

# Caudal Regression Syndrome: A Case Report

Brian Knight, CRNA, MS

*Caudal regression syndrome is a condition that is present in approximately 1:60,000 births. This syndrome can manifest as an absence of a few terminal coccygeal segments to lumbosacral agenesis. The disease is associated with many other abnormalities that can affect one's anesthetic, including those of the cardiopulmonary, vertebral, and genitourinary systems, and may include VACTERL syndrome (abnormality of vertebrae, anus, cardiovascular system, trachea, esophagus, renal system, and limb buds).*

*This case report describes a neonate with caudal regression syndrome undergoing placement of a diverting colostomy, and it includes a review of the literature regarding this syndrome and its possible causes of development.*

**Keywords:** Caudal regression syndrome, sacral agenesis, VACTERL, VATER.

Caudal regression syndrome (CRS) is a rare, sporadic neural tube defect characterized by incomplete development of terminal spinal segments.<sup>1</sup> Presentation varies from absence of a few terminal coccygeal segments to lumbosacral agenesis and more extensive forms such as sirenomelia (congenital anomaly with fusion of lower extremities).<sup>2</sup> This syndrome manifests as loss of bladder and bowel control along with total neurologic deficit in the lower limbs. Several associated anomalies affecting other systems are frequently present and complicate the clinical picture. The incidence is estimated to be approximately 1:60,000 births, with a male-to-female ratio of 2.7:1.<sup>3</sup> Pediatric patients with CRS often require surgery with general anesthesia, and this may be challenging because of the coexistent anomalies, such as musculoskeletal deformities, that could make intubation difficult.<sup>1</sup> This case report describes a neonate with CRS presenting for a diverting colostomy and is followed by a review of the literature.

## Case Summary

A 2-day-old, 2,621-g, male neonate, born of a nonconjuguous marriage at 38 weeks, presented with an imperforate anus for a diverting colostomy. There was no history of antenatal infections or maternal diabetes, hypertension, or drug consumption. The perinatal history was unremarkable. Echocardiogram findings were reported to be within normal limits. An ultrasound examination of the kidneys found them to be within normal limits. X-ray films of the pelvis showed sacral agenesis. Results of routine laboratory investigations were noncontributory.

Physical assessment of the patient revealed a normocephalic neonate with an imperforate anus. The patient was classified as nothing by mouth (NPO) for life and maintained on total parenteral nutrition therapy along with 10% dextrose with electrolytes at 80 mL/kg per day. The

remaining findings of the physical assessment were unremarkable. Preoperative teaching about the procedure, postoperative care, and risks of surgery were discussed with the patient's family by the anesthesiologist and pediatric surgeon. An informed consent was obtained from his mother.

The patient was transported directly from the neonatal intensive care unit (NICU) to the operating room. Once in the operating room, the patient was connected to standard ASA monitors and preoxygenated for 5 minutes. Once preoxygenated, an inhalation induction was started using 8% sevoflurane until the patient was through stage 2 of anesthesia. Sevoflurane was then supplemented by propofol, 14 mg intravenously (IV), and rocuronium, 3.5 mg IV. The patient's eyes were taped to prevent corneal abrasion. A direct laryngoscopy was then performed using a Miller laryngoscope blade size 0, and a grade 1 view of the vocal cords was achieved. A 3.5-cm uncuffed endotracheal tube (ETT) with a stylet was placed without difficulty through the vocal cords. Confirmation of ETT placement was verified via positive end-tidal carbon dioxide tracing and bilateral breath sounds. Immediately on confirmation of ETT placement, the ETT was secured with tape and the patient's anesthesia was maintained with sevoflurane.

The intraoperative period was uneventful. On completion of the surgical procedure, the patient remained intubated and was transported back to the NICU with full monitors and ventilated via a bag valve mask (Ambu bag, Ambu Inc, Glen Burnie, Maryland). In the NICU, the patient recovered without complications and was extubated the following day.

## Discussion

Caudal regression syndrome is characterized by developmental anomalies of the neural tube, caudal vertebrae, urogenital and digestive organs, and hind limbs, the precursors of which are derived from the caudal emi-

nence. The severity varies from absence of the coccyx to lumbosacral agenesis, which is what the neonate in this case had.<sup>2</sup> Associations reported with CRS mostly include genitourinary, anorectal, vertebral, and cardiopulmonary anomalies and may also include VACTERL syndrome (abnormality of vertebrae, anus, cardiovascular system, trachea, esophagus, renal system, and limb buds). Other associations include omphalocele, imperforate anus, bladder exstrophy, spinal anomalies, and Currarino triad syndromic complexes.<sup>3</sup> Therefore, an anesthesia assessment should focus on the possibility of any of these abnormalities, and a plan of care should be individualized based on the coexisting conditions.

Caudal regression syndrome is a consequence of abnormal development of the structures derived from the caudal mesoderm of the embryo before the fourth week of gestation.<sup>3</sup> This anomaly is not thought to be hereditary, and the recurrent risk is very small. It is due to a defect in the *HLBX9* homeobox gene, which is found on chromosome 7q36. This gene is also expressed in the pancreas. Therefore, a possible association between diabetic hyperglycemia and caudal regression is proposed. Insults that may lead to CRS include toxins, hyperglycemia, infections, retinoic acid, and ischemia before the fourth gestational week. Approximately 15% to 20% of the patients have diabetic mothers, and almost 1% of children of diabetic mothers are affected by this disorder. However, because it has been found that only 16% to 22% of the mothers of these patients have diabetes, it is clear that the syndrome is not specific to diabetes.<sup>2</sup> Furthermore, a recent experimental study looked at the role of retinoic acid in producing CRS in mouse fetuses. Retinoic acid, when given in variable dosages to the mouse fetuses, resulted in CRS in most of the survivors.<sup>4</sup> This study along with others like it, strongly suggest the teratogenic role of various chemicals in the genesis of CRS.

Because the primary pathology is irreversible, the

treatment is only supportive, with an aim to achieve as much normalcy as possible. Survival is the rule if the vital systems are unaffected or minimally affected. Each system and its pathology are treated on their own merits; therefore, multiple surgeries may be required. The abnormalities that need special attention are bladder and bowel incontinence, orthopedic deformities, and renal dysfunction. Correction of these abnormalities helps the child to be independent with a better quality of life. These patients have normal intelligence and therefore lead otherwise normal lives except for neuromuscular deficits of the lower limbs and sphincters.<sup>3</sup>

In conclusion, because of the numerous surgeries that patients with CRS will require, anesthesia providers can expect to encounter CRS sometime in their anesthesia career. One must determine the severity of the anomalies that can accompany CRS, which can include the neuroskeletal, cardiac, and renal systems. Tracheal and esophageal abnormalities may also exist, which make intubation difficult or impossible. Therefore, anesthesia in newborns with CRS may be challenging because of a variety of associated anomalies that providers should be prepared to encounter.

## REFERENCES

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## AUTHOR

Brian Knight, CRNA, MS, is a staff nurse anesthetist at Jackson County Memorial Hospital, Altus, Oklahoma. At the time this paper was written, he was a student at Midwestern University, Nurse Anesthesia Program, Glendale, Arizona. Email: bknight33@hotmail.com.