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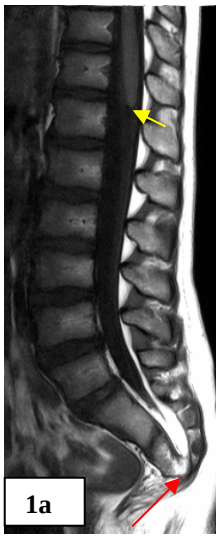
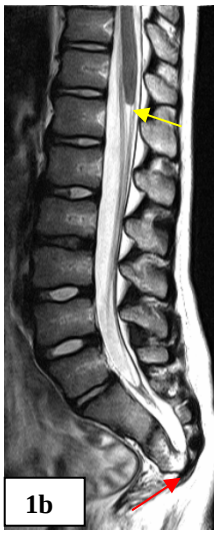
ADVANCED IMAGING CENTER

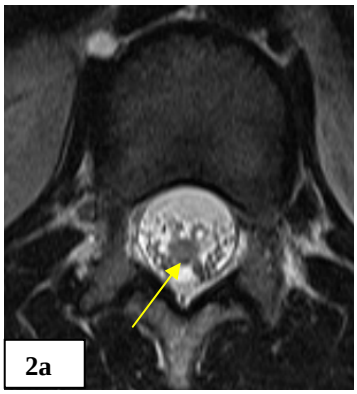
PHYSICIAN NEWS

August 28, 2001

INTERESTING CASE PRESENTATION
Caudal Regression Syndrome (Sacral agenesis)







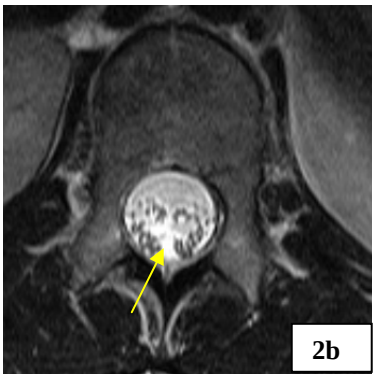


Fig. 1a-b are midline sagittal T1 and T2 weighted images. Fig. 2a-b are successive axial T2 weighted images through the conus.

CLINICAL INFORMATION: The patient is a 12-year-old male who was seen by **Dr. Gerald Edralin***. Patient has a history of colostomy until age two due to anal atresia. He has suffered from bowel incontinence and presents with left lower abdominal pain. Patient was referred to AIC for an MRI of the L-spine to R/O possible spinal dysraphism.

MRI FINDINGS: The MRI was performed on AIC's high-field 1.5 Tesla short-bore Siemens Symphony. They demonstrate partial absence of distal sacrum (red arrows) and abrupt truncation of the tip of the conus, the so-called "**filum terminus**" (yellow arrows). No tethered cord is noted. The above findings are consistent with caudal regression syndrome (also known as sacral agenesis).

DISCUSSION: This is a rare congenital midline closure defect of neural tube, characterized by partial or total absence of the sacrum and defects of variable portions of L-spine and other associated anomalies. **Prevalence:** 0.1-0.25:10,000 in normal pregnancies and 200-250 times higher in diabetic pregnancies. **Etiology:** Unknown, but associated with **maternal diabetes** in 16% of the affected. **Treatment:** Managing renal, cardiac, pulmonary and GI function, preventing renal infection, and achieving continence. Orthopedic surgery in correcting associated malformations. Physical therapy in preventing secondary deformities, skin ulcers, and assisting in achieving some essential functions to improve the quality of life such as ambulating. **Prognosis:** Depending on severity, surviving infants usually have a normal mental function, but require extensive urologic and orthopedic assistance. Their long-term morbidity consists mostly of neurogenic bladder dysfunction resulting in progressive renal damage and disabling neuromuscular deficits.

ASSOCIATED ANOMALIES: **Musculoskeletal:** hip dislocation, foot deformities, hypoplasia of extremities, flexion contractures of hips & knees, pelvic deformity, kyphoscoliosis, absence of ribs, spina bifida, sacral absence; **GI:** anorectal atresia, hernias, abdominal wall defects, gut malrotation; **Fistulas:** rectovaginal, rectourethral, tracheo-esophageal (should be distinguished from VACTER syndrome); **GU:** Lack of bladder/bowel control, neurogenic bladder, vesicoureteral reflux, hydronephrosis, fused kidneys, renal agenesis, ectopic ureters, transposition of external genitalia, mullerian duct agenesis; **Others:** neural tube defects, congenital heart defects, midline facial cleft.

For more information, please call Dr. Edralin or myself at the numbers below.

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