Evaluation of 18 patients with Caudal Regression Syndrome

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E-Poster:

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Authors Disclosure Information

Introduction

- Caudal regression syndrome (CRS) also referred to as caudal dysplasia, and sacral agenesis (SA) and lumbosacral agenesis syndrome is a rare congenital abnormality in which a segment of the sacrum and/or lumbar spine, and spinal cord fails to develop.
- CRS is an uncommon congenital disorder which occurs in 0.01-0.05 per 1,000 live births.
- CRS is associated with Neurologic, Orthopedic, Gastrointestinal, Genitourinary and Cardiac abnormalities.
- Spinal cord and cervical spine abnormalities, imperforate anus, malformed genitalia, renal dysplasia or aplasia and congenital heart defects are commonly seen.
- Since there is not a true cure, treatment is difficult, multidisciplinary, and largely supportive.

Etiology

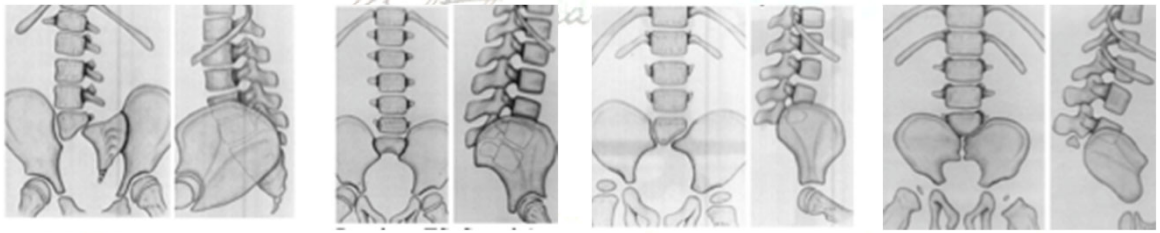
- The etiology of CRS is unknown.
- Maternal diabetes, inherited genetic factors, teratogens, vascular hypoperfusion and failure of early embryonic mechanisms are associated with CRS.
- The syndrome occurs more frequently in the offspring of diabetic mothers (16-50%) than of non diabetic ones.
- Candidate gene mutations have been mapped to 7q36 and T a transcription factor for posterior mesodermal structures.

Diagnosis

- Renshaw classified patients into four types according to the amount of sacrum remaining and to the characteristics of the articulation between the spine and pelvis.

Renshaw Classification (Renshaw, 1978)	
Type I	Partial or total unilateral sacral agenesis
Type II	Partial bilateral, symmetrical sacral agenesis
Type III	Total sacral agenesis with variable lumbar anomaly and iliac wings attached to the last lumbar vertebrae
Type IV	Total sacral agenesis with / lumbar anomaly and iliac wings fused behind the last vertebrae, if they are present.
Mild form	Type I and II. Coccyx agenesis without functional repercussions.
Major form	Type III and IV. Systematic sequelae are present with neurologic impairment. Perinatal death is frequent. Thoracic vertebrae involvement is incompatible with life.

Type I *Type II* *Type III* *Type IV*



Diagnosis

- The classification of CRS of Guille et al considers the absence (Group I) or presence (Group II) of myelomeningocele.
- Three types of spinal deformities in myelomeningocele were described.

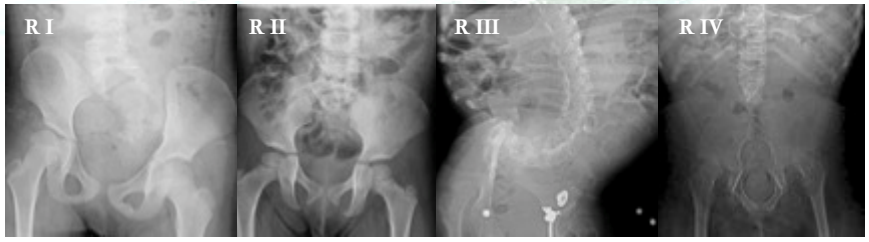
Guille's Classification (Guille et al, 2002)		
Group I	Absence of myelomeningocele.	
Group II	Presence of myelomeningocele.	
	Type A	There is either a slight gap between the ilia or the ilia fused in the midline. One or more lumbar vertebrae were absent. The caudad aspect of the spine articulated with the pelvis in the midline, maintaining its vertical alignment.
	Type B	The ilia are fused together, some of the lumbar vertebrae are absent, and the most caudad lumbar vertebra articulates with one of the ilia, with the most caudad aspect of the spine shifting away from the midline.
	Type C	Total agenesis of the lumbar spine, the ilia are fused together, and there is a visible gap between the most caudad intact thoracic vertebra and the pelvis.

