan to follow up with bisphosphonates - Risk of heart attack, stroke in older adults
Teriparatide [4] (PTH analogue)	Daily Injection	- 2 years then plan to follow up with bisphosphonates

References:

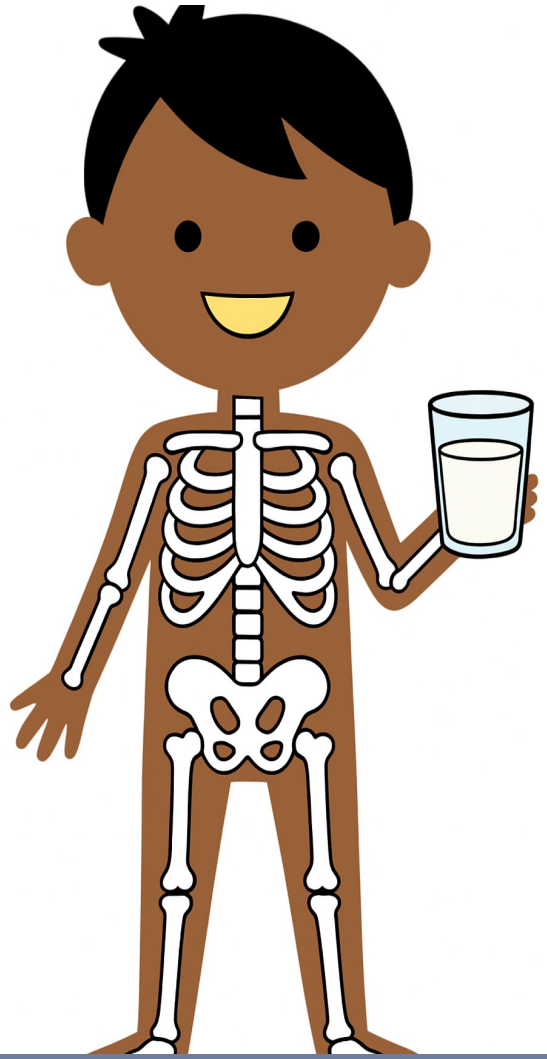
[1] C. Tian et al., “Oral bisphosphonate treatment in patients with Duchenne muscular dystrophy on long term glucocorticoid therapy,” Neuromuscul. Disord. NMD, vol. 30, no. 7, pp. 599–610, Jul. 2020, doi: 10.1016/j.nmd.2020.06.005.

[2] L. M. Ward, K. Kinnett, L. Bonewald, and Parent Project Muscular Dystrophy Bone Health Workshop Collaborators, “Proceedings of a Parent Project Muscular Dystrophy Bone Health Workshop: Morbidity due to osteoporosis in DMD: The Path Forward May 12-13, 2016, Bethesda, Maryland, USA,” Neuromuscul. Disord. NMD, vol. 28, no. 1, pp. 64–76, Jan. 2018, doi: 10.1016/j.nmd.2017.05.012.

[3] C. Hung, K. D. Mathews, and A. Shibli-Rahhal, “Effect of Denosumab on Bone Health in Adult Patients with Duchenne/Becker Muscular Dystrophy: A Report of 2 Cases,” JBJS Case Connect., vol. 12, no. 1, Mar. 2022, doi: 10.2106/JBJS.CC.21.00664.

[4] N. Nasomyont et al., “Safety and efficacy of teriparatide treatment for severe osteoporosis in patients with Duchenne muscular dystrophy,” Osteoporos. Int. J. Establ. Result Coop. Eur. Found. Osteoporos. Natl. Osteoporos. Found. USA, vol. 31, no. 12, pp. 2449–2459, Dec. 2020, doi: 10.1007/s00198-020-05549-z.

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Development
(puberty & growth)

AAP Obesity Clinical Practice Guidelines (Jan 2023)

Comprehensive Treatment	Overweight		Obesity	
	<12 y	≥12 y	<12 y	≥12 y
Motivational Interviewing (KAS 10)	✅	✅	✅	✅
Intensive Health Behavior and Lifestyle Treatment (KAS 11)	✅	✅	✅	✅
Weight loss Pharmacotherapy (KAS 12)				✅

Pharmacotherapy for children ≥ 12 years of age:

- Phentermine / Topomax
- GLP-1 agonist weekly
  - **Muscle loss & Constipation are concerns**
  - **Clinical nutrition for protein intake**
  - **Bowel regimen**

Worksheet for Weight Management

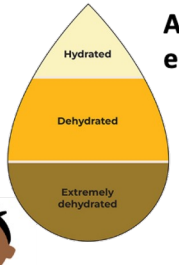
APPETITE ☐ Yes ☐ No

Are you eating when you are hungry?

Do you also eat when you are emotional? Bored? Or just because others are eating?

Pay attention to your hunger cues and *try* to eat based on hunger.

WATER ☐ Yes ☐ No



Are you drinking enough water?





Estimate to drink ½ oz per pound you weigh up to 64 oz (~2L).

SLEEP

☐ Yes ☐ No

Are you getting good sleep?

Sleep affects appetite, so please tell your Neurologist if you have concerns about sleep.

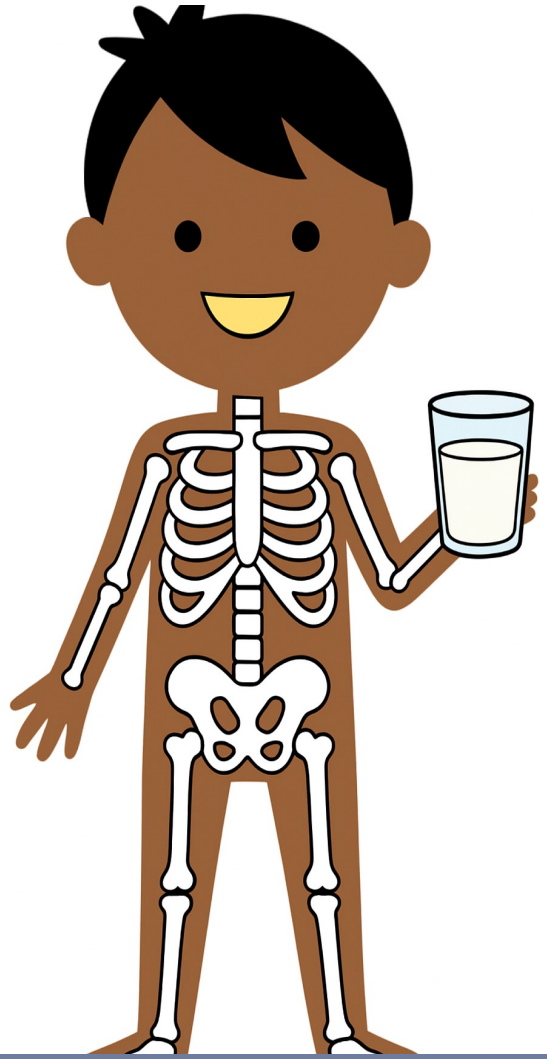


NUTRITION

☐ Yes ☐ No



ENDOCRINE HEALTH PLAN in DMD:



Adrenal
(secondary AI)

Bone
(osteoporosis & fracture)

Carbs
(steroid-induced insulin resistance)

Development
(puberty & growth)

2018 Consensus Guidelines

THE LANCET Neurology

Growth Guidelines

Assessment of height every 6 months until completion of puberty and attainment of final height

→

Impaired growth
- Crossing of height percentile
- Height velocity <4 cm/year
- Height <3rd percentile

→

Recommended
- Bone age x-ray
- Thyroid function tests
- Celiac panel
- Growth factors
- Comprehensive metabolic panel
- Complete blood count

To be considered
Growth hormone stimulation testing



Few data show the safety and efficacy of growth hormone in the DMD population.

PROS

- One retrospective study found short-term benefit on height velocity

"Until more evidence is available, routine use of growth hormone to treat DMD-related growth failure is **not** recommended."

