The Editor’s Workshop

Mayer-Rokitansky-Kuster-Hauser (MRKH) Syndrome: It’s More Than the Anatomy

How do you counsel a patient with vaginal agenesis? The scenario is something like this. You complete your evaluation and the concerned parent says, “Does that mean my daughter will never have children?” Well, then we move to “she can have her own genetic child” through assisted reproduction—in vitro fertilization with a surrogate carrier. So what are the options at this point in time is the next query, and the story goes on from there. Well, we need to focus on MRKH syndrome and prepare you to address the various facets of the clinical challenge.

We begin with the genetics: “polygenic/multifactorial mode of inheritance.”¹ In the study provided by Elias and co-workers it was noted that one of 37 (2.7%) of female sibs over age 16 appeared to have a symptomatic uterine anomaly. The overall prevalence of incomplete müllerian fusion defects is 0.5–4.5%. Management is a challenge. For instance: are you aware of the apparent association of hearing loss with the Connexin 26 gene, demonstrated by genomic DNA isolation and amplification polymerase chain reaction?²

Vertical fusion defects account for imperforate hymen and transverse vaginal septum, both of which can be “obstructive” or “non-obstructive.” The problem of lateral fusion defects brings us to the categories of obstruction and non-obstruction. Didelphic, bicornuate, and unicornuate uteri fall into this clinical categorization.

Then we focus on the “subsets,” such as Goldenhar syndrome and müllerian duct/renal agenesis/cervicothoracic somite dysplasia (MURCS). Perhaps you have seen a patient with CHARGE or VATER syndromes. Brady et al draw attention to new autosomal recessive syndromes which indeed may be associated with müllerian anomalies.³

Next we must focus on the psychosocial aspects of MRKH syndrome. For this we call upon Bean and coworkers at the MAGIC Foundation in Chicago and the State University of New York at Buffalo in this issue of the Journal.⁴ Background to this approach, including surgical and/or vaginal dilators, is a 83% “functional satisfaction” with the end result when “combined with psychosexual rehabilitation.”⁵ We have learned that creation of a neovagina per se does not equal successful psychological outcome. Timing and cooperation with the adolescent is of paramount importance. We must take the time to address the challenge of psychological well-being for our patients, especially with MRKH syndrome. Quality of life reigns supreme. Bean et al proceed with an in-depth review of the databases related to vaginal agenesis. Successful coitus, orgasm, the problem of the “short vagina”—they take us through the best means of addressing partner relationships, infertility, parenthood options, “spirituality,” and give us the basic tenets of psychological evaluation and support. Take note of the “Recommendations” and Quality of Life information. We can address the whole gamut of clinical concerns with MRKH syndrome now that we have the excellent contribution of Bean et al.⁴

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References