Purpose

- We presented 18 cases of CRS among the consecutive pediatric cases that were diagnosed with congenital spinal column deformities between 2006-2012.
- To evaluate magnetic resonance imaging (MRI) and computed tomography (CT) results, radiological and clinical data of pediatric patients with spinal problems related to CRS.

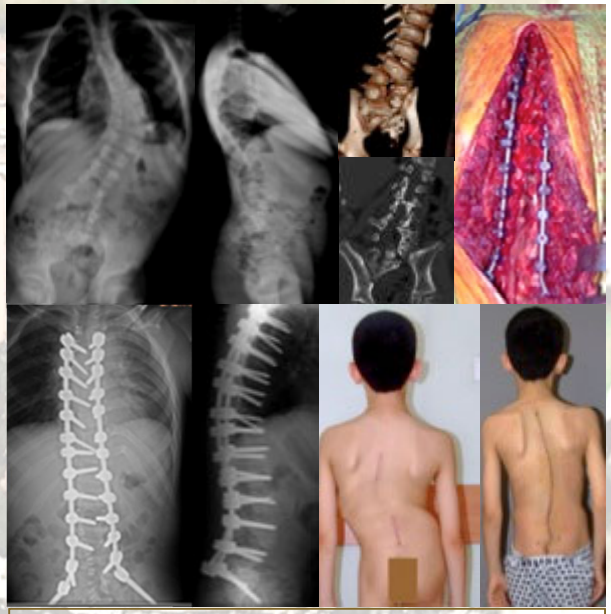
Methods

- Clinical and radiological findings were reviewed to classify each patient into Renshaw's and Guille's classifications.
- Each patient reviewed X-ray, MRI and CT.
- Cardiovascular, urogenital, neurological and genetically abnormality were researched.
- Clinical and radiological findings of these cases along with life time management were outlined and the literature was reviewed.

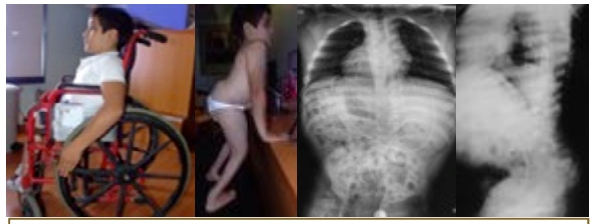


Results

- Our retrospective study allowed us to see the various concomitant conditions which often occur with CRS.
- CRS occurred most often with Spina bifida (55.5%).
- Renshaw classification was seen as type III in 38.8% of the patient and the others were seen 33.3% in type I, 16.6% in type II and 11.1% in IV.
- According to Guille's classification, 8 patients were in Group 1, 10 patients were in Group 2.
- Different type of congenital spinal anomalies were seen associated with CRS.
- One patient was operated for congenital scoliosis associated with SA.
- Others patients were followed up with breys, and needed multidisciplinary approach.
- Some patients operated due to neurosurgical, orthopedic or other organ disorders.

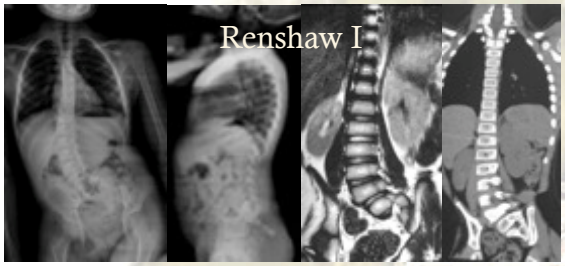


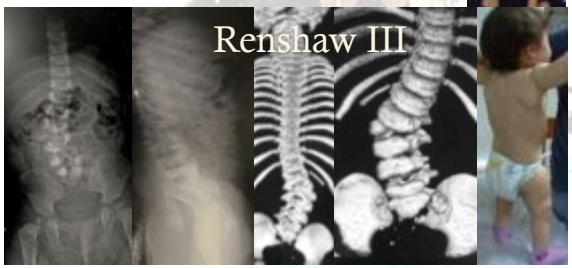
Renshaw I, 11y, M, Lumbar Hemivertebra with SA. Hemivertebra resection and posterior instrumentation was made.