CONS

- Worsening glucose intolerance
- Progression of scoliosis.[99]
- Taller stature might be detrimental to mechanical function in DMD.[100,101]
- No studies have followed patients to their final adult height
- No studies large enough to establish whether GH has a negative effect on muscle function

2018 Consensus Guidelines

THE LANCET Neurology

Puberty Guidelines

Physical assessment of pubertal status by Tanner staging every 6 months starting by age 9 year

→

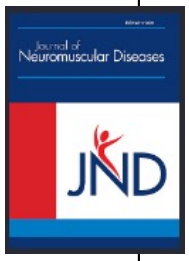
Delayed puberty
Testicular volume < 4 mL at 14 years or older

→

Recommended Laboratory assays:
- Luteinizing hormone
- Follicle-stimulating hormone
- Testosterone
- Bone age x-ray

Treatment for confirmed hypogonadism:
- T at low doses → gradually increase
- Recommended for age >13 years; can be considered from age 12 years

Strongly consider 13 years old



Check for updates

Managing growth and puberty concerns in individuals with Duchenne muscular dystrophy: Updated guidance and future directions

Journal of Neuromuscular Diseases
2026, Vol. 0(0) 1–30
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Meilan M. Rutter, MBBCh, FRACP[1,2], Janet L. Crane, MD[3,4], Funmbi Babalola, MD, MSc, FRCPC[5], Robert W. Benjamin, MD[6], Carol Lam, MD, MSc[7,8], Laura McAdam, MD, FRCPC[9], Stefan Nicolau, MD[10,11], Anne Marie Sbrocchi, MD, FRCPC[12], Mena Scavina, DO[13], Julia Sorbara, MD, MSc, FRCPC[7,8], Alexandra Ahmet, MD[14], Susan Apkon, MD[15], Chiara Arpaia, MD[16,17], Tommaso Aversa, MD, PhD[18], Shipra Bansal, MBBS[19], Luca Bello, MD, PhD[20], Diana X. Bharucha-Goebel, MD[10,11], Sabrina Corbetta, MD, PhD[21,22], Natascia Di Iorgi, MD, PhD[23,24], Ryan Farrell, MD[25], Alberto Ferlin, MD, PhD[26,27], Danilo Fintini, MD[28], Melissa Fiscaletti, MD, MSc, FRCPC[29], Rana Halloun, MD[30], Maria-Elena Lautatzis, MD, MSc, FRCPC[31], Hugh J. McMillan, MD, MSc[32], Nadia Merchant, MD[33], Patrick Moeschen[13], Nat Nasomyont, MD, MS[1,2], Maria Fernanda Ochoa Molina, MD[34], Chiara Panicucci, MD, PhD[35], Nora E. Renthal, MD, PhD[36], Luisa De Sanctis, MD, PhD[37,38], Jaclyn Tamaroff, MD, MSCI[39], Cuixia Tian, MD[40], Aravindhan Veerapandiyan, MD[41], Leanne M. Ward, MD FRCPC[14,30], David R. Weber, MD, MSCE[42], Sze Choong Wong, MRCPCH, DMedSci[43,44], Ilaria Zito, PhD[45], and Claire L. Wood, MRCPCH, BMedSci, PhD[46,47]

Abstract

Long-term glucocorticoid therapy is standard of care for individuals with Duchenne muscular dystrophy (DMD) but often results in impaired growth and delayed or incomplete puberty, impacting their physical and psychosocial health. Although

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Pubertal Induction as part of Adolescent Sexual Healthcare

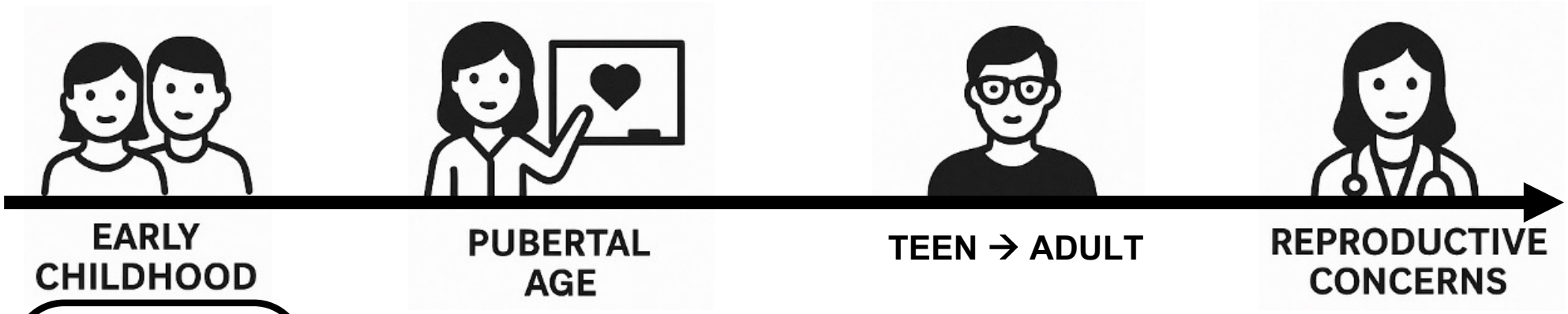
When discussing testosterone therapy for **puberty induction** in DMD, several topics related to **sexuality and fertility** should be addressed to ensure informed decision-making, support, and holistic care.



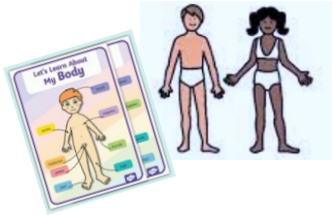
Pubertal Induction as part of Adolescent Sexual Healthcare

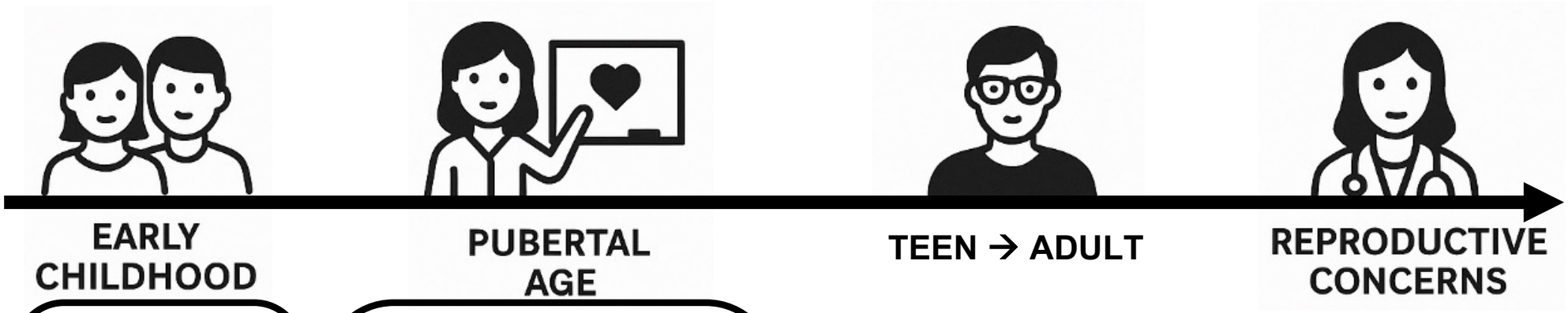


1. Pubertal Development Goals
2. Fertility Implications
3. Psychosocial-Emotional Considerations
4. Sexual Education & Support Systems

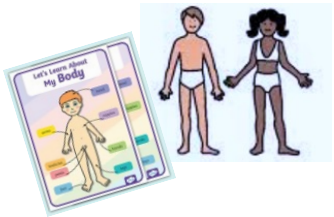


Parents/guardians talk about privacy, body part names, and body safety rules.





Parents/guardians talk about privacy, body part names, and body safety rules.



