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VEPTR Treatment of Patients with Cerebro-Costo-Mandibular Syndrome (CCMS)

CHRISTUS SANTA ROSA
Spinal and Thoracic Treatment and Research Center

University of Texas Health Science Center at San Antonio (UTHSCSA)
San Antonio, Texas

Children's Hospital of Philadelphia,
Philadelphia, PA

Ajeya Joshi, MD
Robert Campbell, MD
Vishwas Patil, MD
Davin Cordell, MD
James W. Simmons, DO, PhD
Kent Reinker, MD
William Koeck, MD

Introduction

- CCMS is a rare disease
  - First described by Smith et al in 1966
  - AR inheritance pattern with some sporadic cases
  - Approximately 80 cases identified in the literature
- Characteristic findings
  - Variable cerebral impairment
  - Multiple rib malformations (pseudarthroses & hypoplastic chest)
  - Severe micrognathia with or without palatal defects
  - Frequent scoliosis
- High mortality rate due to respiratory insufficiency
  - 40% mortality at one-year; 24/33 at five-years

Background

- Thoracic Insufficiency Syndrome (TIS)
  - Inability of thorax to support normal respiration or lung growth
- CCMS presents with TIS due to:
  - Type 3B volume-depleting deformity (narrow chest)
  - ‘Implosion’ of posterior rib pseudarthroses creates a Jeune syndrome-like chest on cross-section
  - Flail chest due to rib pseudoarthroses
  - Scoliosis that is often rapidly progressive
- VEPTR is established as an effective treatment for TIS
- We hypothesize that treatment with VEPTR may stabilize the flail chest, control the spinal deformity, and thereby address TIS in these patients.

Objectives

1. Describe the surgical technique used with CCMS
2. Assess changes in the respiratory status
3. Quantify changes in spinal and thoracic architecture
4. Identify complications associated with treatment

Materials & Methods

- Retrospective cohort study
- Inclusion criteria: Diagnosis of CCMS & minimum 2 years follow-up
- Exclusion criteria: prior spinal or thoracic surgeries
- Chart review
  - Demographic data: age at initial implant and time in program
  - Operative technique and frequency
  - Respiratory status: Assisted Ventilatory Rating (AVR) scale, respiratory rate, capillary blood gas (CBG)
  - Complications
- Radiographic Assessment
  - Cobb angle
  - Chest width: inside diameter of thorax at T6 in PA radiograph
- Data analyzed using paired *t*-test and Mann-Whitney U test

Operative Techniques

- Bone grafting
  - Used to establish stable rostral and caudal rib anchor points prior to VEPTR implantation
- Implantation
  - Staged bilateral VEPTR rib-to-rib devices
  - Lateral placement to create chest expansion
  - Rib osteotomies dependent on extent of pseudarthroses
  - Medial hybrid rib-to-lamina or pelvis device to address scoliosis
- Replacement
  - Due to maximum expansion, migration, or infection of device
- Expansion
  - Based on clinical and radiographic assessment at 6 month intervals during the course of treatment

Results

- Demographics
  - Six patients met the inclusion criteria; none were excluded
  - 2 males; 4 females
  - Age at first surgery: 49.8 months (range, 8-112)
  - Length of follow-up: 48.3 months (range, 24-91)
- Complications
  - Device migration: 4 (in 2 patients)
    - 3 superior cradle migrations (1 acute); 1 laminar hook migration
    - treated with re-seating (3) or replacement (1)
  - Wound dehiscence: 1
    - no infection; treated at home with dressing changes
  - Deep infection: 1
    - treated with I&D, iv antibiotics, & device removal; device re-implanted 3 months after infection resolved

Results

- Surgical Technique
  - Bone grafting
    - 3/6 patients (total of 6 procedures) using autologous iliac
    - 2 graft recipients required multiple grafting procedures
  - Implantation
    - 6/6 underwent staged bilateral rib-to-rib device implants
    - 4/6 received hybrid rib-to-spine or pelvis devices to address scoliosis
      - 3 rib-to-lamina; 1 rib-to-pelvis (dependent on length of curve)
    - Mean interval between implants: 17.0 months (range, 8-25)
  - Replacements
    - 2/6 underwent a total of 6 replacements
    - 4 for maximum expansion; 1 for migration; 1 for infection
  - Expansions
    - Mean of 13.2 (range, 0-17) during 6.6 procedures (range, 0-9)

Results

- Respiratory Status
  - AVR: (4=full-time mechanical ventilation; 0=no assistance)
    - 2 unchanged (AVR 4 & AVR 3)
    - 4 improved (2 from AVR 4 to AVR 3; 2 from AVR 3 to AVR 2)
      - p > *0.05 using Mann-Whitney U test*
  - Respiratory rate: 36 (pre); 30 (post) ($p > 0.05$)
  - CBG:
    - pO2: 62.8 (pre); 58.6 (post) ($p > 0.05$)
    - pCO2: 41.8 (pre); 38.4 (post) ($p > 0.05$)
- Spinal & Thoracic Architecture
  - Cobb angle: 48 (pre); 49 (post) ($p > 0.05$)
  - Chest width: 117mm (pre); 141mm (post) (p=0.009)

Conclusions

- VEPTR efffectively addresses TIS in CCMS by
  - stabilizing the flail chest
  - reversing the ‘implosion’ deformity and increasing chest width
  - arresting scoliosis progression
- Bone grafting is necessary in some patients to create stable anchor points prior to implantation
- Most patients experienced an improvement in ventilatory status during treatment
- Complications were limited and manageable

References

Campbell RM. Spine deformities in rare congenital conditions. *Spine* (2009) 34(17):1815-27.

Flodmark P, Wattsgard C. Cerebro-costo-mandibular syndrome. *Pediatr Radiol* (2001) 31:36-7.

Ozkinay F, Cogulu O, Gunduz C, Cankaya T, Ozkinay C. Cerebro-costo-mandibular syndrome: a case report. *Pediatr International* (2001) 43:522-4.

Plotz FB, van Essen AJ, Bosschaart AN, Bos AP. Cerebro-costo-mandibular syndrome. *Am J Med Genet* (1996) 62:286-92.

Silverman FN, Strefling AM, Stevenson DK, Lazarus J. Cerebro-costo-mandibular syndrome. *J Pediatr* (1980) 97:406-17.

Tachibana K, Yamamoto Y, Osaki E, Kuroki Y. Cerebro-costo-mandibular syndrome: a case report and review of the literature. *Hum Genet* (1980) 54:283-6.