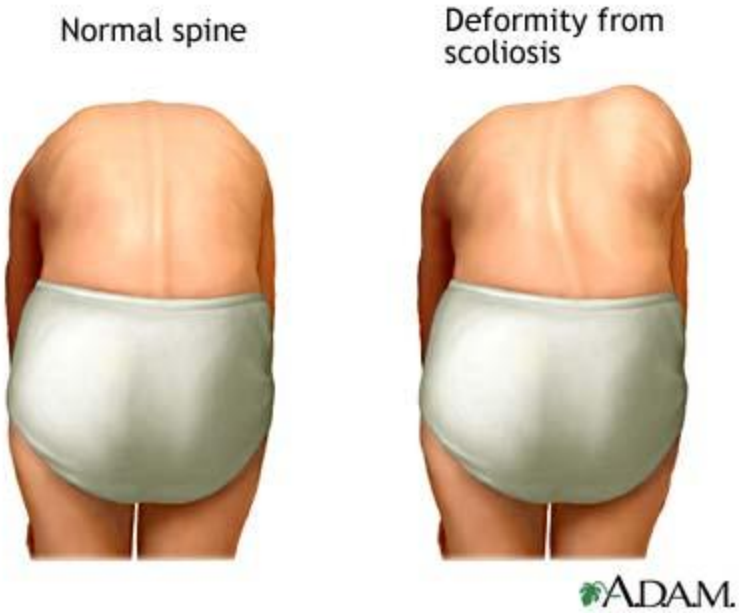
SCOLIOSIS IN SMA

SCOLIOSIS

- **Scoliosis** is a curvature of the spine from side to side, out of the normal vertical plane

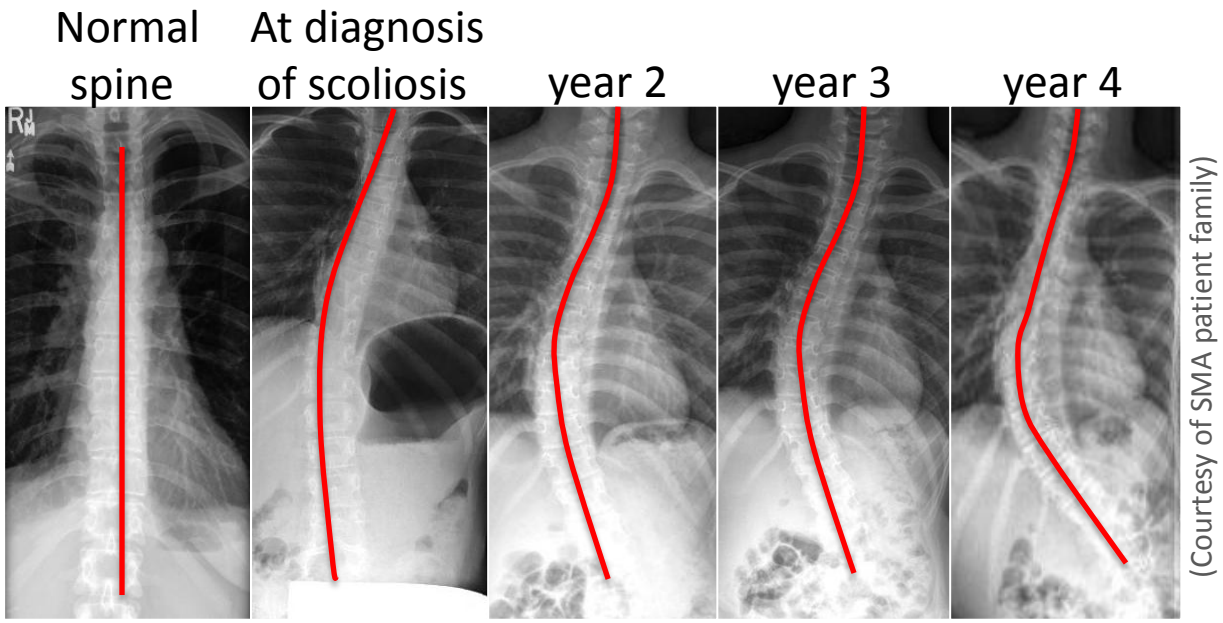


The forward bend test is used most often in schools and doctor's offices to screen for scoliosis.

(http://www.umm.edu/patiented/articles/how_scoliosis_diagnosed_000068_6.htm#ixzz2IuMZJK6m)

SCOLIOSIS IN SMA

- In SMA, as muscle weakness progresses in the back muscles while kids continue to grow, gravity pulls down on the spine and curves develop into scoliosis