- **School**-based sex education
- **Peer**-shared "knowledge"
- **Family** shares values, normalizes topic



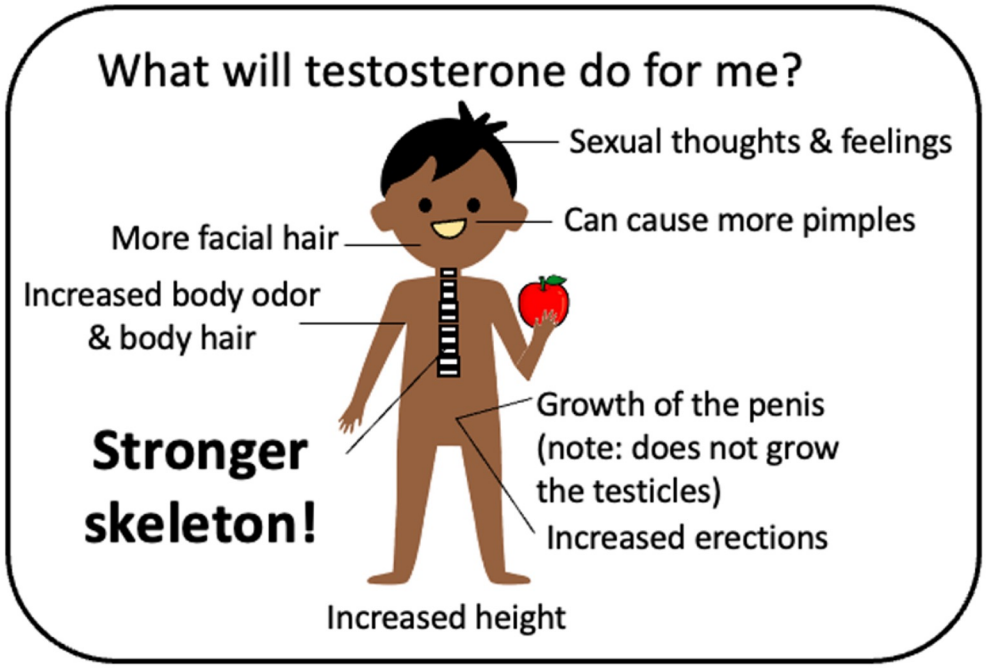
- **Physician** discusses puberty

→With delayed puberty, physician assesses gender goals & discusses fertility prior to T.

1. Pubertal Development Goals

Discuss Expected / Desired Physical Changes

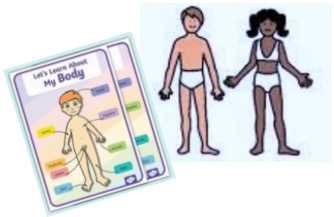
- Discuss what testosterone therapy will achieve, such as increased deepened voice, growth of body and facial hair, and genital maturation.
- Listen to the patient's goals for puberty.
- Normalize that puberty is a time of new sexual thoughts and feelings.
- Normalize that it's OK to talk to the medical team about these topics.





EARLY CHILDHOOD

Parents/guardians talk about privacy, body part names, and body safety rules.



PUBERTAL AGE

- **School**-based sex education
- **Peer**-shared "knowledge"
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- **Physician** discusses puberty

→With delayed puberty, physician assesses gender goals & discusses fertility prior to T.

TEEN → ADULT

Standard Care:

- STI screening, HPV vaccine
- Condoms, Abuse screening

DMD-Specific Add-ons:

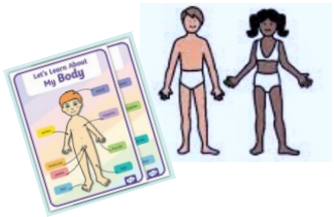
- Sexual activity check-in (e.g., positioning, heart health)
- Psychological response to T.
- Body image & isolation screening
- Internet safety support

REPRODUCTIVE CONCERNS



EARLY CHILDHOOD

Parents/guardians talk about privacy, body part names, and body safety rules.



PUBERTAL AGE

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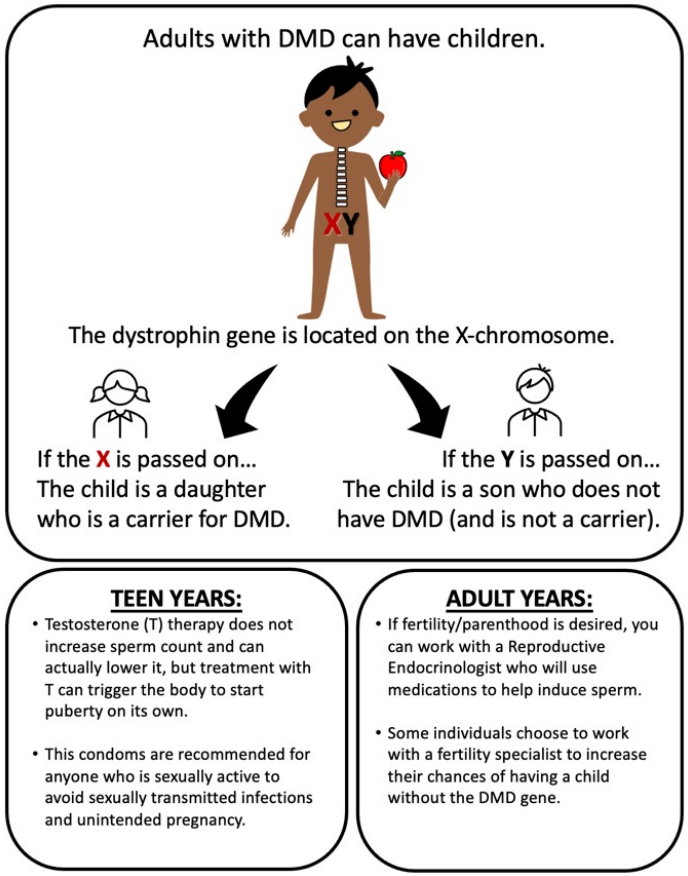
- Sexual activity check-in (e.g., positioning, heart health)
- Psychological response to T.
- Body image & isolation screening
- Internet safety support