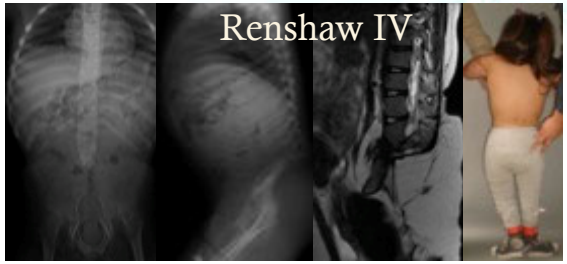


Renshaw III, 10y, M, Lumbosacral agenesis.

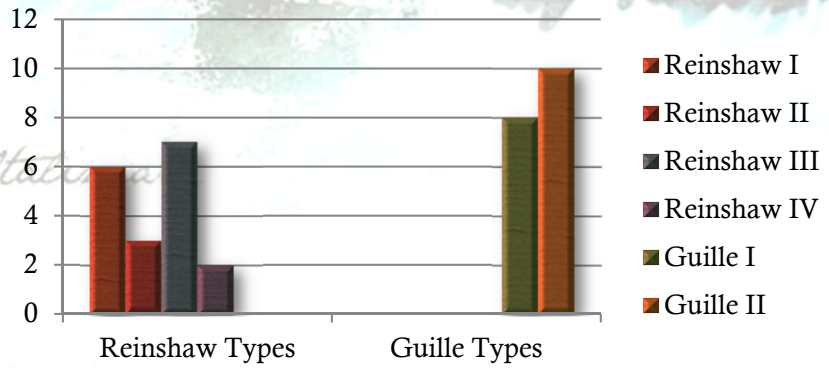
Results







N	Age	Gender	Renshaw Type	Vertebral Anomaly
1	5 y 2 m	M	III	Spina Bifida
2	8 y 3 m	F	I	Congenital Scoliosis
3	6 m	F	III	Spina Bifida
4	13 y 11 m	M	I	Congenital Scoliosis and Kyphosis
5	9 y 10 m	F	I	Spina Bifida, Congenital Scoliosis and Kyphosis
6	4 y 11 m	F	II	Spina Bifida, Congenital Anomalies
7	15 y	F	III	Spina Bifida, Congenital Scoliosis and Kyphosis
8	12 y 9 m	F	II	Spina Bifida, Congenital Scoliosis and Kyphosis
9	7 y 1 m	F	III	Spina Bifida, Congenital Scoliosis and Kyphosis
10	9 y 5 m	F	I	Spina Bifida, Congenital Scoliosis
11	11 y 8 m	M	III	Spina Bifida, Congenital Anomalies
12	6 y 2 m	F	IV	Congenital Vertebral Anomalies
13	9 m	F	IV	Congenital Vertebral Anomalies
14	8 y 7 m	F	I	Spina Bifida, Congenital Scoliosis and Kyphosis
15	18 y	F	II	Congenital Skoliosis and Anomalies
16	13 y 2 m	M	III	Congenital Scoliosis
17	10 y 3 m	F	III	Congenital Scoliosis
18	5 y 2 m	M	I	Congenital Scoliosis
	8.9 y (6 m-18 y)	**13 F 5 M**	**I (6), II (3), III (7), IV (2)**	



Conclusions

- The goal of this study was to show in which variations may CRS patients come into the picture and to provide insight into the CRS and management of this disorder.
- CRS remains associated with structural and systematic problems including genitourinary, gastrointestinal, orthopedic, neurological, respiratory and cardiac anomalies.
- First step of the early management of CRS should be an accurate prenatal diagnosis.
- As a result, our aim was to point out the congenital spinal disorders and especially spina bifida patients associated with CRS.
- CRS need a careful investigation, evaluation, preoperative planning and follow up.
- We need larger studies to determine the utilities of the classifications and to improve them.

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