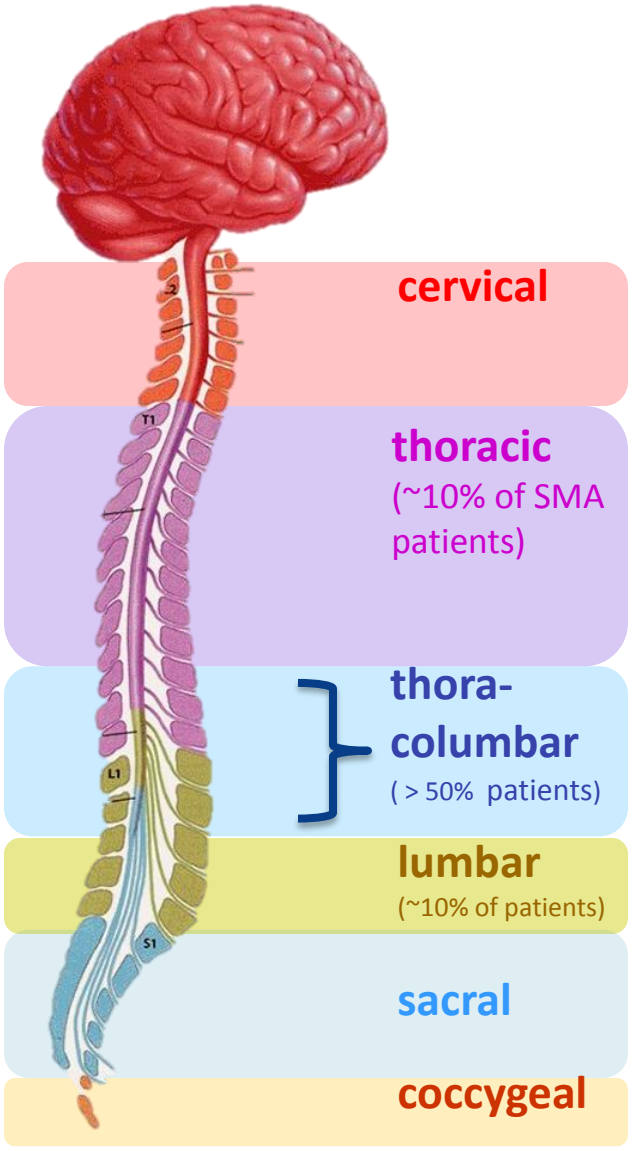
Progression of scoliosis in SMA

SMA PATIENTS DEVELOP SCOLIOSIS

- Almost all children with SMA (90%) develop scoliosis over the course of the disease (Granata et al, 1989)
  - Starts as early as 8-9 months of age (Grayhack, 2012)
  - Average age of onset is between 6-8 years
  - Most Type 2 patients develop scoliosis (Mullender et al, 2008)
- Progression of scoliosis is slower in patients affected by mild forms of the disease
- Progression is slower in walkers than in patients who have stopped walking
- In most ambulant patients with mild SMA, scoliosis does not impact their daily functioning and does not worsen greatly with age

SCOLIOSIS CAN OCCUR IN DIFFERENT SECTIONS OF THE SPINE

- In natural history studies, 75% of SMA patients with scoliosis have a single curve (Granata et al, 1989)
  - Thoracolumbar: >50%
  - Thoracic: ~10%
  - Lumbar: ~10%
- Roughly one fourth of patients have a double curve (a curve occurring in more than one location of the spine)



(http://sifoundation.webs.com/spinalcordinjury.htm)

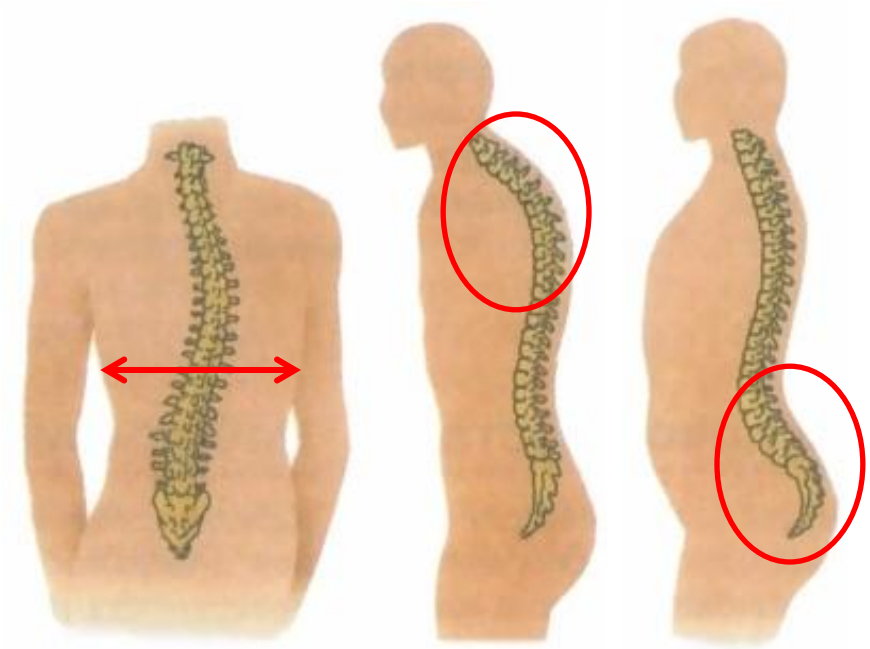
SMA PATIENTS CAN HAVE ADDITIONAL CURVATURES OF THE SPINE WITH SCOLIOSIS

The spine can curve in several directions:

Scoliosis – side to side curve

Kyphosis – backwards (or hunchback) curve

Lordosis – curve forward or inward



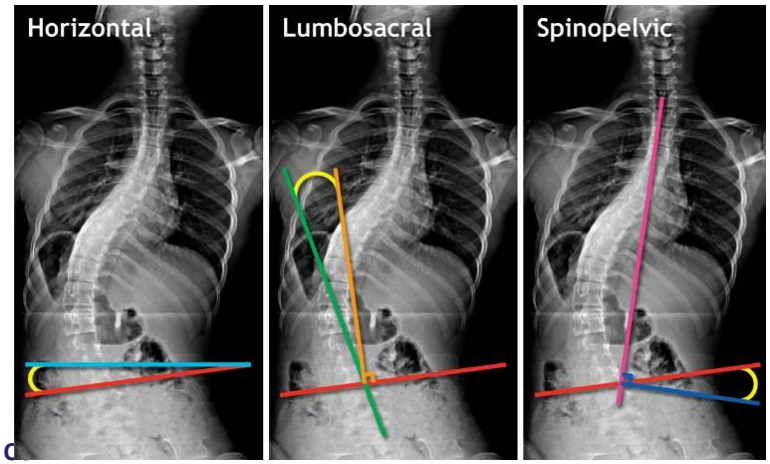
scoliosis kyphosis lordosis

(http://doctorumit.com/kifozeng.html)

Roughly one third of patients have kyphosis, lordosis, and/or pelvic tilt coupled with scoliosis
(Granata et al, 1989)

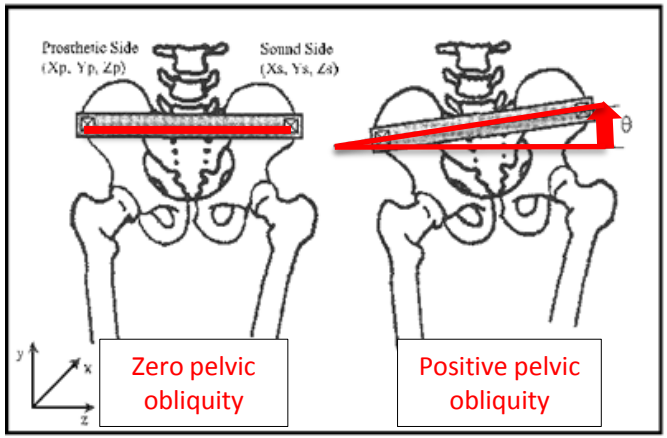
SMA PATIENTS' PELVIS AND HIPS CAN BE AFFECTED