REPRODUCTIVE CONCERNS

Fertility Desired?
Refer to reproductive endocrinology for parenthood planning

MD will tailor guidance based on pubertal history & current testosterone use

Fertility Implications

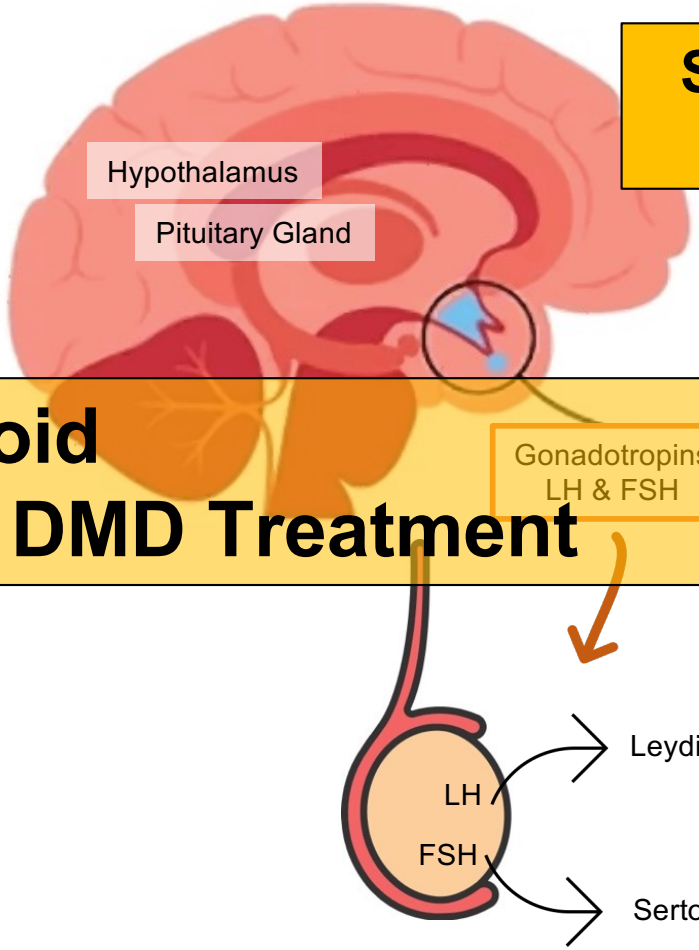


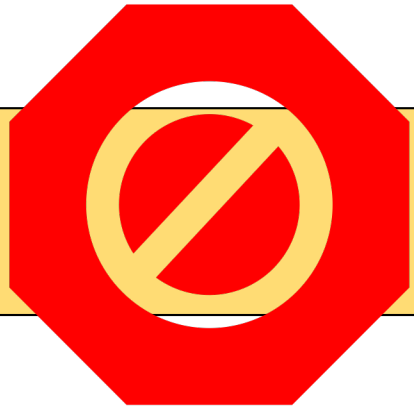
Discussion of Fertility and Parenthood

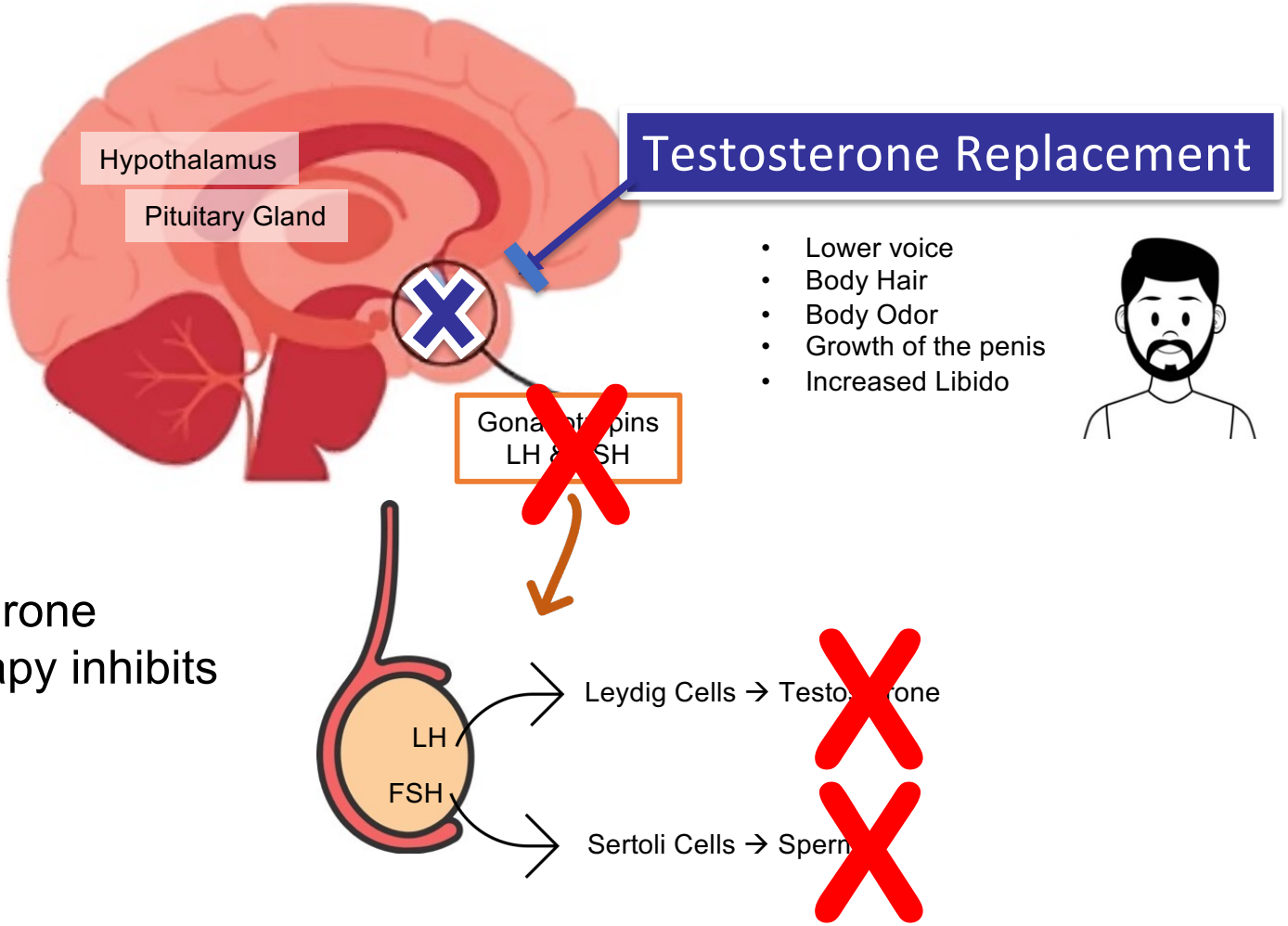
- Explain that T supports secondary sexual characteristics, it does not directly induce spermatogenesis or guarantee fertility.
- Some patients experience endogenous puberty with monthly testosterone.
    - Use this as a chance to say, that in sexually active teens a condom is always recommended.
- Long-term testosterone may suppress spermatogenesis.
    - Assisted reproductive interventions may help restore spermatogenesis after long-term testosterone use.

Glucocorticoid Steroids for DMD Treatment

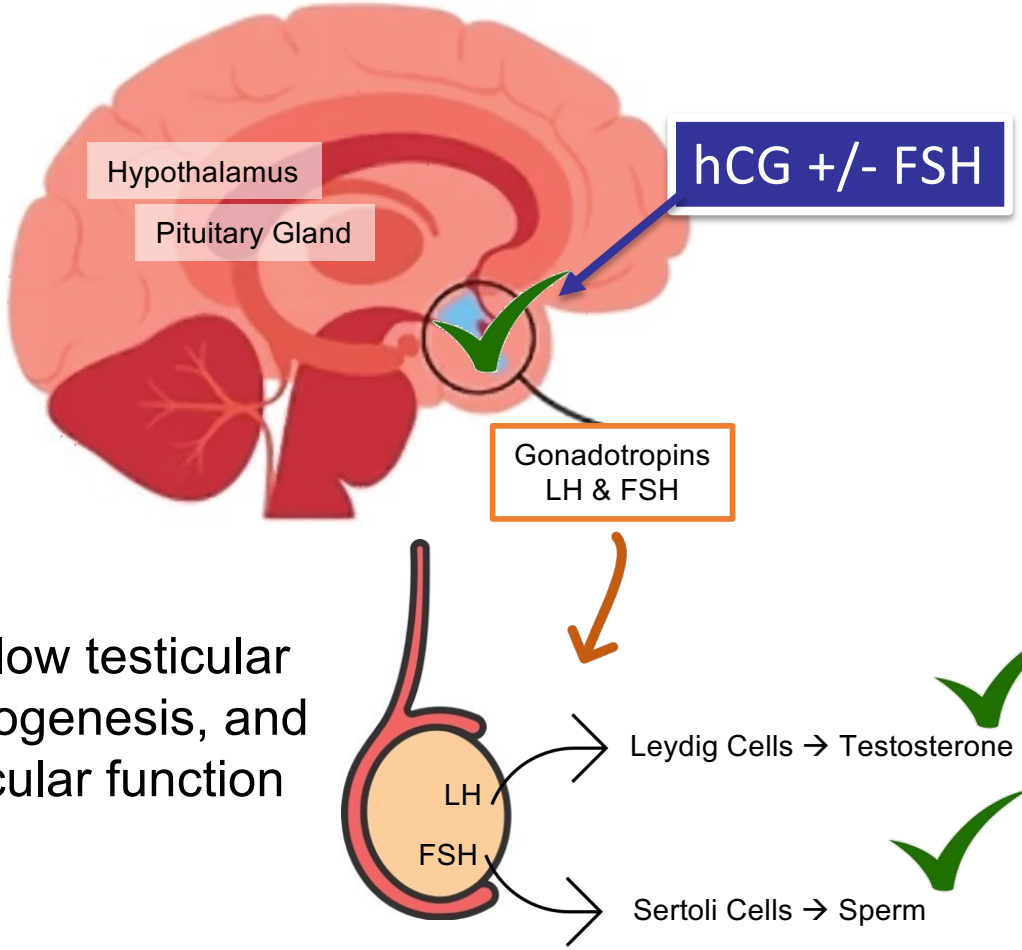
Secondary hypogonadotropic hypogonadism



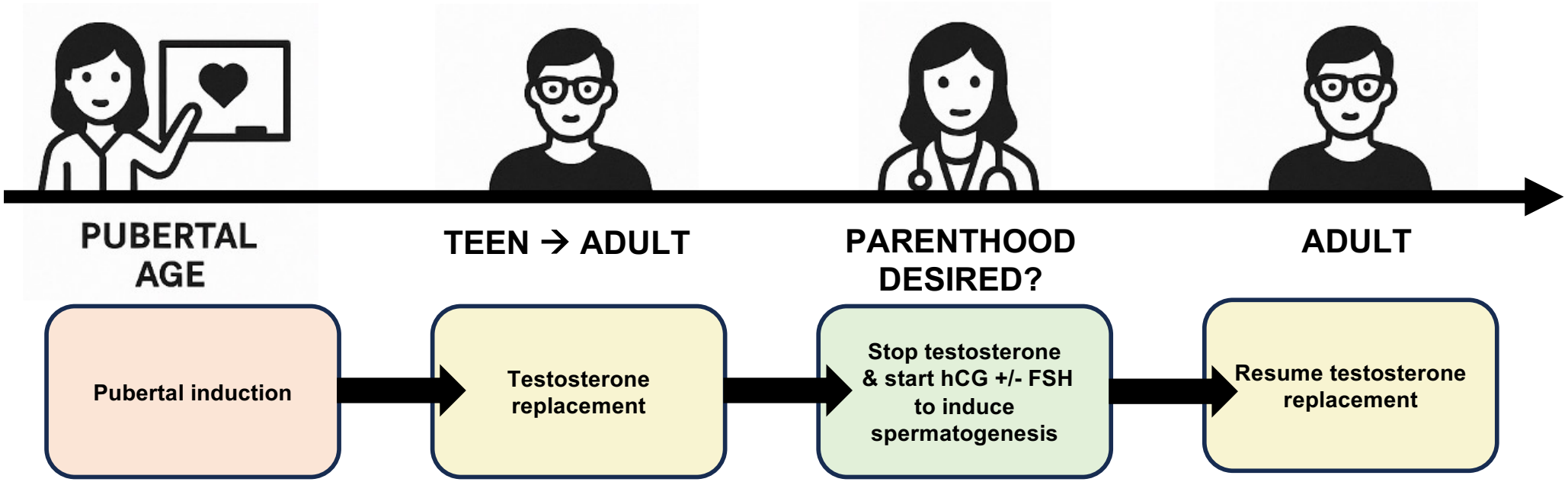




Long-term Testosterone Replacement Therapy inhibits testicular growth & spermatogenesis



hCG +/- FSH allow testicular growth, spermatogenesis, and endocrine testicular function


