VEPTR Treatment of Jarcho-Levin Syndrome

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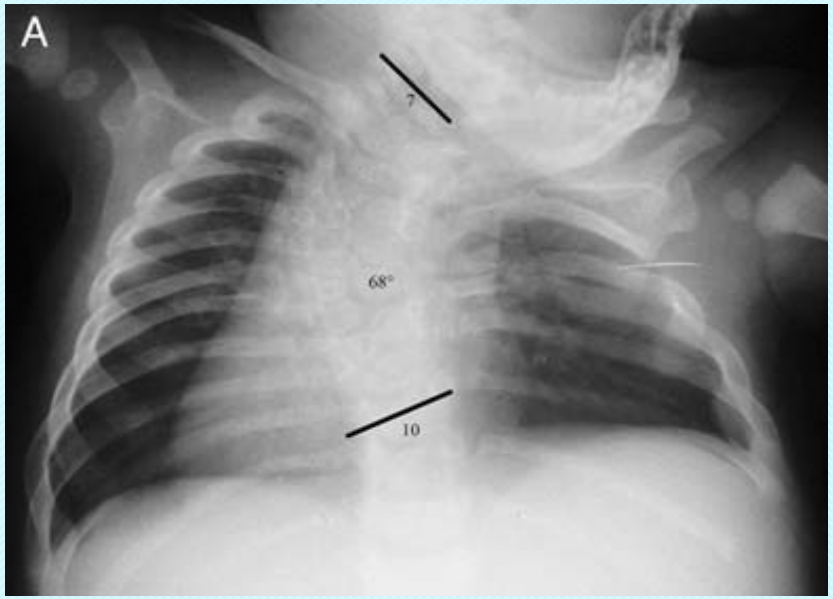
Background

- Jarcho-Levin Syndrome
  - Associated with thoracic insufficiency syndrome (TIS)
  - Historically an eponym for a multitude of radiographic and skeletal deformities.
  - Currently subtyped via phenotype by
    - distribution of skeletal anomalies
    - inheritance pattern
    - prognosis

Background

Spondylocostal dysostosis (SCD)

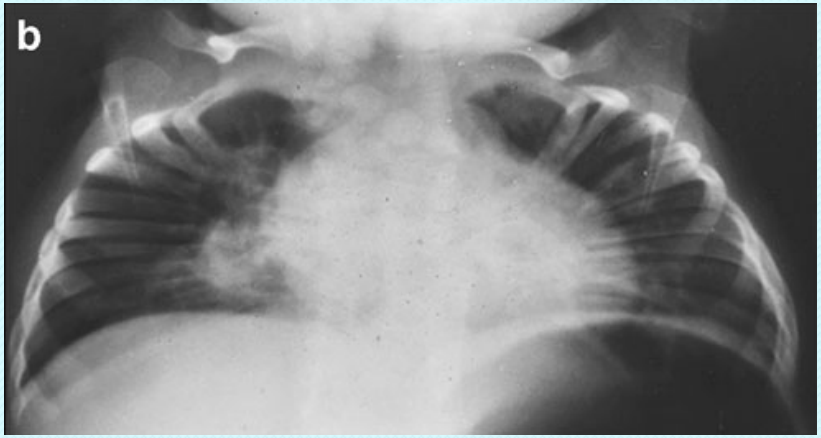
- Intrinsic rib anomalies such as broadening, bifurcation, and fusion with no symmetric fusion of the ribs.
- Also known as Lavy-Moseley syndrome.
- Scoliosis and early demise due to respiratory failure secondary to TIS is common.



