Currarino Syndrome

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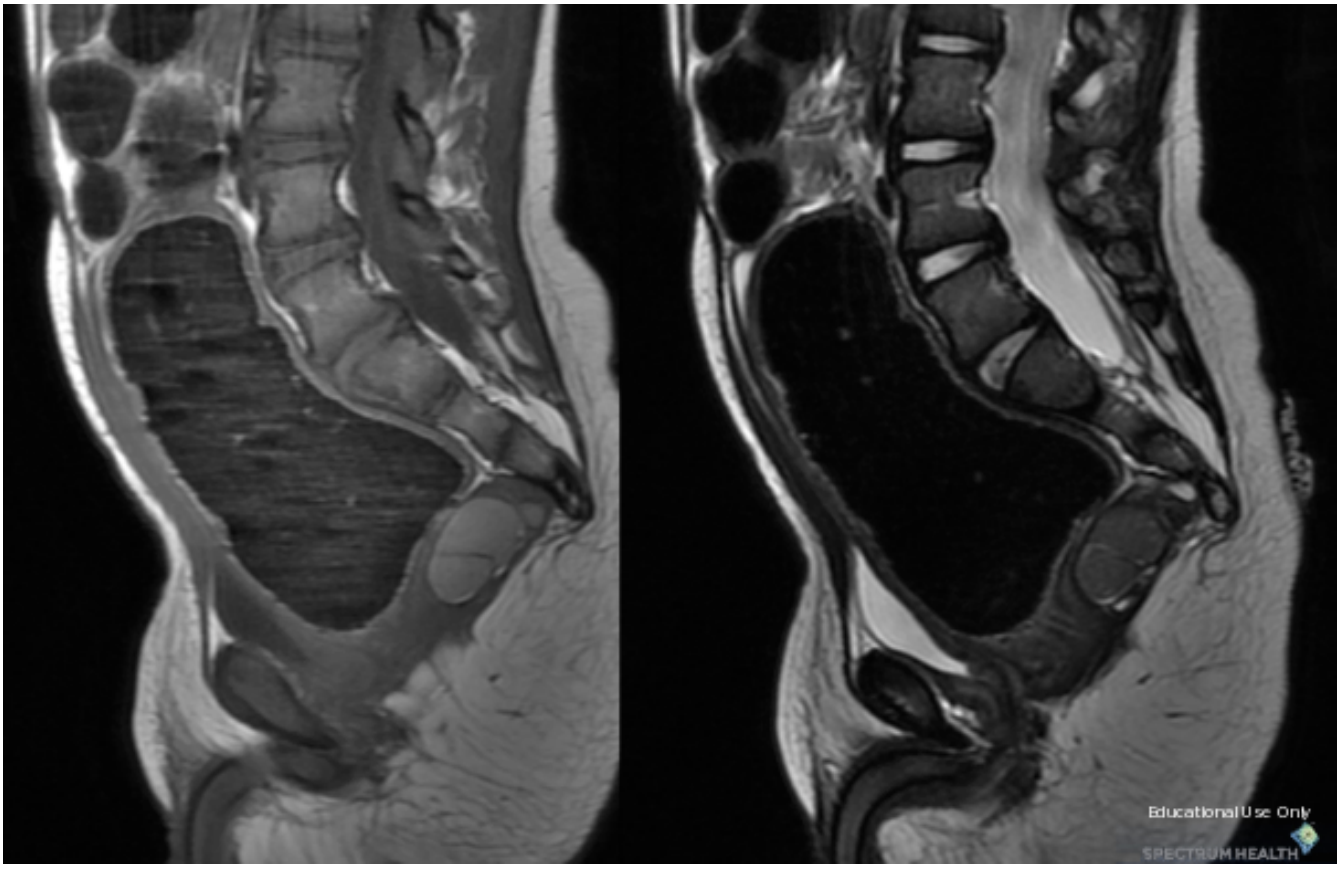
History
6 year old male with constipation