PUBERTAL AGE
TEEN → ADULT
PARENTHOOD DESIRED?
ADULT
Pubertal induction
Testosterone replacement
Stop testosterone & start hCG +/- FSH to induce spermatogenesis
Resume testosterone replacement

What is happening from the individual's perspective? What are the lived experiences of sexual healthcare and sexual education among youth with disabilities?





Interviews of Young Adults with Muscular Dystrophy

MUSCLE &NERVE

CLINICAL RESEARCH ARTICLE | Open Access

Sexual health and fertility in Duchenne muscular dystrophy—An exploratory study

Janet Hoskin PhD, Timothy D Cheetham MD, Rod T Mitchell PhD, Sze Choong Wong MD, Claire L Wood PhD

First published: 11 July 2024 | https://doi.org/10.1002/mus.28201 | Citations: 2

- 6 men in the UK with DMD aged 24 – 48 years
- 2 online focus groups
- Opinions on sexual health, fertility and testosterone treatment
- Constructivist methodology and Thematic Analysis (Braun and Clarke 2006).

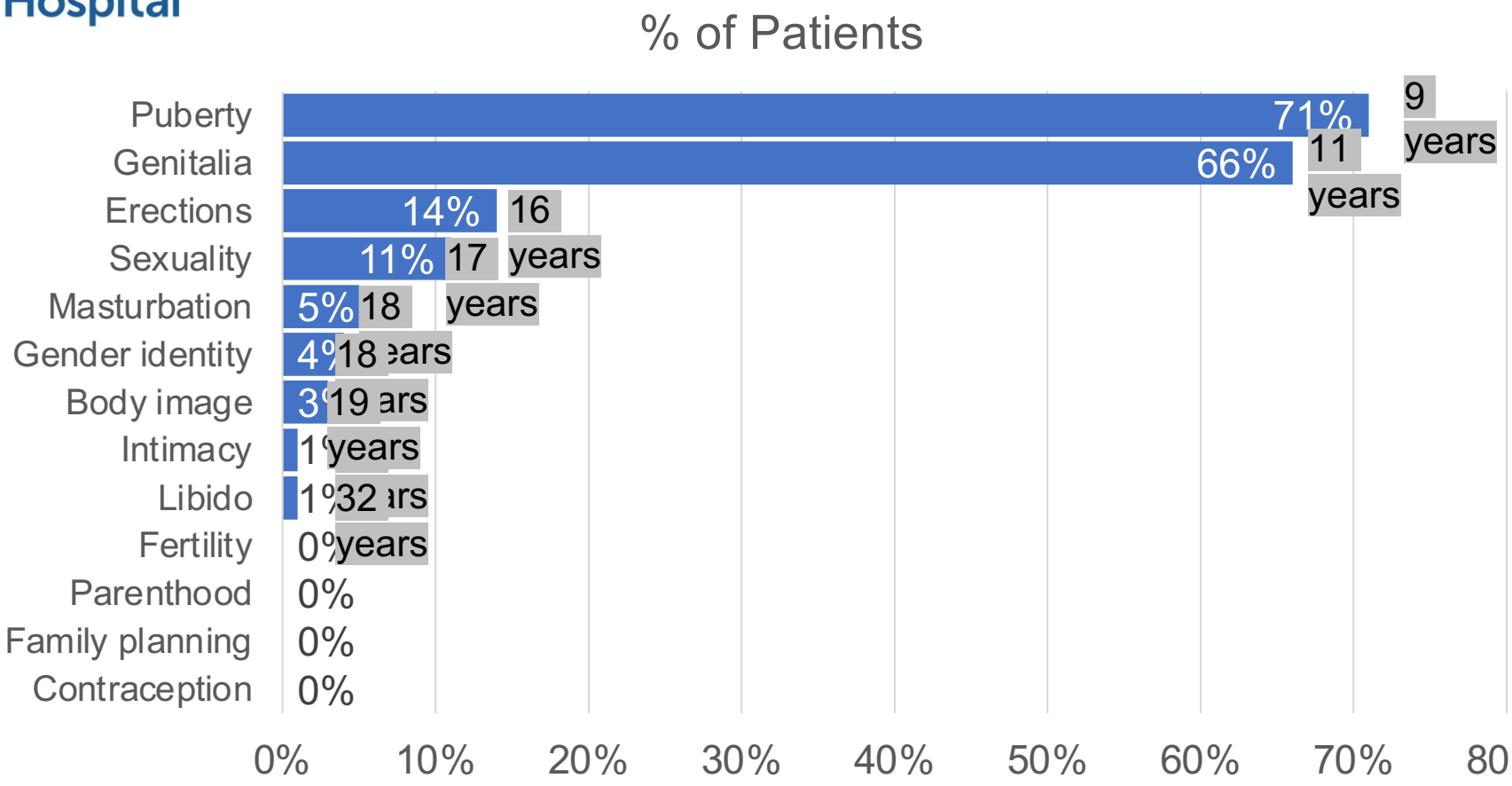
5 key themes

- **Need for info/guidance**
- **Fear of rejection**
- **Physical barriers**
- **Testosterone treatment**
- **Parenthood and fertility**

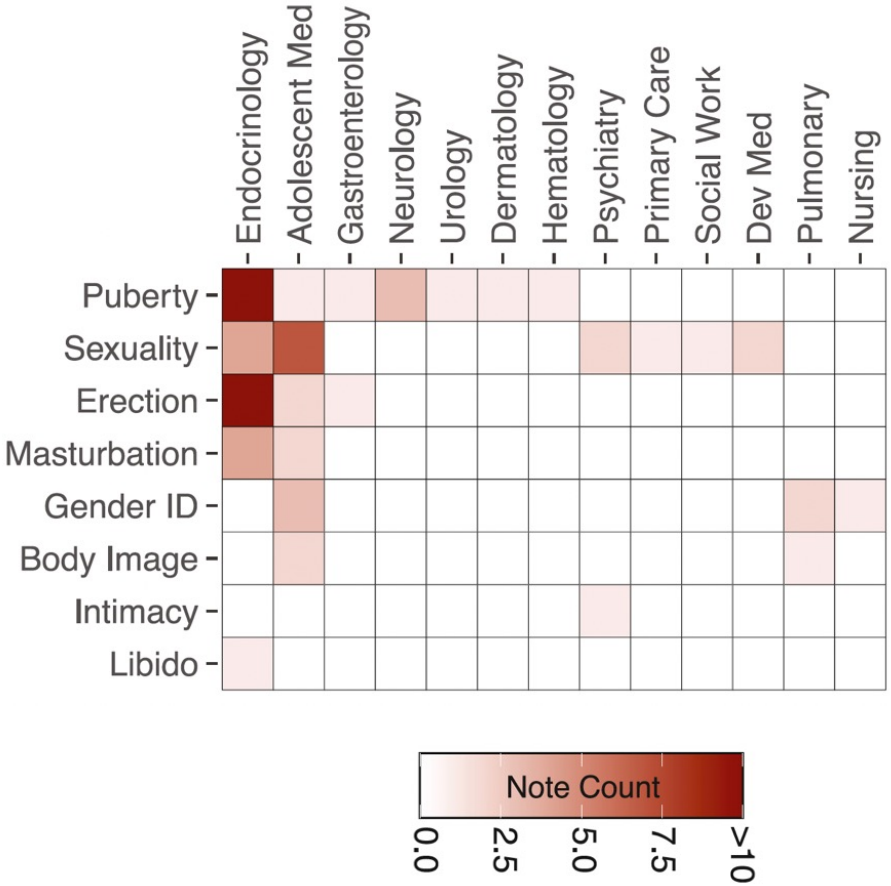
Hoskin et al. Muscle & Nerve 2024.