Background

Spondylothoracic dysplasia (STD)

- Fusion of all the ribs at the costovertebral joints bilaterally due to segmentation and formation vertebral defects throughout spine without intrinsic rib anomalies
- "Crab-like" radiographic distinction.
- More severe subtype, estimated 25% survive to adulthood, those surviving have a lung volume 28% normal.
- Scoliosis and No Scoliosis subtypes observed



Objective

Jarcho-Levin patients with VEPTR treatment

- Quantify the changes in thoracic architecture
- Assess the changes in respiratory status
- Identify complications associated with treatment

Methods

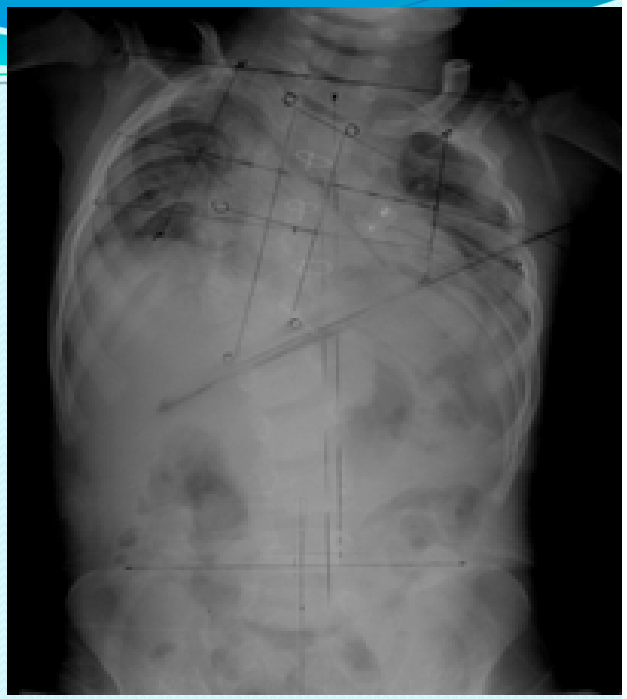
- IRB approved retrospective study
- Chart Review
  - Demographic: Age at initial implant
  - Respiratory Status: Assisted Ventilation Rate (AVR) scale
  - Complication
- Radiographic Assessment
  - Cobb Angle
  - Thoracic Height
  - Hemi- and Thoracic Widths
  - Space Available for lungs (SAL)
- Data analyzed using paired *t*-test and Mann-Whitney U test

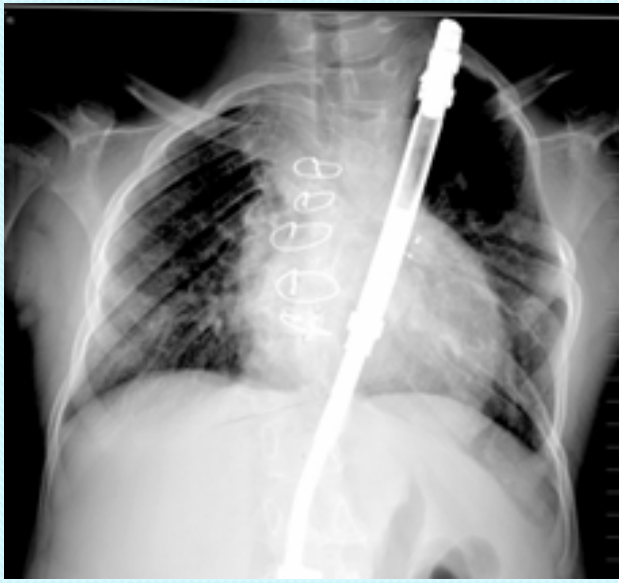
Results

	Cohort	Spondylocostal Dysostosis (SCD)	Spondylothoracic Dysplasia with Scoliosis (STD-S)	Spondylothoracic Dysplasia without Scoliosis (STD-N)
Patients	29	10	9	10
Age - 1st Implant	3.7	3.07	4.67	3.50
Age - Latest Follow-Up	10.4	11.00	11.86	8.38
Mean TIP	6.7	7.94	7.19	4.88

Spondylocostal Dysostosis (SCD)