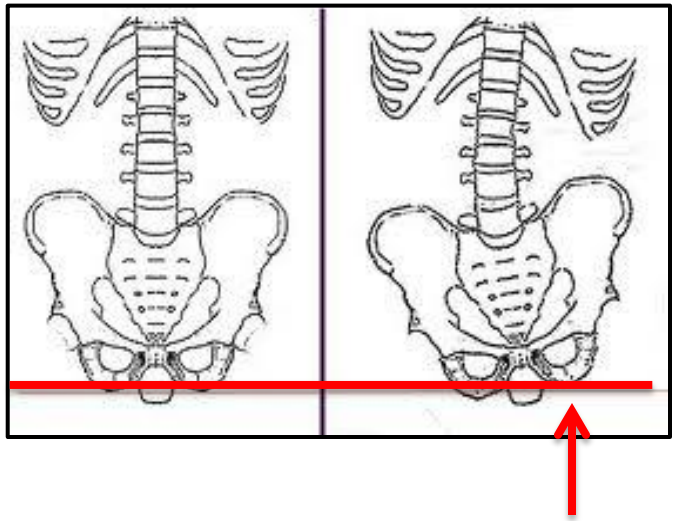
- **Pelvic obliquity** is a condition in which the pelvis is uneven, rotated downward on one side
  - Caused by muscle weakness, contractures in hips, or a combination of the two
- Early development of progressive scoliosis with pelvic obliquity is the most significant orthopedic problem in non-ambulatory SMA



Dayer et al, 2012

Methods of measuring pelvic obliquity





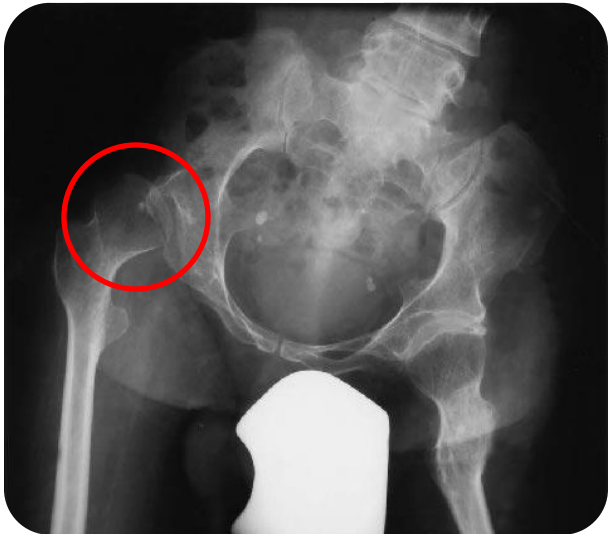
http://www.rehab.research.va.gov/jour/00/37/1/michaud.html

PELVIC OBLIQUITY CAN LEAD TO HIP DISLOCATION, A COMMON PROBLEM IN SMA

- Natural history studies report 10-30% of Type 2 and Type 3 patients had dislocated hips; 50% of Type 2 patients in a separate study (Sporer et al, 2003; Granata et al, 1990; Evans et al, 1981)
- Hip subluxation, a partial dislocation, is also common (50%) (Sporer et al, 2003)
- Taken together ~60% of SMA Type 2 patients had either hip subluxation or dislocation
- In patients who are able to stand or walk, no hip dislocation is present (Granata et al, 1990)

Right hip dislocation in an SMA patient

http://emedicine.medscape.com/article/



Right hip subluxation in an SMA patient

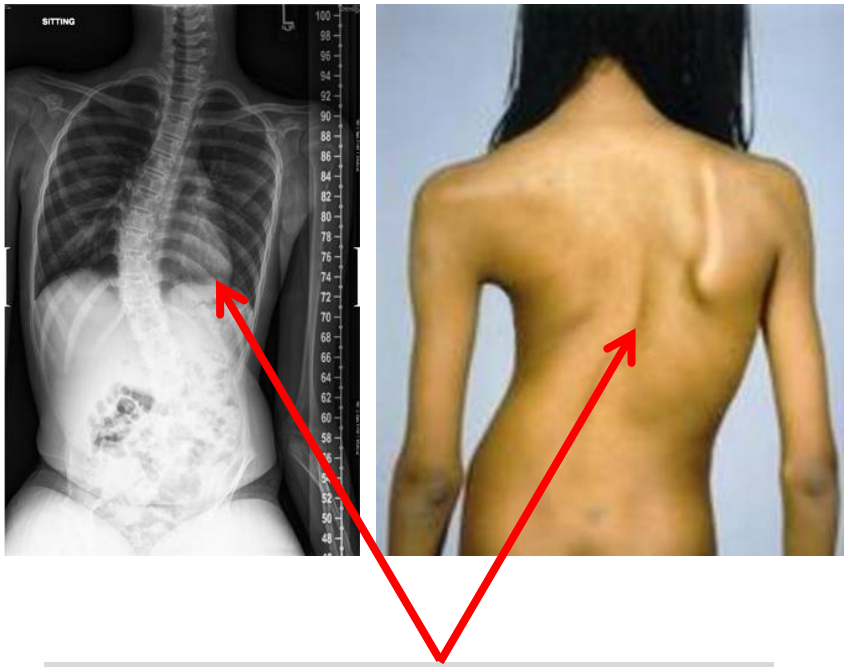
Sporer et al, 2003

HIP SURGERY MAY BE AN OPTION FOR HIP DISLOCATIONS: ASSESSED ON A CASE BY CASE BASIS

- In a natural history study, 9 out of 30 Type 2/3 patients had hip surgery during the course of the disease (Sporer et al, 2003)
- Advocates of surgical treatment believe that a located hip is required to improve balance, maintain pelvic alignment, minimize pain and reduce the incidence of ischial (pelvic) pressure (Zenios et al, 2005)
- Others have questioned the benefits of operative treatment because the recurrence of subluxation may require further surgeries (Thompson et al, 1990)

SCOLIOSIS HAS DETRIMENTAL EFFECTS

- Affects appearance
- Functional effects
  - Interferes with use of arms
  - Loss of sitting balance (arms needed to support sitting)
- Pulmonary – change in shape of chest cavity
  - **Can lead to respiratory dysfunction and reflux**
  - Studies have shown a correlation between scoliosis severity and lung capacity