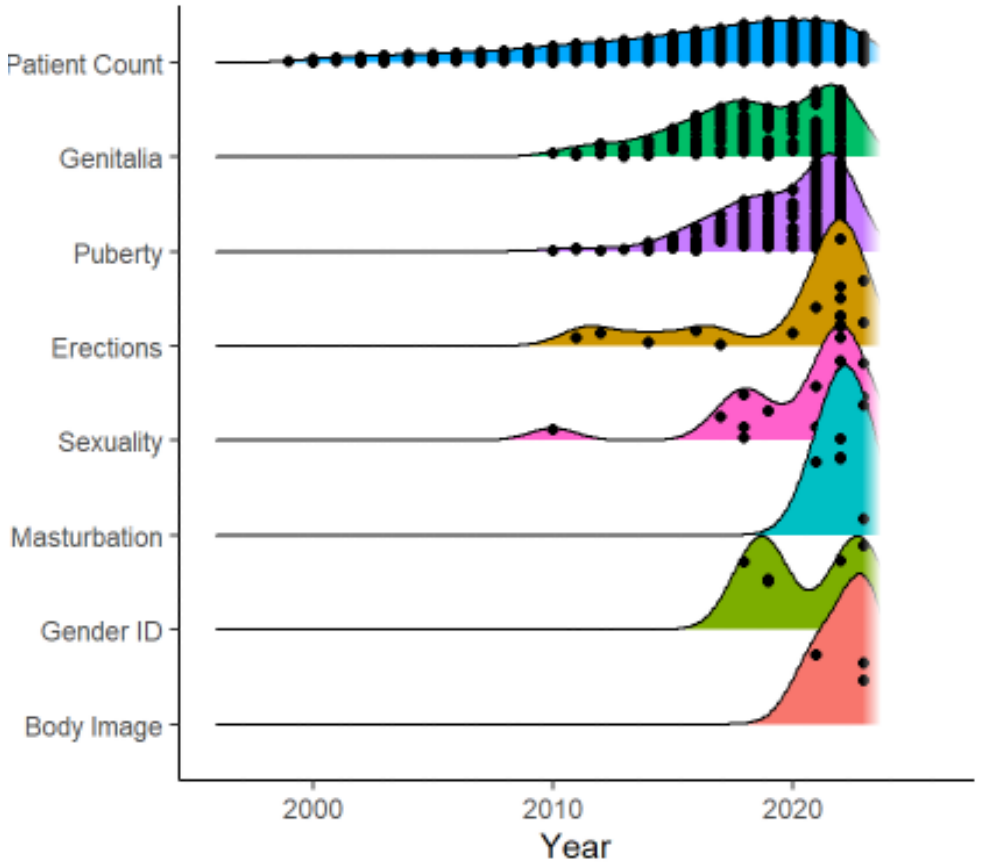
Boston Children's Hospital

Were the topics " _____" documented across any visits?



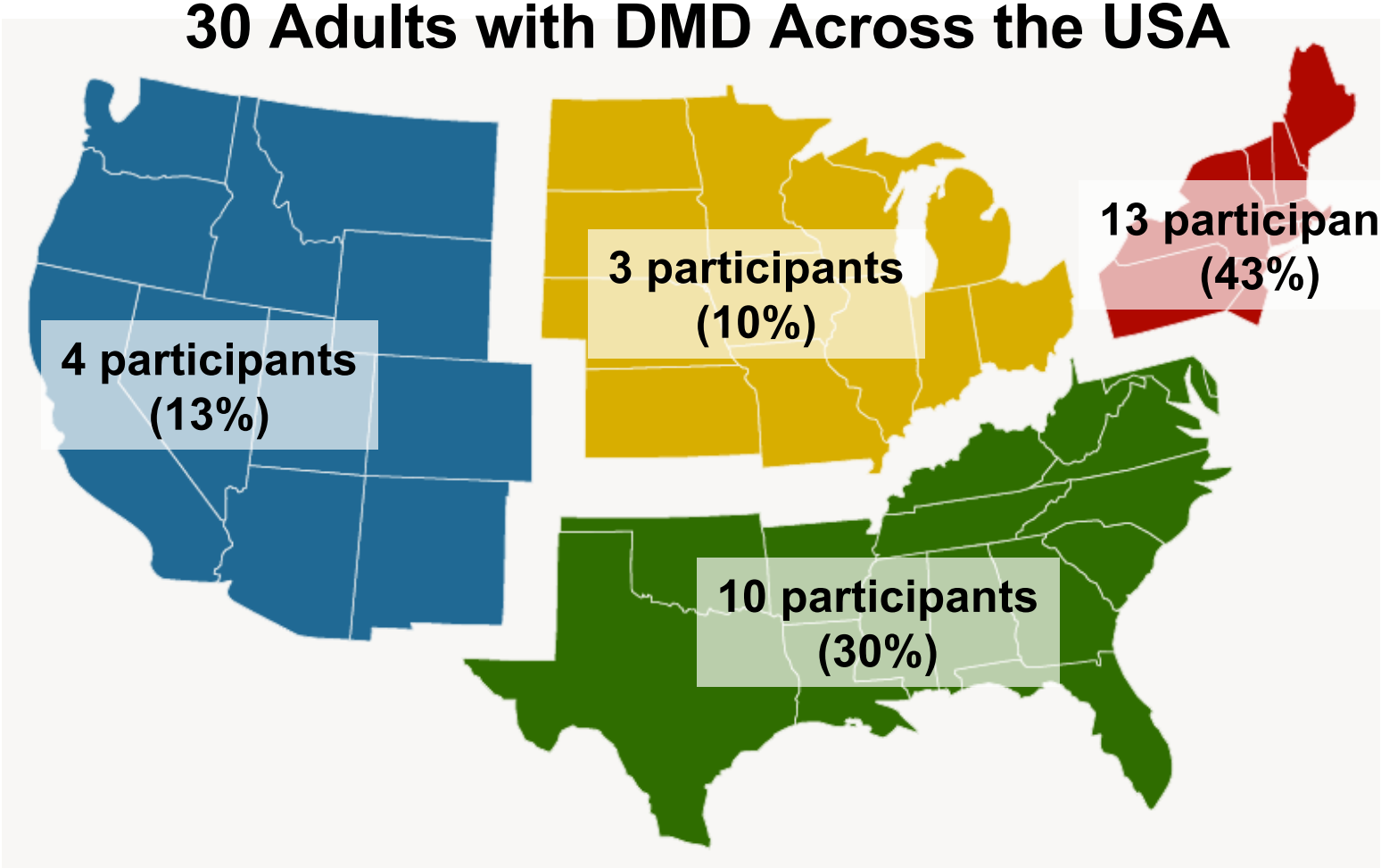
Ellis Barrera, Baronas JM, Sutherland S, Rohan A, Margolin RE, Boskey ER, Renthal NE. Journal of Pediatrics 2024.