Diagnosis
Currarino syndrome

Discussion
In 1838, Bryant described a case of deficiency of the anterior sacrum with a thecal sac in the pelvis, similar to the tumour of spina bifida. In 1981, Currarino noted a triad of anorectal stenosis, sacral agenesis and presacral mass (anterior meningocele, enteric cyst or teratoma). In 1983 Yates reported a familial tendency of this association. Subsequently, this autosomal dominant sacral dysgenesis was linked to the homeobox gene mutation (HLXB9) on the long arm of chromosome 7-all patients with familial and 30% of sporadic cases show this mutation. The function of HLXB9 is not known but probably a transcription factor. Currarino hypothesized that an adhesion between the endoderm and ectoderm results in a notochord split and sacral deformity and neuro-enteric persistence. All patients have a normal S1 and abnormal S2-S5 (scimitar -75%, bifid with central defect -22% or pepper pot sacrum and coccygeal defect -<3%. All 1st degree relatives should undergo sacrum radiography; if XR is abnormal MR of the lumbosacral spine and pelvis should be performed.

Findings
CR-Deficienct distal right hemisacrum.
MR-Cystic pre sacral mass displacing the rectum anteriorly.

Reference
Kochling J, Pistor G, Marzhausen BS, et al. The Currarino syndrome—hereditary transmitted syndrome of anorectal, sacral and presacral anomalies. Case report and review of the literature. Eur J Pediatr Surg. 1996 Apr;6(2):114-9.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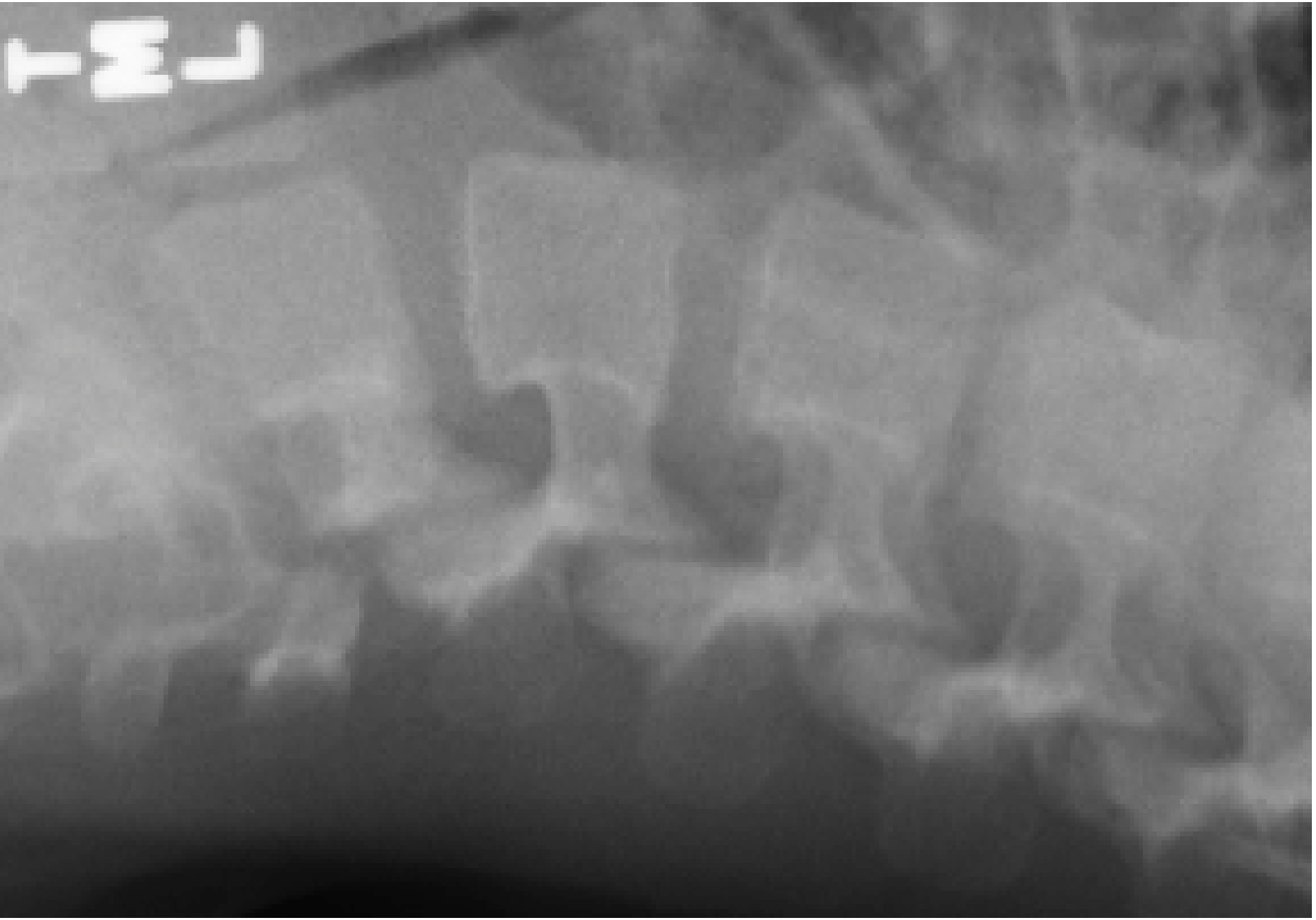