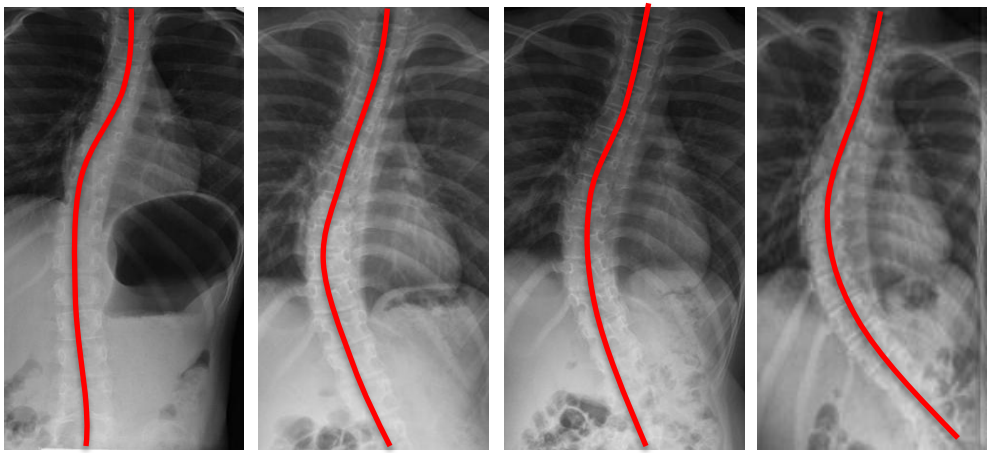


Spinal deformity crushes lungs and leads to respiratory dysfunction

SCOLIOSIS IN SMA IS PROGRESSIVE: NATURAL HISTORY STUDY

- In intermediate SMA forms, the average expected increase in scoliosis is roughly 10° per year (Granata et al, 1989)
- In mild patients who stopped walking, the average increase is 3° per year

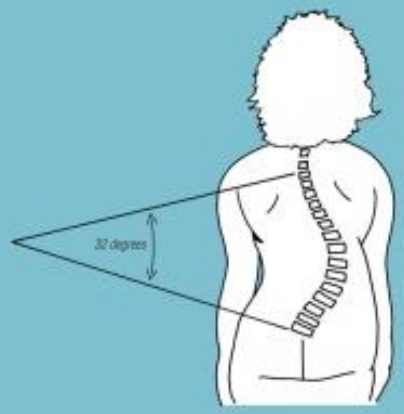
Case study of an intermediate Type 2 SMA with progressive scoliosis



8 years old	10 years old	11 years old	12 years old
9°, T1-T6	10°, T1-T5	12°, T1-T5	35°, T5-T12
34°, T6-L1	42°, T6-L1	43°, T6-L1	45°, T12-L4
17°, L1-S1	32°, L1-S1	24°, L1-S1	

Degree of scoliosis is measured by the Cobb method from radiographs (Cobb, 1948)

Cobb Angle



TREATMENTS FOR SCOLIOSIS: BRACING

- Currently, there is controversy in the field about whether bracing has an effect on slowing curve progression in children with SMA (Communication with Dr. Vitale)
- Some physicians use braces to improve quality of life, i.e. helps to support trunk and makes care and positioning easier
- Some physicians believe that bracing can play an important role in postponing scoliosis surgery
- However, bracing does not prevent development of scoliosis (Cheung et al, 2012; Mehta, 2005; Robinson et al, 1996, Sponseller et al, 2009)
- Can cause respiratory difficulty or complications (Robinson et al, 1995)

SMA patient that wore brace at ages 4 – 9 before surgery

http://quest.mda.org/article/in-focus-straightening-growing-spine

http://www.hhsc.ca/body.cfm?id=355

TREATMENTS FOR SCOLIOSIS: BRACING TYPES

- Several types of braces:
  - The Boston brace, or thoraco-lumbar-sacral orthoses (TLSO), is commonly prescribed for SMA patients
  - The Milwaukee brace, or cervico-thoraco-lumbo-sacral orthosis, is generally used to hold an advancing curve of 25° to 40°; intended to minimize the progression to an acceptable level
  - Soft bracing has also been recommended in neuromuscular scoliosis which helps to prevent progression and also improves quality of life (Letts et al, 1992)

A Boston brace (above). A custom version can be seen below, usually used after surgery and made to measurement.

http://www.choa.org/Childrens-Hospital-Services/Orthopaedics/Programs-Services/Orthotics-and-Prosthetics/Spinal-Bracing

TREATMENTS FOR SCOLIOSIS: SURGERY

- **Scoliosis surgery** – studies show that surgery can preserve pulmonary function, improve nursing care, comfort, appearance and overall quality of life
- **Pros for surgery**
  - Improved sitting balance, stable spine and trunk; results in greater functional freedom of the arms
  - Aesthetic improvement – improvement in sitting height, relieves back pain
  - Pelvic obliquity can also be ameliorated with surgery
- **Cons for surgery**
  - Decline in functional activities (such as movement of the head) after spinal surgery has been reported by some authors (Hart and McDonald, 1998)
  - Risk of complications during surgery include blood loss, respiratory distress, infection, pain or nerve damage

SCOLIOSIS SURGERY: NEED AND TIMING IS SPECIFIC TO INDIVIDUAL

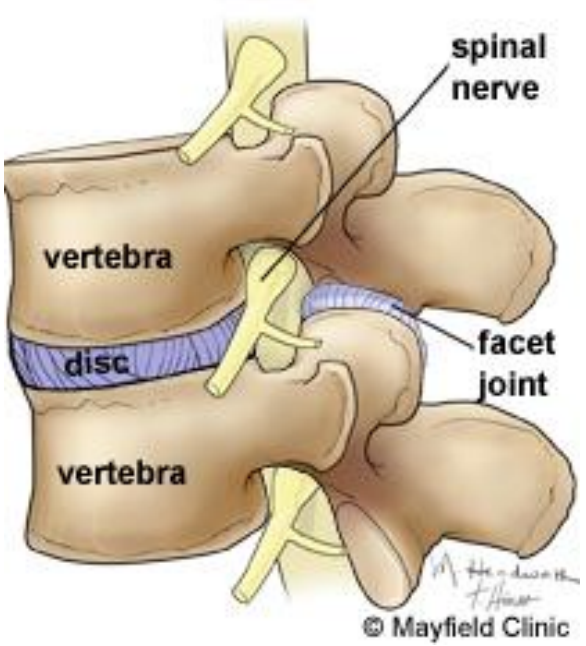
- Because almost all cases of scoliosis progress, leading to poor posture with pulmonary and functional limitations, surgery is generally recommended
- In general, surgery conducted at an age of 10 years or older when patients have stopped growing:
  - Can achieve 100% correction of the curve in some cases
  - Results have been satisfactory for posture – patients tend to report improvements in sitting comfort and balance
  - Several studies show a slowing of decline in pulmonary function and others an improvement in function postoperatively (Chng et al, 2003, Granata et al, 1993, Robinson et al, 1995)
- There are surgical options for younger patients that develop rapid scoliosis