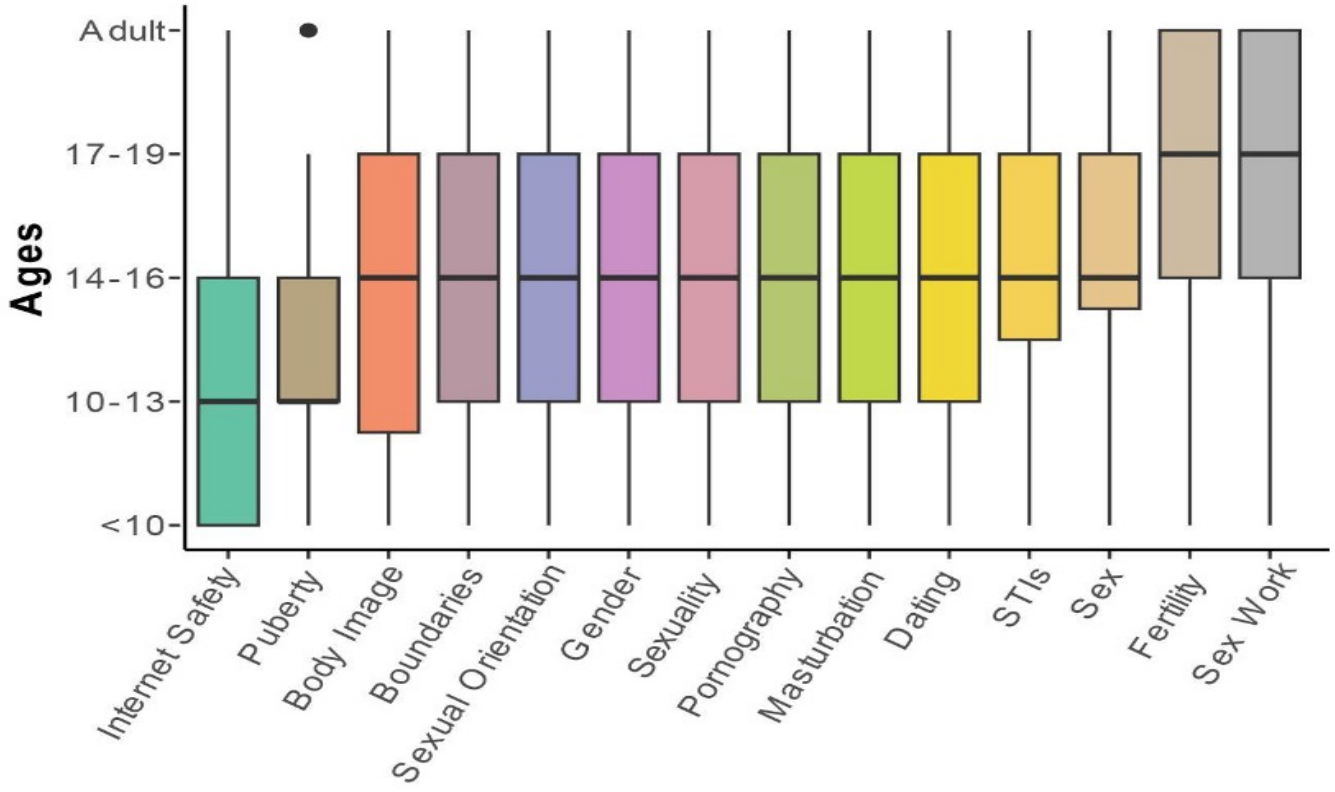
Ellis Barrera, Baronas JM, Sutherland S, Rohan A, Margolin RE, Boskey ER, Renthal NE. Journal of Pediatrics 2024.

30 Adults with DMD Across the USA



Kim J, Rohan A, Margolin RE, Pagan A, Tantivit N, Jama N, Barrera E, Baronas JM, Boskey ER, Renthal NE. J Sex Med. 2025

If it were up to you, what topics would be discussed with youth with DMD & when?



Kim J, Rohan A, Margolin RE, Pagan A, Tantivit N, Jama N, Barrera E, Baronas JM, Boskey ER, Renthal NE. J Sex Med. 2025

Patient Narratives – Interviews of 30 Young Adults with DMD

"I wish doctors had talked about sexual health… but it makes *them* uncomfortable." – age 19

"I would have found it helpful if they [doctors] asked me the questions, but this is stuff they didn't like talk about…" - age 31

"No doctor or person in my life has said, "You can't become a parent; you can't get married," but the amount of people in the world that are married with the same disabilities [as me] is microscopic.

On top of that, what you see on the internet is hate and people being like, "You aren't normal; you aren't pretty; you aren't okay because you're different or disabled." And so, I've never been told directly that I can't do it [have children], but it seems like the world is telling me that I can't do it." – age 19

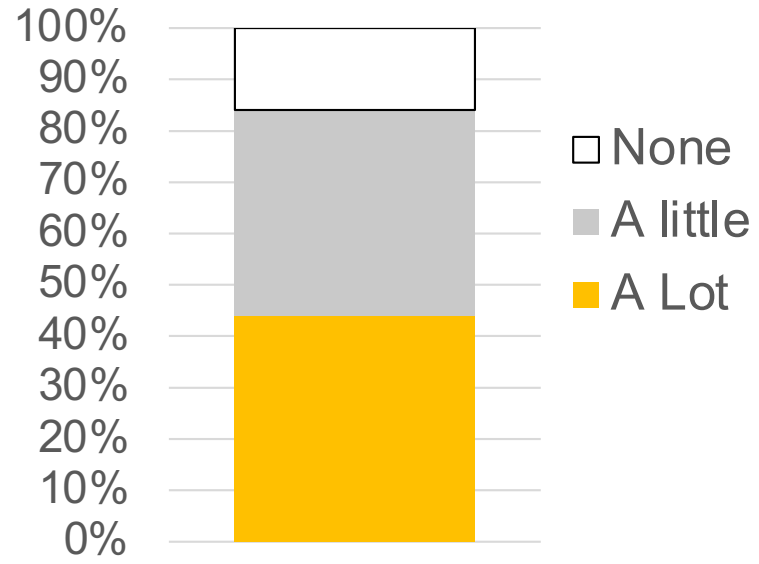
Kim J, Rohan A, Margolin RE, Pagan A, Tantivit N, Jama N, Barrera E, Baronas JM, Boskey ER, Renthal NE. J Sex Med. 2025

Have you ever had conversations with your medical providers about sexual health?		
No	18	60%
Yes, but rarely	8	27%
Yes, many times	1	3%
Non-disclosed	3	10%

Kim J, Rohan A, Margolin RE, Pagan A, Tantivit N, Jama N, Barrera E, Baronas JM, Boskey ER, Renthal NE. J Sex Med. 2025

Fertility and Parenthood

How much have you thought about the possibility of becoming a parent?



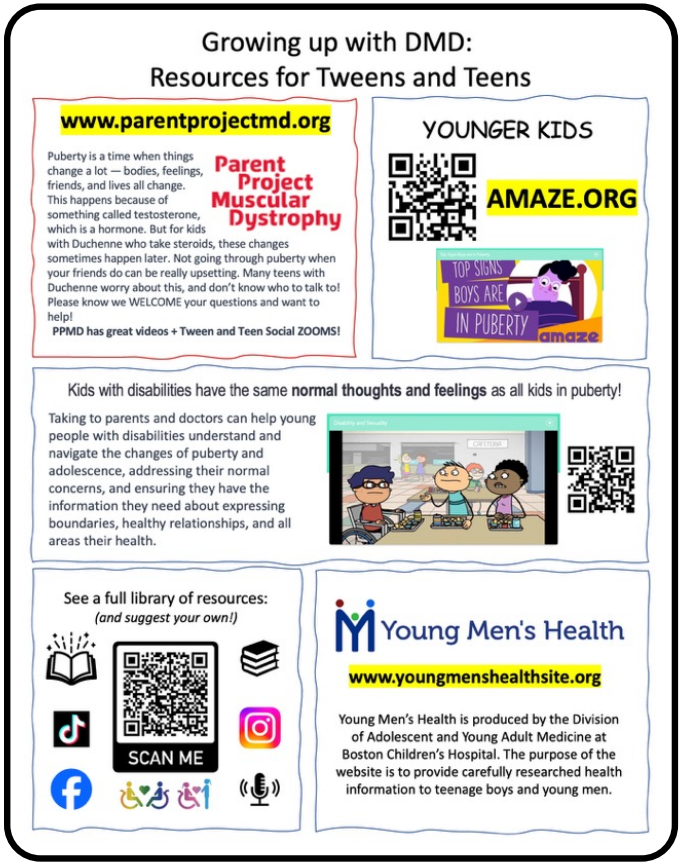
With whom have you discussed the possibility of becoming a parent?	
No one	14
Parents/guardians	11
Romantic partner(s)	8
Friends	6
Doctors/medical providers	3
Non-disclosed	3

Kim J, Rohan A, Margolin RE, Pagan A, Tantivit N, Jama N, Barrera E, Baronas JM, Boskey ER, Renthal NE. J Sex Med. 2025

How can we improve?



Sexual Education & Support Systems



Discussion of Sexual Education & Social Support

Sexual Education

- Assess school curriculum
- Discuss frequency/depth of conversations with parents/caregivers.
- Ask about use of internet/pornography
- Provide resources relevant to your patient population.

