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ORIGINAL REPORT

Radiological findings in Currarino syndrome☆

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KEYWORDS
Sacral agenesis;
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Abstract
Objective: To describe the clinical, radiological and genetic findings of a family affected by Currarino syndrome (CS) (agenesis of the sacrum, presacral mass, and anal-rectal anomalies), and to familiarize the radiologist with this condition that, although uncommon, could be suspected by its characteristic images.
Materials and methods: A study was made of 8 out of 9 family members (the parents, 7 siblings, 4 males and 3 females) suspected of having CS. The clinical and genetic findings are described. Using simple X-rays, ultrasound and magnetic resonance imaging, the presence of agenesis of the sacrum, a presacral mass and anal-rectal anomalies were investigated. Furthermore, a genetic analysis of the HLBX9 gene was performed. Permission by the Ethics Committee was not requested as all the family members gave their consent by signing a document.
Results: The mother with a scimitar-shaped sacrum confirmed that she was the transmitter of the genetic mutation. One of the seven siblings had complete CS (sacral agenesis, anorectal stenosis, and anterior meningocele). Four siblings had an incomplete CS: 3 with sacral agenesis and a presacral mass (two anterior meningoceles and one teratoma) and the fourth with sacral agenesis and anorectal stenosis. One sibling had no anomalies. The mother, as well as four siblings, did not have the HLXB9 gene mutation.
Conclusion: When there is sacral agenesis, the possibility of presacral masses and anorectal changes should be investigated. Likewise, if there is familial association, they should be investigated for a CS.

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PALABRAS CLAVE
Agenesia sacra;
Masa presacra;
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Triada Currarino

Hallazgos radiológicos en el síndrome de Currarino

Resumen
Objetivo: Describir los hallazgos clínicos, radiológicos y genéticos de una familia afecta de síndrome de Currarino (SC) (agenesia del sacro, masa presacra y anomalías anorrectales) y familiarizar al radiólogo con esta entidad que, aunque infrecuente, podemos sospechar por sus imágenes características.
Material y métodos: Se estudiaron 8 de los 9 miembros de la familia con sospecha de SC: los padres y 7 hermanos (4 varones y tres mujeres). Se detallaron los hallazgos clínicos y genéticos; y mediante radiografía simple, ecografía y resonancia magnética se investigó la agenesia del sacro y la presencia de masas presacras y anomalías anorrectales. Además, se realizó un análisis del gen HLBX9. No se solicitó el permiso al comité de ética aunque todos los miembros de la familia dieron su consentimiento.
Resultados: La madre con un sacro en cimitarra confirmado era la transmisora de la mutación genética. Uno de los 7 hermanos era un SC completo (agenesia sacra, estenosis anorrectal y meningocele anterior). Cuatro hermanos presentaron un SC incompleto, tres con agenesia del sacro y masa presacra (dos meningoceles anteriores y un teratoma) y el cuarto una agenesia sacra y estenosis anorrectal. Un hermano no tenía alteraciones. Tanto la madre como 4 hermanos presentaban la mutación en el gen HLXB9.
Conclusión: Ante una agenesia sacra se deberían investigar posibles masas presacras y alteraciones anorrectales. Así mismo, en caso de asociación familiar habría que descartar un SC.

Introduction

The Currarino syndrome (CS) is a rare autosomal dominant disorder characterized by a triad of anorectal stenosis, sacral agenesis and presacral mass.[1] It was first described by Kennedy in 1926, but in 1981 Currarino established the ''scimitar sacrum'' as the defect required for CS and recognized these disorders as a complex syndrome that can be ascribed to a common developmental anomaly.[1] The prevalence of the CS is unknown.[2] In the general population, the prevalence of total or partial sacral agenesis is estimated to be 0.09–0.43%.[3] The first case of CS was described in 1926 and up to 2005 less than 250 cases of complete or incomplete syndrome have been reported in the literature.[2,4] In 2000, genetic studies revealed that the locus involved in normal anorectal and sacral development is located in chromosome 7 (7q 36).[5] Mutations within the locus of the HLXB9 gene,[2,5,6] involved in the development of anterior motor neurons, is responsible for CS. The objective of this study is to describe the clinical, radiological and genetic findings in CS and familiarize the radiologist with this entity that, albeit uncommon, can be suspected by the characteristic imaging findings.

Materials and methods

From the index case who gave rise to the suspicion of CS, all the first-degree relatives were invited for CS screening. They were the mother and seven siblings (3 women and 4 men) with ages ranging from 30 to 65 years. All the participants underwent an evaluation that involved a detailed clinical history, plain X-ray of the pelvis (AP projection) (Axiom Aristos FX plus Siemens), abdominal ultrasound (Acuson Antares Siemens), and an MRI (magnetic resonance imaging) scan of the pelvis and lumbosacral spine (Symphony Siemens 1.5) with the following sequence protocol: axial and sagittal T1-weighted sequences, with and without gadolinium with fat saturation; axial and sagittal T2-weighted sequences with fat saturation. Pelvic X-ray was selected as the screening method for sacral agenesis.[6] Pelvis and lumbar spine MRI is the technique of choice for the detection and characterization of presacral masses.[6,7] The different types of sacral agenesis have been described and classified into the following categories[5]: (1) total sacral agenesis with normal or short transverse pelvic diameter and some lumbar vertebrae possibly missing; (2) total sacral agenesis without involvement of lumbar vertebrae; (3) partial sacral agenesis with intact S1; (4) hemisacrum; (5) coccygeal agenesis. Anterior meningocele is defined as the herniation of the dural sac through a defect in the sacrum, and sacrococcygeal teratoma is a germ cell tumor that contains tissue from all three germinal layers.[8] Additionally, the index case underwent an abdominal computed tomography (CT) with iv contrast agent (LightSpeed General Electric) when diagnosed in the emergency department. Lastly, to evaluate the genotype–phenotype correlation of CS, given the variable phenotype expression described in the syndrome, molecular testing, involving bi-directional sequencing of the HLXB9 gene and 50 pb of the flanking region of the gene, was performed on an ABI3130×l automatic sequencer.[9] Ethics Committee approval was not sought, but all family members gave written consent to genetic testing and verbal consent to imaging examinations.

Results

Eight out of nine members of the family with suspected CS (the parents and 7 siblings, 4 males and 3 females) were