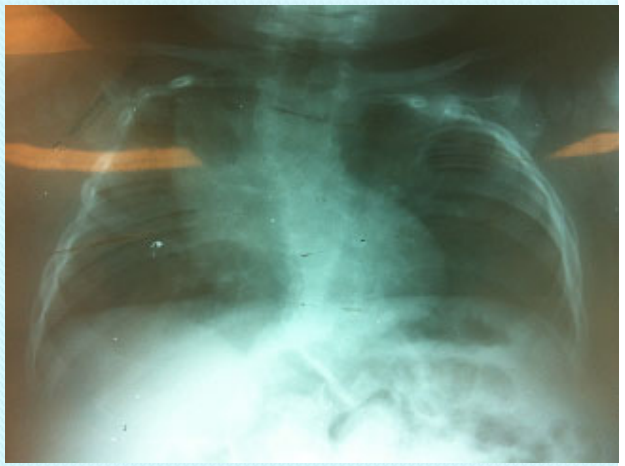
	Cobb	Thoracic Height (mm)	Thoracic Width (mm)	SAL
Pre-operative	54.3	98.6	135.8	0.774
Post-operative	35.8	108.8	143.2	0.894
***t*-test**	*0.04*	*0.02*	*0.14*	*0.04*
Latest Follow-Up	32.3	143.2	178.0	0.934
***t*-test**	*0.014*	*0.00007*	*0.00094*	*0.0098*

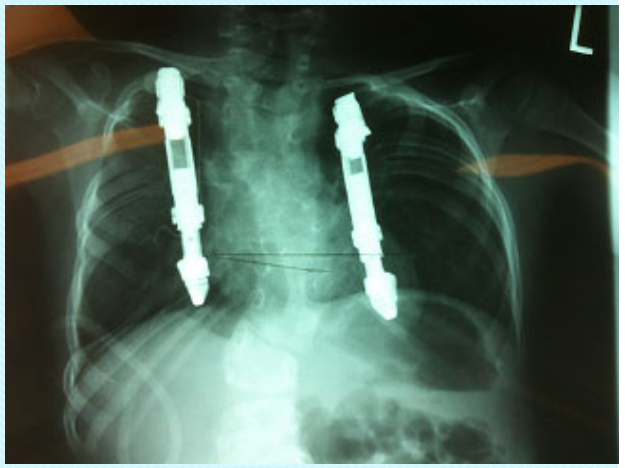




Spondylothoracic Dysplasia with Scoliosis (STD-S)

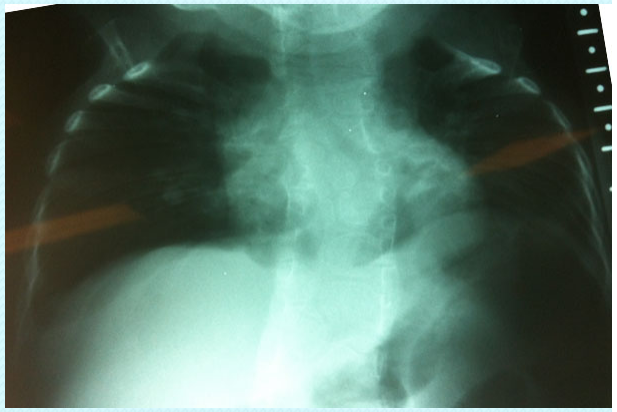
	Cobb	Thoracic Height (mm)	Thoracic Width (mm)	SAL
Pre-operative	23.9	106.7	157.7	0.907
Post-operative	18.8	112.7	163.6	0.937
***t*-test**	*0.003*	*0.0004*	*0.1*	*0.4*
Latest Follow-Up	19.2	135.7	192.7	0.965
***t*-test**	*0.01*	*0.0004*	*0.0016*	*0.12*

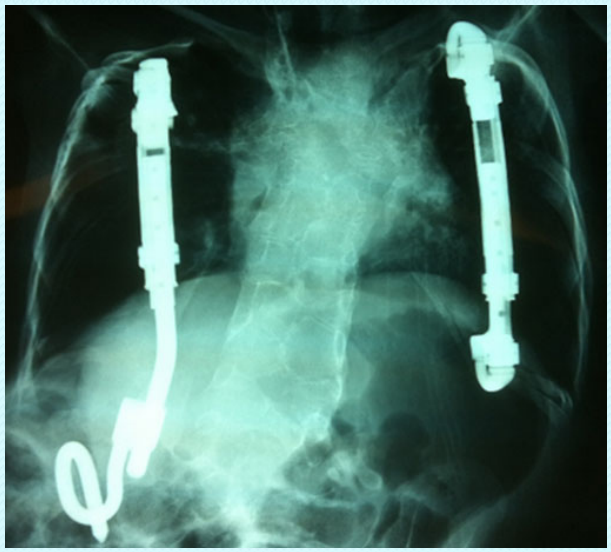




Spondylothoracic Dysplasia without Scoliosis (STD-N)

