A late diagnosis of MURCS association MRKH syndrome with possible familial cluster

Baxter L.
University of Notre Dame Fremantle, Fremantle, WA, Australia

**Definition**

Mullerian agenesis, renal dysgenesis /agenesis, cervicothoracic somite association (MURCS) (type 2, B or atypical Mayer-Rokitansky-Küster-Hauser syndrome (MRKH)) is a rare subset of MRKH syndrome with an incidence of 1-9/100 000 in the general population (1) amongst other congenital malformations(2). And whilst no causative gene locus has been identified current evidence is that autosomal dominant inheritance with incomplete penetrance and highly variable expressivity plays a role in some instances (3). Likewise some have suggested a relationship between azospermia, renal abnormalities, cervical stomatitis (ARCS) syndrome in men, however none has been proven(4).

**Clinical Presentation**

Patient presented as a 36-year-old P1G1 in a private teaching hospital respiratory ward for management of an infective exacerbation of bronchiectasis. Secondary sexual characteristics were normal with onset menarche at age 13. The patient’s pregnancy was obtained via IVF in order to overcome a bicorniculate uterus and associated tubal abnormalities. At age 16 the patient had a Harrington rod inserted to partially correct severe thoracic scoliosis. It was at this time that it was discovered the patient suffered from unilateral renal agenesis with normally placed left kidney. On auscultation and confirmed by trans-esophageal echocardiogram a moderate degree of mitral regurgitation was present. There were no abnormalities of the ears, face or upper limbs and hearing was normal. On questioning she has a family history of congenital deafness and unilateral renal agenesis and two paternal aunties known to be infertile.

**Discussion**

This case demonstrates a rare presentation of type 2 MRKH syndrome exemplified by both the presence of respiratory hypoplasia and mitral regurgitation and MRKH malformations. Likewise the absence of stereotypical features and the scattered presentation to varying specialty practices greatly delayed the diagnosis. Whilst not confirmed genotypically the family history is strongly suggestive of a ARCS like syndrome and other occurrences of MRKH possibly supporting a link between the two syndromes which has been proposed elsewhere and lends weight to the suggestion that GRES (Genital Renal Ear Skeletal) syndrome is a more appropriate nomenclature when dealing with both sexes in a familial setting.

**Conclusion**

Cases of MURCS association MRKH syndrome whilst not uncommon, rarely present with the wide ranging cardiovascular and respiratory involvement seen in this case. Likewise in cases of MURCS the investigation of male relations to the proband should always be considered in light of the possible association between ARCS and MURCS and the possibility for familial clustering and hopeful further elution of the genetic underpinning of the syndrome.

**References**