SCOLIOSIS SURGERY: TREATMENT OPTIONS

- Depending on several factors, a specific type of surgical procedure will be selected
  - Severity and location of the curve
  - Age of patient and the degree of spinal maturity
  - Pulmonary function
  - Clinical opinion of the surgeon and preferences of the child and parent
- Often a combination of these treatments are considered
  - **Spinal fusion**
  - **Growing rods**
  - **Vertical expandable prosthetic titanium ribs (VEPTR)**
- Generally, surgery is considered for children >10 years and with a curve >40°
  - The goal is to prevent progression and to maintain balance; this is most often achieved with spinal fusion
- For children <5 years who develop severe and rapid scoliosis, growing rods and VEPTR are commonly used

SCOLIOSIS SURGICAL PROCEDURES: SPINAL FUSION

- Spinal fusion is surgery to permanently connect two or more vertebrae in the spine, eliminating motion between them
- Involves techniques designed to mimic the normal healing process of broken bones – placing a bone or bone like material within the space of two vertebrae
- Fusion prevents further growth of the spine

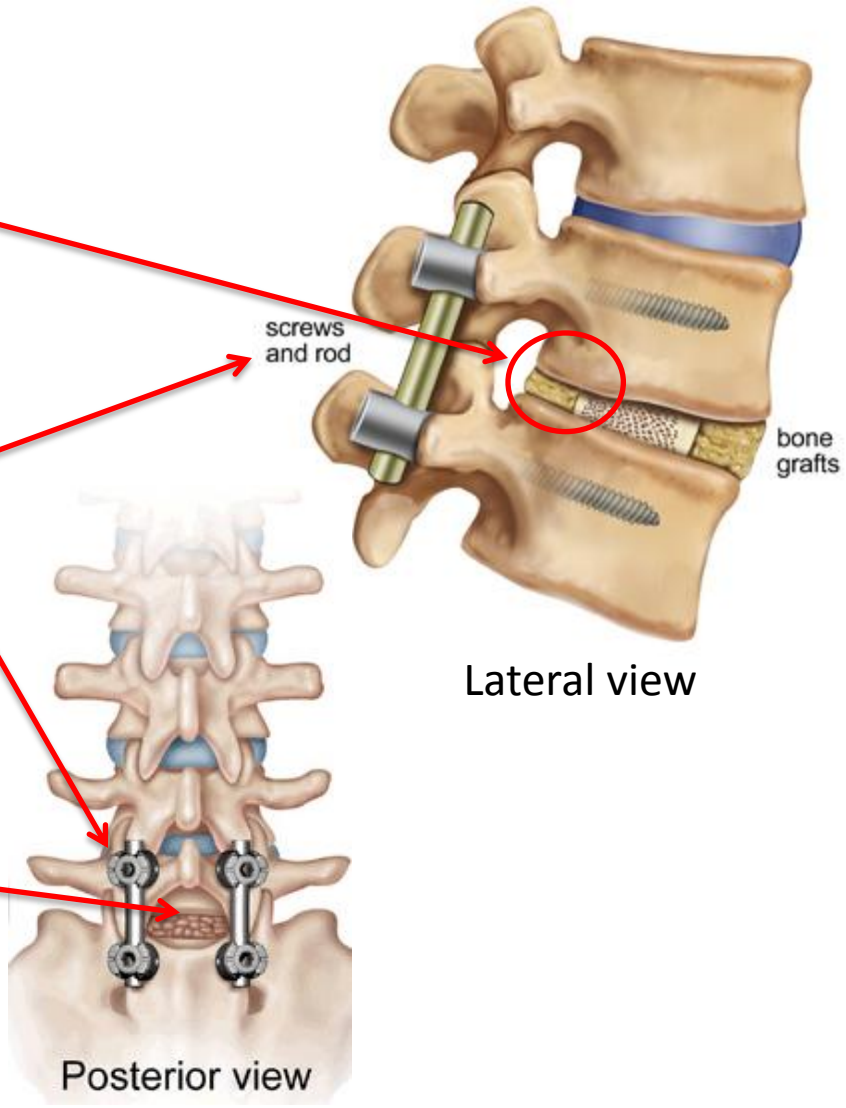


Normal spinal anatomy

http://www.mayfieldclinic.com/PE-FusionPreparing.htm

SCOLIOSIS SURGICAL PROCEDURES: SPINAL FUSION

- In fusion, bone grafts from pieces of bone or bony material are placed in between the vertebrae to help create a solid piece of bone
- Rods, hooks, screws or wires may be used to hold the vertebrae together while the fusion heals
- Over several months the bones and grafts fuse together to become one solid unit