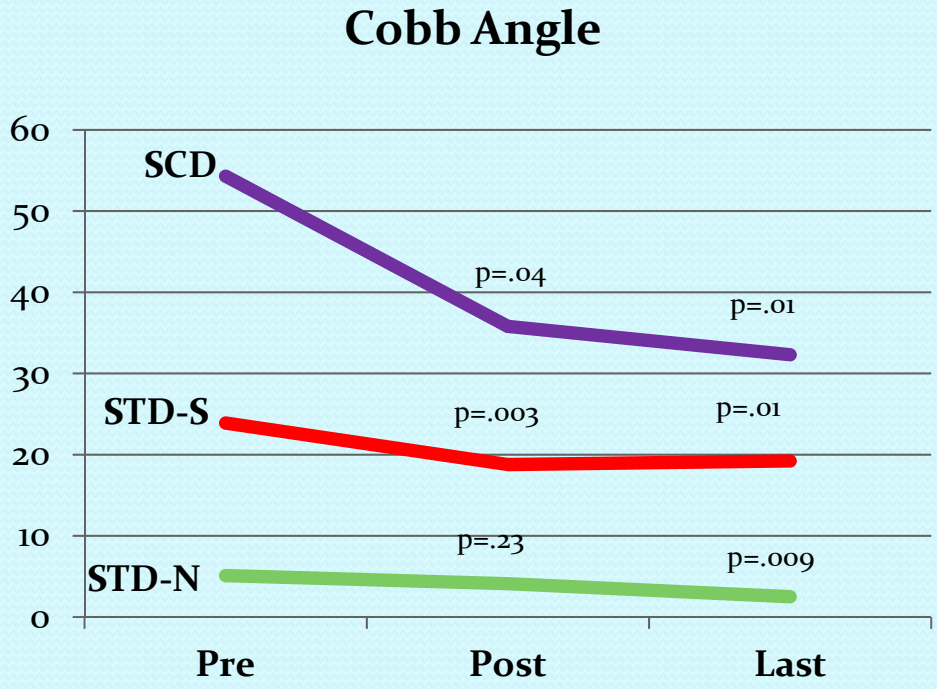
	Cobb	Thoracic Height (mm)	Thoracic Width (mm)	SAL
Pre-operative	5.1	91.2	156.3	0.964
Post-operative	4.1	93.6	165.5	1.006
***t*-test**	*0.2*	*0.017*	*0.01*	*0.09*
Latest Follow-Up	2.5	112.0	188.0	1.015
***t*-test**	*0.009*	*0.00012*	*0.00003*	*0.04*

