International Journal of Research in Medical Sciences
Sendhil CA et al. Int J Res Med Sci. 2014 Feb;2(1):344-346
www.msjonline.org
pISSN 2320-6071 | eISSN 2320-6012

DOI: 10.5455/2320-6012.ijrms20140268

Case Report

Caudal regression syndrome: a rare case report

Sendhil Coumary A[1]*, Sunitha V C[2], Vasavi Kolluru[1], Janani Shankar[1]

[1]Department of Obstetrics & Gynaecology, [2]Department of Radiodiagnosis, Mahatma Gandhi Medical College & Research Institute, Pillaiyarkuppam, Cuddalore road, Pondicherry 607402, India

Received: 28 September 2013
Accepted: 10 October 2013

***Correspondence:**
Dr. Sendhil Coumary A,
E-mail: sendhilcoumary1975@yahoo.in

ABSTRACT

Caudal regression syndrome is a rare disorder characterised by abnormal development of structures in the caudal region of the embryo like lower lumbar and sacral vertebrae, urogenital and lower gastrointestinal system. It is secondary to abnormal development of mesoderm. Multiple hypotheses like genetic, metabolic and vascular hypoperfusion have been proposed as etiologies. It can be picked up in early second trimester by ultrasound. It has a higher incidence in diabetic pregnancies.

Keywords: Caudal regression syndrome, Sacral agenesis

INTRODUCTION

Caudal regression syndrome is a rare syndrome characterised by malformations of the structures derived from the caudal region of the embryo like the caudal spine, spinal cord, lower limbs, urogenital system & hindgut.[1] Caudal regression syndrome has an incidence of 1:25000 live births, though it is much more common among overt diabetic mothers.[1]

CASE REPORT

An overt diabetic on irregular treatment with oral hypoglycaemic agents presented to the antenatal clinic. She was a second gravida with irregular cycles with a good obstetric outcome in the previous pregnancy. The present pregnancy was an unplanned one and she was not sure of her dates. On examination she was of normal BMI, not pale with uterus of 16 – 18 weeks size. Her blood investigation showed high blood sugars and her HbA_1C was 5.6. Her routine obstetric USG revealed caudal regression syndrome which included complete lumbosacral agenesis, a flat sheet of bone in midline posteriorly in the lumbosacral region with absent movements of right lower limb (Figure 1 & 2). After a detailed counselling patient wanted to discontinue pregnancy and a medical termination of pregnancy was carried out. She delivered a female fetus weighing 500 grams with placenta weighing 150 grams. Fetus had type four sacral agenesis with the iliac bones fusing in the midline. It also had fixed flexion deformity of bilateral hip joints with genu varum of right knee joint (Figure 3 & 4).

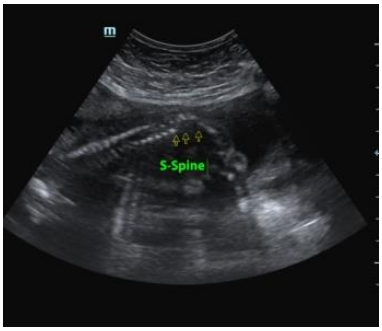

Figure 1: Two dimensional USG image showing the absence of lumbosacral spine.

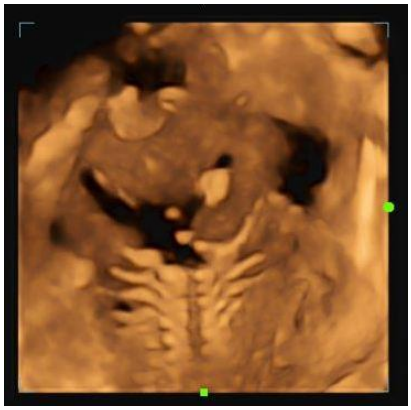

Figure 2: Three dimensional USG image showing the absence of lumbosacral spine.

Figure 3: Picture of fetus showing fixed flexion deformity of right lower limb with a small pelvic girdle.

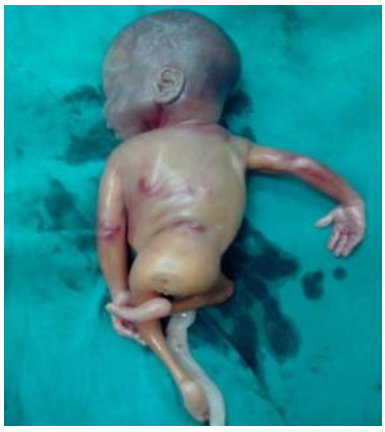

Figure 4: Picture of the posterior aspect of the fetus showing the fusion of iliac bones in the midline.

