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REFERENCES AND DIAGNOSES

MODERATOR: E. Tessa Hedley-Whyte, M.D.

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Case 2005-6

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Diagnosis: Currarino triad with concurrent terminal myelocystocele of filum terminale and sacral hamartoma

Comment: The Currarino triad is a syndrome complex defined by the combination of an ano-rectal malformation with a sacral defect and a presacral mass. Although in 60% of case reports the presacral mass has been described as a benign teratoma, in the case presented, Dr. Johnson is of the opinion that the mass represents a hamartoma associated with a myelocystocele of the filum terminale. In familial cases of Currarino triad, mutations in the homeobox gene HLXB9 located at the terminal portion of chromosome 7q have been reported, suggesting that there is a migrational defect in the pathogenesis of this disorder.

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