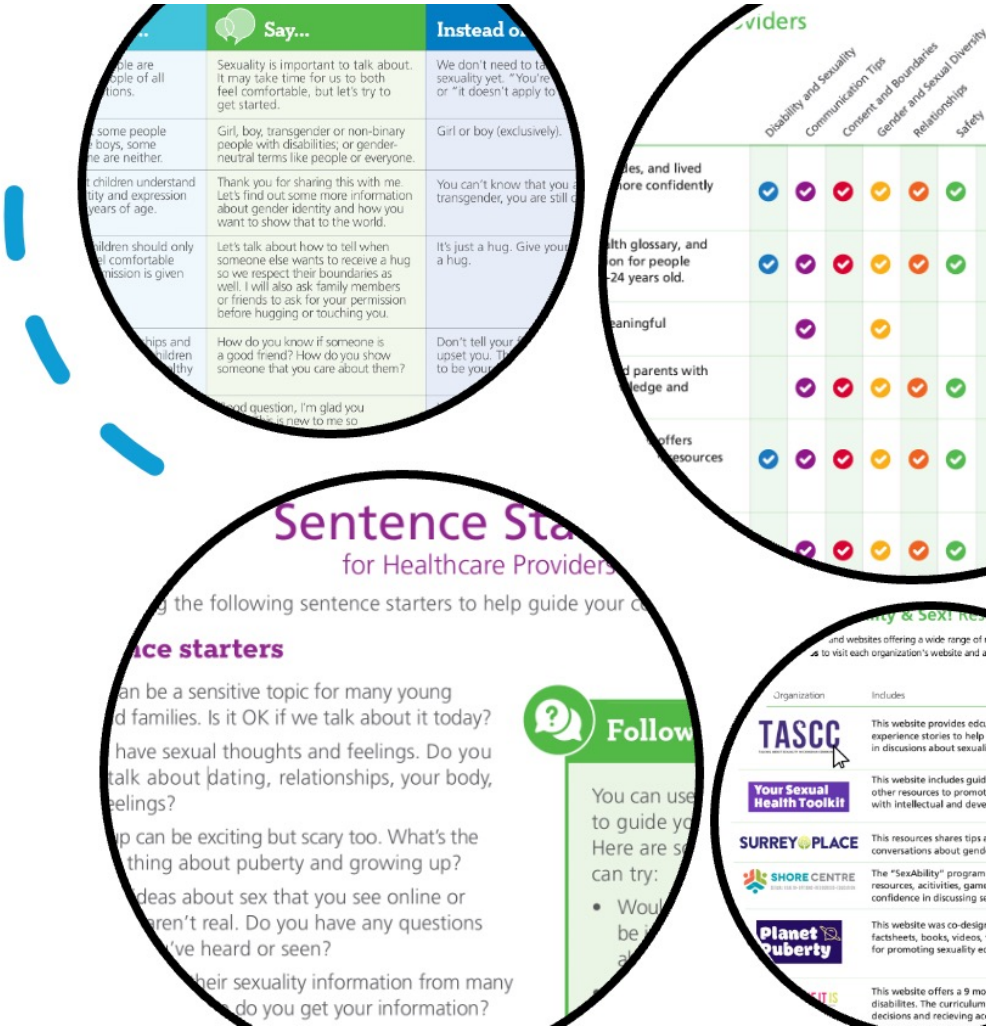
Social Supports

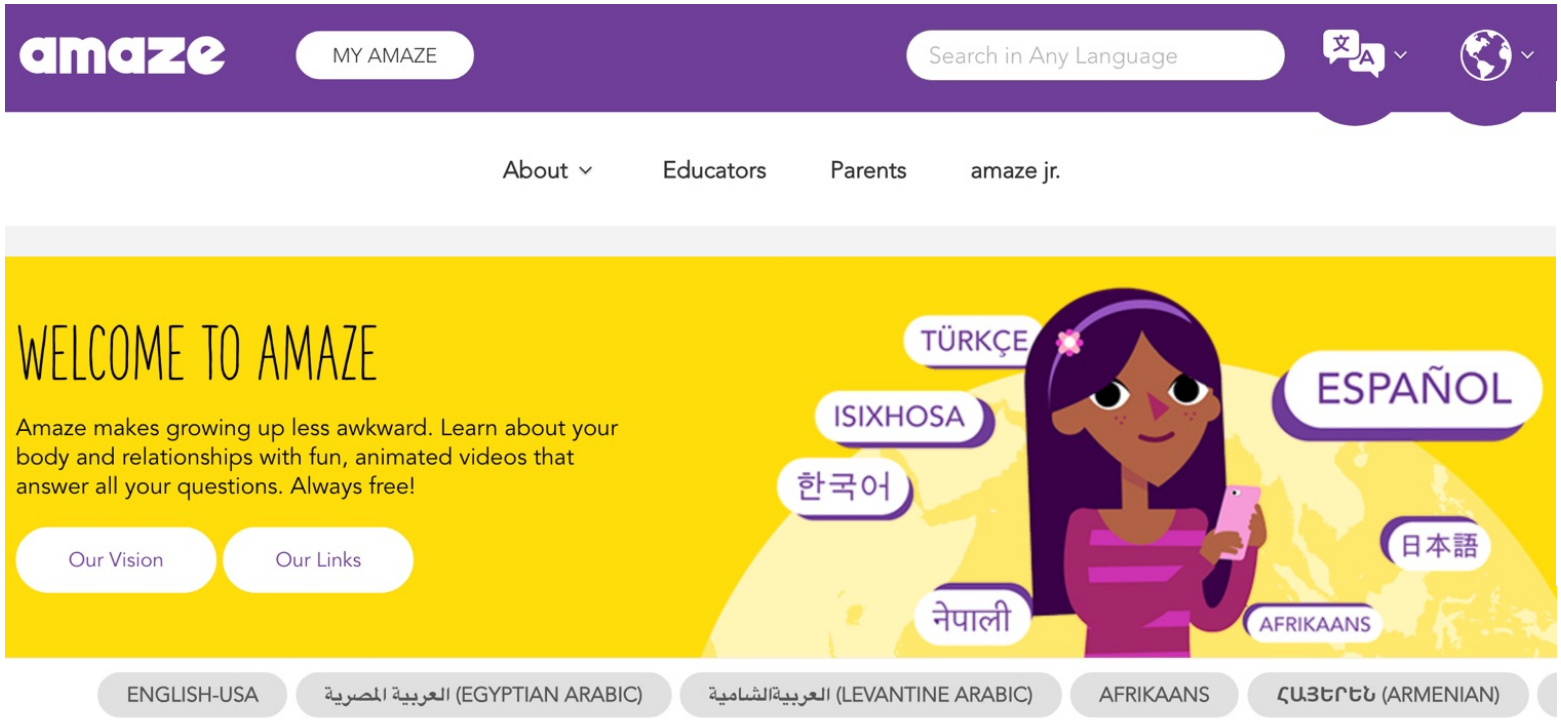
- Parent-child communication: comfort and openness.
- Family dynamics: cultural/religious factors.
- External support: therapist, peers, or community resources.

Holland Bloorview

Kids Rehabilitation Hospital

hollandbloorview.ca/disability-sexuality-resource-hub




amaze
MY AMAZE
Search in Any Language
About
Educators
Parents
amaze jr.
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AFRIKAANS
ENGLISH-USA
العربية المصرية (EGYPTIAN ARABIC)
العربيةالشامية (LEVANTINE ARABIC)
AFRIKAANS
ՀԱՅԵՐԵՆ (ARMENIAN)



OPTIMIZE DMD: An international, collaborative network dedicated to optimizing endocrine and bone healthcare for people with DMD through education, research, and advocacy.

OPTIMIZE DMD
Est. 2023

OPTIMIZING ENDOCRINE AND BONE HEALTH MANAGEMENT IN DUCHENNE MUSCULAR DYSTROPHY



SUMMARY OF ENDOCRINE HEALTH IN DMD:

BONE HEALTH

- ❑ Vitamin D Level (goal >30 ng/dL)
- ❑ Vit. D Supplement
- ❑ Calcium from diet / supplement
- ❑ DXA Bone Density Scan
- ❑ Spine Fracture Screening
- ❑ Bone strengthening medication

IF TAKING STEROIDS…

Steroids are extra important during illness.
Continue oral steroids for illness

- ❑ Emergency Solu-Cortef Injection
- ❑ School Sick-Day Plan

PUBERTY

Puberty hormones are good for bones!

- ❑ Monitor exam and morning labs

LOW SUGAR, HIGH CALCIUM FOODS

- ❑ Diabetes Screening
- ❑ Nutrition

