Revisiting the significance of proper imaging in Müllerian ducts anomalies

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The Müllerian or paramesonephric ducts are the primordia of the female reproductive tract (FRT). Their development and differentiation give rise to the fallopian tubes, uterus, cervix, and the upper vagina. Properly developed FRT plays an important role in promoting successful process of fertilization and pregnancy (1).

The Müllerian ducts are made of the epithelial lumen surrounded by a mesenchymal layer. Early development of the ducts is marked by thickening of the anterior mesonephric coelomic epithelium, forming a placode of two distinct progenitor cells, which undergo partial epithelial-mesenchymal transition to form immature Müllerian luminal cells and Müllerian mesenchymal cells. After specification, these cells eventually invaginate and proliferate to form the Müllerian ducts regardless of genotypic sex (2). In males, Müllerian ducts undergo regression under the influence of the anti-Müllerian hormone (AMH) produced by the Sertoli cells in the testes, preventing the development of Müllerian-derived organs (3).

Developmental defects in the formation of the Müllerian ducts, also known as the Müllerian duct anomalies (MDA) could result in failure of development of Müllerian-derived organs. The prevalence of Müllerian duct anomalies in the general population has been estimated to be between 1–7%. Patients with MDA have higher risks of infertility, recurrent miscarriages, fetal intrauterine growth restriction, fetal malposition, preterm birth and retained placenta. The American Society of Reproductive Medicine classifies the MDA into seven classes based on the embryology of the Müllerian system and the severity (4). In class I, for example, uterine agenesis and hypoplasia are due to early developmental dysfunction of the Müllerian ducts during early development with either no identifiable uterus or identification of solely rudimentary tissue. Patients may present with primary amenorrhea with normal secondary sex characteristics during puberty due to properly functioning ovaries. This anomaly has no reproductive potential. Alternatively, in class VII, the uterine abnormalities include hypoplasia and a T-shaped uterine cavity. Patients may also present with stenoses of the cervix. The Mayer-Rokitansky-Küster-Hauser syndrome (MRKH), an autosomal dominant condition occurring in 1 in 4500 female livebirths is defined as agenesis of the uterus, cervix and upper 1/3 of the vagina and has been described well in literature as a form of MDA (5).

Imaging studies, including hysterosalpingogram (HSG), ultrasonography (US), magnetic resonance imaging (MRI), or a combination have been used to aid in diagnosis. MRI has been considered the gold standard. MRI is noninvasive, uses non-ionizing radiation, has multiplanar ability, can interrogate greater field, and has excellent soft-tissue characterization (6). Use of MRI for evaluation of MDAs can better guide management decisions because it is able to distinguish between anomalies that are surgically corrigible from the inoperable ones besides picking other associated anomalies such as unilateral renal agenesis (7). MRI is therefore an important tool that can spare patients from unnecessary surgery and to plan for potential pregnancy-associated complications in those able to carry pregnancies.

References

1. Chandler TM, Machan LS, Cooperberg PL, Harris AC, Chang SD. Mullerian duct anomalies: from diagnosis to intervention. *Br J Radiol*. 2009;82(984):1034-1042. doi:10.1259/bjr/99354802

2. Mullen RD, Behringer RR. Molecular genetics of Müllerian duct formation, regression and differentiation. *Sex Dev*. 2014;8(5):281-296. doi:10.1159/000364935

3. Roly ZY, Backhouse B, Cutting A, et al. The cell biology and molecular genetics of Müllerian duct development. *Wiley Interdiscip Rev Dev Biol*. 2018;7(3):e310. doi:10.1002/wdev.310

4. The American Fertility Society classifications of adnexal adhesions, distal tubal occlusion, tubal occlusion secondary to tubal ligation, tubal pregnancies, müllerian anomalies and intrauterine adhesions. *Fertil Steril*. 1988;49(6):944-955. doi:10.1016/s0015-0282(16)59942-7

5. Morcel K, Camborieux L; Programme de Recherches sur les Aplasies Müllériennes, Guerrier D. Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome. *Orphanet J Rare Dis*. 2007;2:13. Published 2007 Mar 14. doi:10.1186/1750-1172-2-13

6. Behr SC, Courtier JL, Qayyum A. Imaging of müllerian duct anomalies. *Radiographics*. 2012;32(6):E233-E250. doi:10.1148/rg.326125515

7. Maciel C, Bharwani N, Kubik-Huch RA, et al. MRI of female genital tract congenital anomalies: European Society of Urogenital Radiology (ESUR) guidelines. *Eur Radiol*. 2020;30(8):4272-4283. doi:10.1007/s00330-020-06750-8