DISCUSSION

Sacral agenesis is listed as a rare disease by the office of rare disease (ORD) of National institute of health. It is usually sporadic. It occurs at 3rd-7th week of embryonic development during gastrulation.[2] Sacral agenesis results from abnormal development of mesoderm. This disruption of the mesoderm leads to motor and neurologic impairment. Though the etiology is not known various hypotheses have been suggested like interaction of multiple genetic & environmental factors. It is suggested that an abnormal artery in the abdomen diverts the blood flow away from the lower areas which may be the primary cause or this artery itself may be secondary to abnormal mesoderm.[1] Increased blood sugar levels and drugs like retinoic acid[3] have been implicated.

Renshaw[4] classified sacral agenesis into four types:

Type I: Total or partial unilateral sacral agenesis.

Type II: Partial sacral agenesis but bilaterally symmetrical defect and a stable articulation between the ilia and a normal or hypoplastic first sacral vertebra (most common).

Type III: Variable lumbar and total sacral agenesis with the ilia articulating with the sides of the lowest vertebra present.

Type IV: Variable lumbar and a total sacral agenesis, the caudal end-plate of the lowest vertebra resting above either fused ilia or an iliac amphiarthrosis.

Diagnosis in the first trimester may be difficult due to incomplete sacral ossification.[1] Features of caudal regression syndrome range from complete absence of sacrum along with abnormal lumbar spine. There may be associated clubfoot and contraction of the knees and hip. Typically there is absence of a few vertebrae, shield like iliac wings and decreased space between femoral heads. Decreased movement of the leg are often observed. A short crown rump length and abnormal yolk sac have been proposed as early USG features of caudal regression syndrome. It may be associated with abnormalities of the CNS, musculoskeletal, genitourinary, gastrointestinal systems.[1]

Sirenomelia is a common differential diagnosis, which was initially considered to be a severe variant of caudal regression syndrome. Sirenomelia is characterised by fused lower limbs, severe renal anomalies with oligohydramnios and is now considered to be a separate entity. It is frequently associated with VACTERL syndrome.[2] A triad of hemisacrum, anorectal malformation and presacral mass is often described as Currarino syndrome. It is an autosomal dominant sacral agenesis involving S2-S5 only and the underlying gene defect being localised to 7q36. Currarino syndrome has a familial tendency.[5]

Incidence of major fetal malformations is 5-10% among women with pregestational diabetes. Severe malformations result from poor peri conceptional control of blood sugars.[6] In comparison to the general population, the incidence of caudal regression is many hundred folds more common among diabetic mothers. Even a slight

increase in HbA_1C is associated with malformations which mandate a rigorous blood sugar control among diabetic women planning pregnancy. A conscious planning of pregnancy with peri conceptional control of sugars would avoid such malformations, thereby reducing mental and physical trauma to patients.

Prognosis of caudal regression syndrome depends on the severity of spinal defect and associated anomalies. Survivors are mentally normal though it requires extensive urologic and orthopaedic interventions at a tertiary centre. Severe forms associated with cardiac, renal and respiratory problems result in early neonatal death. When caudal regression syndrome is encountered early, pregnancy termination can be offered.[1] If pregnancy is continued standard prenatal care is offered.

REFERENCES

1. Fetal syndromes. Juliana Lete, Roberta Granese, Philippe Jeanty, Sandra Silva Herbst. Ultrasonography in obstetrics and gynaecology, 5th edition, 2008, pg 112 to 180.
2. M. Valenzano, R. Paoletti, A. Rossi, D. Farinini, G. Garlaschi, E. Fulcheri Sirenmelia. Pathological features, antenatal ultrasonographic clues and a review of current embryogenic theories. Human reproduction update 1999, Vol 5, no 1, pp 82 – 86.
3. Padmanabhan R. Retinoic acid-induced caudal regression syndrome in the mouse fetus. Reprod Toxicol. 1998 Mar-Apr;12(2):139-51.
4. Renshaw TS. Sacral Agenesis. The Pediatric Spine - Principles and Practice. 1:2214,1994,Raven Press, New York.
5. Sally Ann Lynch, Yiming Wang, T Strachan, John Burn, Susan Lindsay Autosomal dominant sacral agenesis: Currarino syndrome Review article J Med Genet 2000;37:561–566.
6. Inkster ME, Fahey TP, Donnan PT, Leese GP, Mires GJ, Murphy DJ. The role of modifiable pre-pregnancy risk factors in preventing adverse fetal outcomes among women with type 1 and type 2 diabetes. Acta Obstet Gynecol Scand. 2009;88(10): 1153-7.

DOI: 10.5455/2320-6012.ijrms20140268
Cite this article as: Sendhil CA, Sunitha VC, Vasavi K, Janani S. Caudal regression syndrome: a rare case report. Int J Res Med Sci 2014;2:344-6.