

What makes women women? - A case of discordant monozygotic twins with Mayer-Rokitansky-Kuster-Hauser syndrome

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We report on an 18-year woman with typical Mayer-Rokitansky-Kuster-Hauser syndrome (MRKH syndrome). On physical examination, the patient had Tanner stage IV breast development and escutcheon. The vagina was absent, and there was a small dimple between the urethra and anus. Rectovaginal examination revealed no uterus. MRKH syndrome was diagnosed by laparoscopy and pelvic and abdominal MRI. They revealed müllerian dysgenesis involving congenital absence of the vagina and a rudimentary uterus. Other studies revealed a 46,XX karyotype, normal ovarian function and normal secondary sexual characteristics, but any renal or skeletal congenital abnormalities were not detected. Follicle-stimulating hormone, luteinizing hormone and prolactin levels were all normal. This patient had a monozygous twin who had normal müllerian/mesonephric development without any abnormalities of the urinary tract, kidney or skeleton. The patient will have a sigmoid colpopoiesis this summer that will allow her a normal sexual life. Furthermore the patient could have her own offspring with the aid of surrogacy. The etiology of this syndrome remains obscure. If its etiology is genetic one, her future daughters may have a risk for the same disorder. We review 6 other discordant monozygotic twins with MRKH syndrome, but could not find any concordant cases in the literature. The presence of discordant cases and the absence of concordant pairs make a genetic etiology unlikely. In this study we analyzed the WNT4, DAX-1, Sox9 genes for mutations that may impair the development and maintenance of the female genitalia.