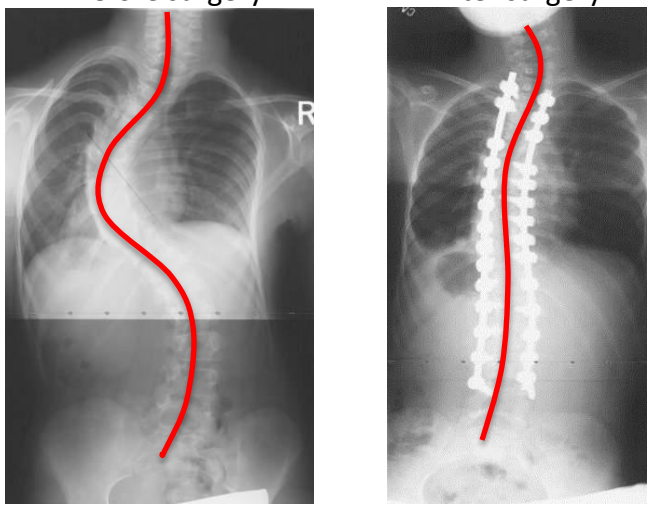
http://www.lyerlyneuro.com/spinal-fusion.html

http://www.allaboutbackandneckpain.com/explore/spinal-fusion.asp#

SCOLIOSIS SURGICAL PROCEDURES: SPINAL FUSION

Case study of a Type 2 SMA patient with spinal fusion

- Patient achieved over a 50% correction in her curve
- Patients in a separate study were monitored for 6 to 12 months post surgery and their vital capacity (measure of lung function) remained constant (Fugak et al, 2012)

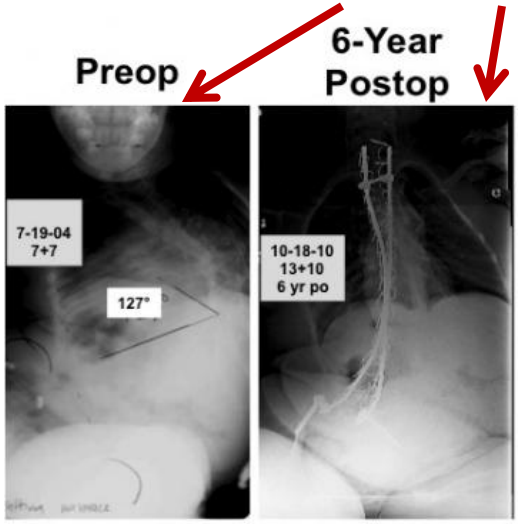


Side view

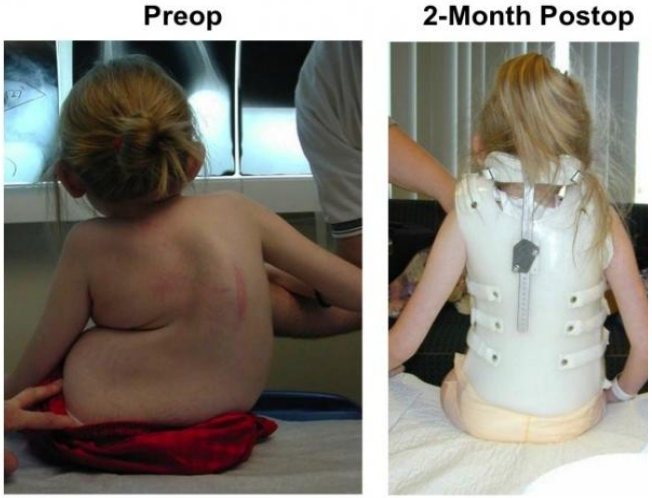
(Courtesy of Dr. Michael Vitale, Columbia University Medical Center)

CASE STUDY: 7 YEAR OLD SMA PATIENT WITH SEVERE CURVE (127°) RECEIVED A SPINAL FUSION

Curve correction 127° to 52°



Because of severe curve, frail nature and fragile bones, spinal correction was protected by braces



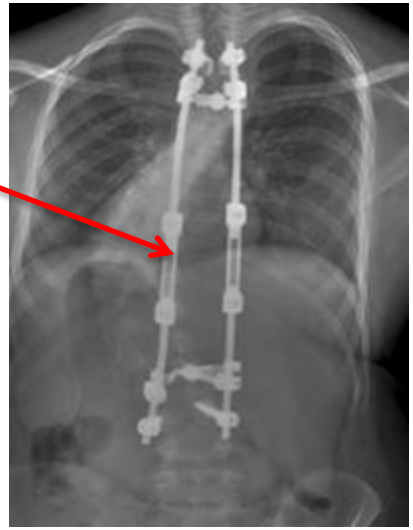
Patient 2 years post surgery has maintained correction and can breathe better as well

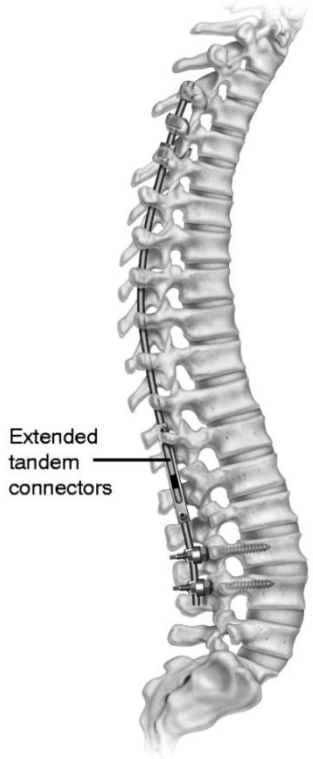
(Treated by Dr. Keith Bidwell in St. Louis, MO)

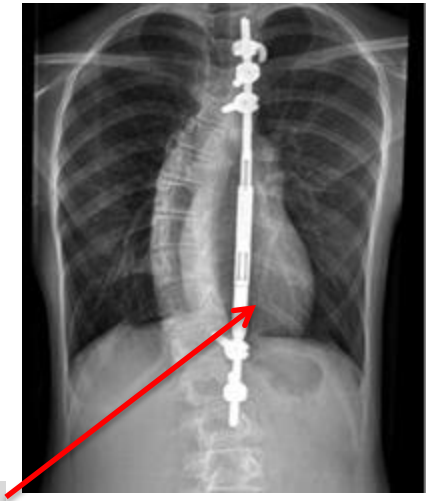
SCOLIOSIS SURGICAL PROCEDURES: GROWING RODS

- Growing rods are expandable devices that are attached to the top and base of the spine using screws or hooks
  - Surgery is required every 6 months to lengthen the rods
- Effective on patients with curves >40°
- Study of 11 SMA Type 1 and 2 patients found growing rods with pelvic fixation is an effective option with few complications (Chandran et al, 2011)
  - Average of 60% reduction in curve
  - Pelvic fixation is a surgical technique used in neuromuscular scoliosis that fuses the rods to the pelvis in order to address pelvic obliquity
- Growing rods allow for further growth of spine

Growing rod construct with fusion at the top and at the bottom of the spine.