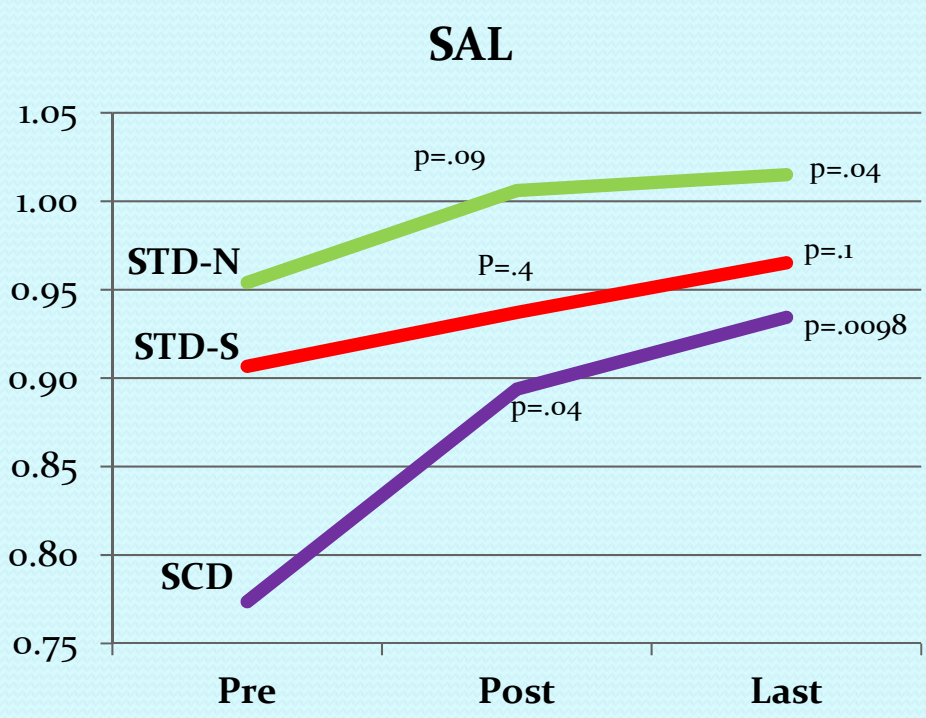




Results

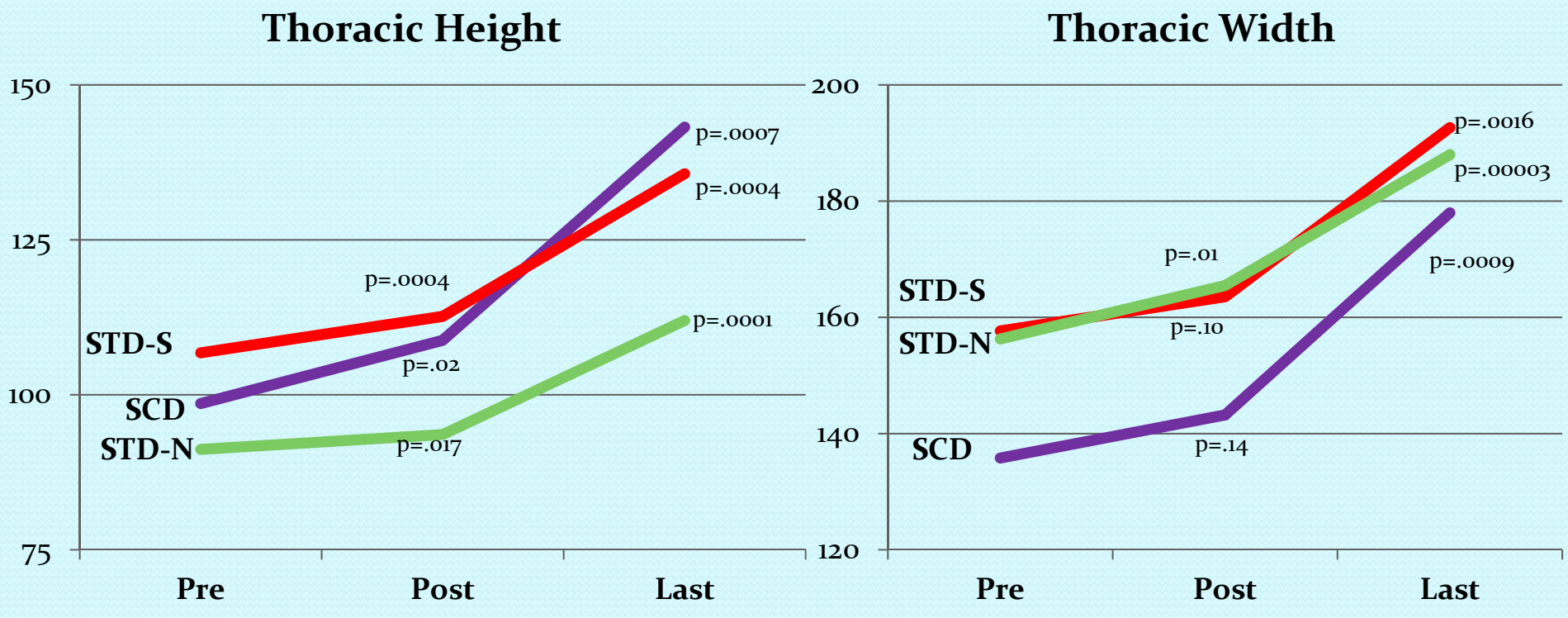
Increased **symmetry** was achieved with initial VEPTR treatment and maintained throughout course of treatment.