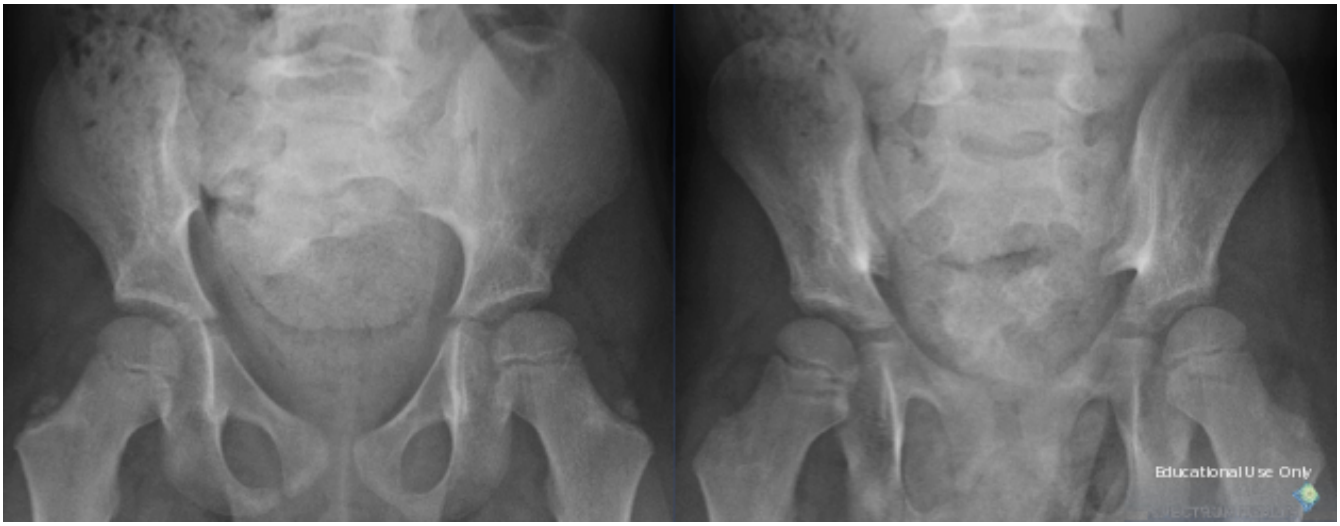
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