Magnetically Controlled Growing Rod (MCGR) in a patient with idiopathic early onset scoliosis is a new treatment

http://www.srs.org/patient_and_family/scoliosis/early_onset_scoliosis/treatment/graphics/growing-rods.jpg

MAGNETICALLY CONTROLLED GROWING RODS (MCGR)

- New device approved in Europe and Asia to adjust rods (not FDA approved in the U.S.)
- Allows for remote, frequent outpatient procedures to adjust rods with growth
  - No need for sedation or anesthesia
  - No need for follow up surgeries
  - 10 minute adjustment procedure
- Study of 5 patients in Hong Kong showed no rod or wound complications (Cheung et al, 2012)
- Study of 14 patients in Hong Kong, Cairo and Ankara showed 1 superficial infection (Akbarnia et al, 2013)
- 50% curve correction after 24 and 10 months, respectively (Cheung et al, 2012; Akbarnia et al, 2013)
- Potential to implant rods at early stage (mild scoliosis), act as internal brace, maintain growth
- Informational video

http://www.srs.org/patient_and_family/scoliosis/early_onset_scoliosis/treatment/graphics/growing-rods.jpg

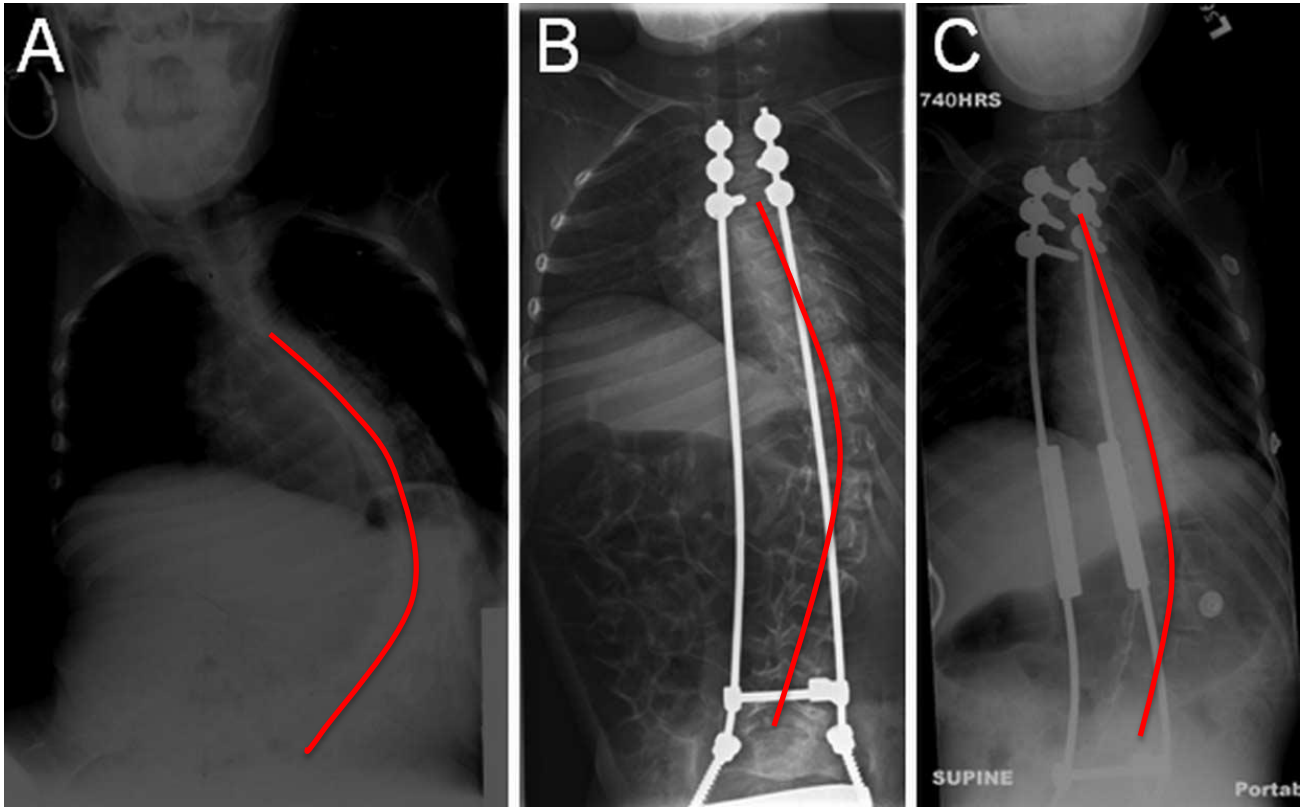
MCGR in a patient with idiopathic early onset scoliosis

MAGEC External Remote Control – to adjust rods

CASE STUDY OF GROWING RODS PROCEDURE

Case study of a 9 year old patient who had a curve correction of 60˚post surgery with growing rods

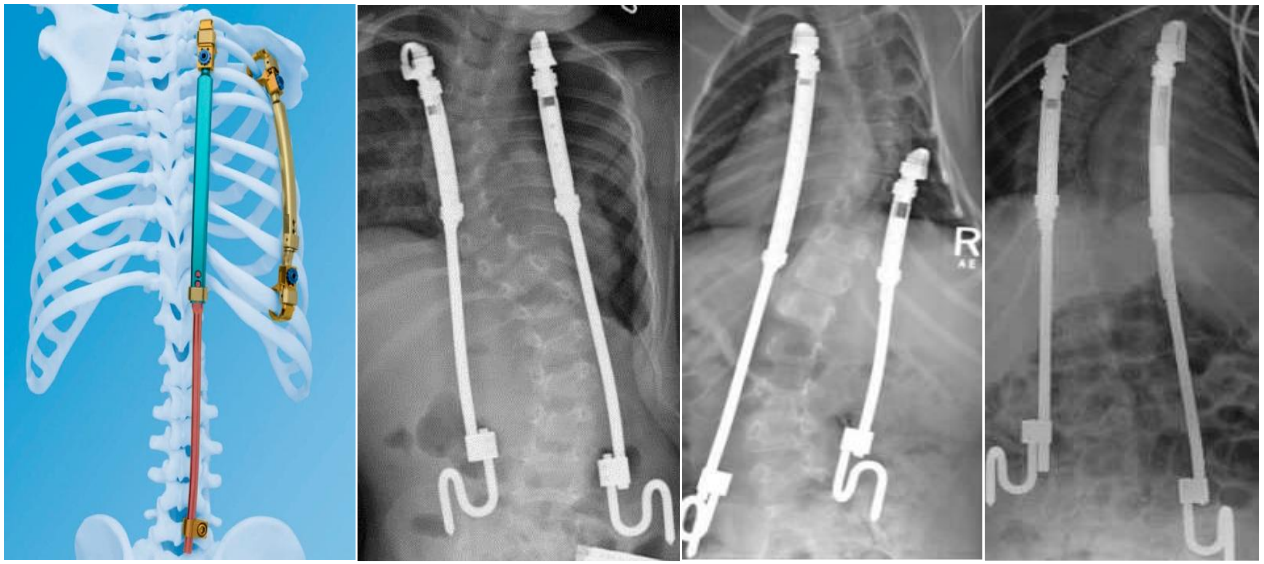
Before surgery, 102° Initial postop, 38° 2 years postop, 42°