Results

Increased **thoracic height and width** were achieved with initial VEPTR treatment and maintained throughout course of treatment.



Discussion

- There were significant gains in thoracic height, at 31% of normal.
- AVR performance increased (7) or remained at room air (20) in 94% of the cohort (27 of 29).
  - No subtype showed a statistical difference in AVR improvement.
  - 2 unchanged patients still under active treatment.
- 55% (15 of 27) complications were migrations/ dislodgements.
- VEPTR achieved success in treating all Jarcho-Levin patients, while its most beneficial impact varied between subtypes according to that subtypes perceived impairment and thus, planned course of treatment.

References

1. Solomon L, Bosch-Jime´nez R, Reiner L. Spondylothoracic dysostosis. Arch Pathol Lab Med. 1978;102:201–205.
2. Campbell RM, Hell-Vocke AK. Growth of the thoracic spine in congenital scoliosis after expansion thoracoplasty. J Bone Joint Surg Am. 2003;85:409–420.
3. Campbell RM, Smith M. Thoracic insufficiency syndrome and exotics scoliosis. J Bone Joint Surg Am. 2007;89:108–122.
4. Franceschini P, Grassi E, Fabris C, et al. The autosomal recessive form of spondylocostal dysostosis. Pediatr Rad. 1974;112:673–675.
5. Turnpeny PD, Bulman MP, Frayling TM, et al. A gene for autosomal recessive spondylocostal dysostosis maps to 19q13.1-q13.3. Am J. Hum Genet. 1999;65:175–182.
6. Walter E. Berdon, Lampl, Brooke S. et al. Clinical and radiological distinction betweenspondylothoracic dysostosis (Lavy-Moseley syndrome) and spondylocostal dysostosis (Jarcho-Levin syndrome). Pediatr Radiol. 2011. 41:384–388
7. Teli M, Hosalkar H, Gill I, et al. Spondylocostal Dysostosis. Thirteen new cases treated by conservative and surgical means. Spine. 2004;29:1447–1451.
8. Cornier AS, Ramirez N, Arroyo S et al. Phenotype characterization and natural history of spondylothoracic dysplasia syndrome: a series of 27 new cases. Am J Med Genet A. 2004. 128A:120–126
9. Cornier AS, Staehling-Hampton K, Delventhal KM et al. Mutations in the MESP2 gene cause spondylothoracic dysostosis/Jarcho-Levin syndrome. Am J Hum Genet. 2008. 82:1334–1341
10. Kim, Daniel H. et al. Surgery of the Pediatric Spine. ISBN 978-1-58890-342-6
11. Campbell RM, Smith MD, Mayes TC, et al. The characteristics of thoracic insufficiency syndrome associated with fused rib and congenital scoliosis. J Bone Joint Surg Am. 2003;85:399–408.
12. Betz R, Mulcahey M, Ramirez N, et al. Mortality and lifethreatening events after Vertical Expandable Prosthetic Titanium. Rib surgery in children with hypoplastic chest wall deformity. J Pediatr Orthop. 2008;28:850–853.
13. Ramirez, Norman et al. Vertical Expandable Prosthetic Titanium Rib as Treatment of Thoracic Insufficiency Syndrome in Spondylocostal Dysplasia. J Pediar Ortho. 2010. 30:6:522,526.
14. Ramirez, Norman et al. Vertical Expandable Prosthetic Titanium Rib as Treatment of Thoracic Insufficiency Syndrome in Spondylocostal Dysplasia. J Pediar Ortho. 2010. 30:6:522,526.

THANK YOU