McElroy et al, 2011

SCOLIOSIS SURGICAL PROCEDURES: VEPTR

- Vertical expandable prosthetic titanium ribs (VEPTR): rods that are attached to the ribs or the spine; there are few differences from growing rods
  - Helps straighten the spine
  - Helps separate the ribs to prevent deterioration of breathing function
- Small adjustments are made every 6 months to allow for growth
- No peer-reviewed studies published on VEPTR in SMA



http://www.synthes.com

(Courtesy of Dr. Michael Vitale, Columbia University Medical Center)

TREATMENT OPTIONS

Surgical procedure	Pros	Considerations
Spinal Fusion – fusion of the bone to stabilize and straighten the spine requiring rods, hooks, screws and wires	• Permanent • Requires one procedure • Tend to achieve good correction in 6-12 months • Does not prevent intrathecal delivery of drugs	• Surgery is most invasive, although new endoscopic procedures are emerging • Can cause infection post surgery • Back pain after surgery • Less mobility in back, neck • Only for nonambulatory patients > 10 years
Growing rods – expandable devices that are attached at the top and base of spine using screws and hooks	• **Option for younger children with curves >40°** • Allows for growth of the spine • Less invasive surgery • Achieve good correction • Does not prevent intrathecal delivery of drugs	• Surgical adjustments needed • May require spinal fusion at later date; less mobility in back and neck • Infections and loosening of device can occur after surgery
VEPTR – rods that are attached to the ribs or to the spine in the same way as growing rods	• **Option for younger children with >40° curves** • Good spine correction + separation of ribs may help prevent respiratory decline • Does not prevent intrathecal delivery of drugs	• Fewer published studies on effects • Surgical adjustments every 6 months • May require fusion at later date • Infections and loosening of device can occur after surgery

CONCLUSIONS

- Scoliosis is common for children with SMA
- There are several types of scoliosis and several treatment options
- The right treatment for a person is specific to the individual and includes factors like age, degree of scoliosis, pulmonary status, etc.
- See a specialist early to discuss bracing and review your child's status

PATIENT RESOURCE PAGES

- Quest Muscular Dystrophy Association page general information on treatment of scoliosis in patients with neuromuscular disorders: http://quest.mda.org/article/in-focus-straightening-growing-spine

- Families of SMA website on orthopedics including articles by clinicians with experience treating scoliosis: http://www.fsma.org/FSMACommunity/MedicalIssues/Orthopedic/

- Information on orthopedic care from FightSMA (taken from the consensus statement on care: http://www.fightsma.org/sma-guidebook/spinal-muscular-atrophy-treatment-symptoms/sma-orthopedic-care/

REFERENCES

- Akbarnia, B., K. Cheung, et al. (2013). “Next generation of Growth-Sparing Techniques.” Spine **38** (8): 665-670.
- Chandran, S., J. McCarthy, et al. (2011). "Early treatment of scoliosis with growing rods in children with severe spinal muscular atrophy: a preliminary report." J Pediatr Orthop **31**(4): 450-454.
- Cheung, Cheung et al. (2012). “Magnetically controlled growing rods for servere spinal curvature in young children: a prospective case series.” Lancet 379: 1967-1974.
- Chng, S. Y., Y. Q. Wong, et al. (2003). "Pulmonary function and scoliosis in children with spinal muscular atrophy types II and III." J Paediatr Child Health **39**(9): 673-676.
- Cobb JR (1948). “Outline for the study of scoliosis.” Instr Course Lect AAOS **5**: 261 - 75.
- Dayer R., J.A. Ouellet, et al. (2012). “Pelvic fixation for neuromuscular scoliosis deformity correction.” Curr Rev Musculoskelet Med 5:91-101.
- Fujak, A., W. Raab, et al. (2012). "Operative treatment of scoliosis in proximal spinal muscular atrophy: results of 41 patients." Arch Orthop Trauma Surg.
- Granata, C., S. Cervellati, et al. (1993). "Spine surgery in spinal muscular atrophy: long-term results." Neuromuscul Disord 3(3): 207-215.
- Grayhack, J (Accessed 2012). “The Hidden Twist…Scoliosis,” FSMA website: http://www.fsma.org/FSMACommunity/MedicalIssues/Orthopedic/links/index.cfm?ID=2847&TYPE=1500
- Granata, C., L. Merlini, et al. (1989). "Spinal muscular atrophy: natural history and orthopaedic treatment of scoliosis." Spine (Phila Pa 1976) **14**(7): 760-762.
- Hart, D. A. and C. M. McDonald (1998). "Spinal deformity in progressive neuromuscular disease. Natural history and management." Phys Med Rehabil Clin N Am **9**(1): 213-232, viii.
- Mercuri, E., E. Bertini, et al. (2012). "Childhood spinal muscular atrophy: controversies and challenges." Lancet Neurol **11**(5): 443-452.
- Mullender, M., N. Blom, et al. (2008). "A Dutch guideline for the treatment of scoliosis in neuromuscular disorders." Scoliosis **3**: 14.
- Robinson, D., C. S. Galasko, et al. (1995). "Scoliosis and lung function in spinal muscular atrophy." Eur Spine J **4**(5): 268-273.
- Yazici (ed), “Non-Idiopathic Spine Deformities in Young Children,” DOI: 10.1007/978-3-642-19417-7_7

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