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Complete androgen insensitivity syndrome: An anatomic evaluation and sexual function questionnaire pilot study

Jason M. Wilson*, Anne Arnhym, Angie Champeau, Michele Ebbers, Fergus Coakley, Laurence Baskin

Urology, UCSF, San Francisco, CA 94143-0738, USA

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Abstract *Purpose:* To further characterize the anatomy and sexual function of women with CAIS compared to normal females, and assess the utility of magnetic resonance imaging (MRI) to distinguish anatomical differences.
Materials and methods: In a prospective cohort pilot study, five individuals with androgen insensitivity syndrome and six, normal, nulliparous women underwent an interview, physical examination, questionnaire completion and MRI of the pelvis. Statistical analysis was performed with emphasis on determining significant differences in anatomical findings and sexual satisfaction.
Results: MRI demonstrated statistically significant differences in vaginal depth and size that were not confirmed on physical exam. MRI and physical exam demonstrated a non-significant difference in average phallic thickness between the two groups, although the CAIS group clitoral width tended to be smaller. Physical exam demonstrated a higher average erect height and longer arm span in the CAIS patients but this was not statistically significant. No significant differences were noted in categories designed to assess satisfaction with ability to achieve orgasm, vaginal appearance and frequency of sexual intercourse between the two groups.
Conclusions: The women with CAIS were as satisfied with sexual function as were the women within the control group. Physical exam and MRI did not find any statistically significant clinically relevant differences between the two groups.

* Corresponding author. University of New Mexico, MSC 10 5610, Albuquerque, New Mexico 87131
E-mail address: jmwilson@salud.unm.edu (J.M. Wilson).

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Our clinical experience suggests that individuals with CAIS have a unique phenotype, differing from 46, XX females and 46, XY males. Other groups have also noted distinctive morphologic traits in patients with CAIS, including tall stature, atypical axillary and pubic hair patterns, and small clitoral and vaginal sizes [1–3]. It has been suggested that vaginal hypoplasia and/or anxiety arising from the self-awareness of anatomic differences may lead to altered psychosexual or sexual function, including genital perception, sexual frequency, problems with vaginal penetration and perception of compromised womanhood [3,4].

A murine homologue of CAIS has further illuminated the concept of a unique phenotype, and challenges the belief that androgen in the presence of a functional androgen receptor portends male genital development and the derangement of the androgen/receptor interaction will result in female genital development. High resolution histologic comparisons of the genital tubercle of wild-type male, wild-type female and androgen receptor knock-out (ARKO) mice have shown that the ARKO mouse genital tubercle has a unique phenotype. More specifically, the ARKO mouse exhibits a more generous corpora spongiosum and smaller glans and corporal bodies [5]. Reproduction of this type of histological analysis in humans is not possible.

In this pilot study, we further characterized the anatomy and sexual function in humans with CAIS compared to controls, using magnetic resonance imaging (MRI), physical exam and validated sexual function questionnaires.

Materials and methods

In this nonrandomized prospective cohort study, after institutional review board approval, patients with CAIS were invited to participate in an MRI examination of the pelvis, history and physical, questionnaire and interview. The same invitation was extended to the public within and around the community of UCSF, attempting to identify an age-matched, nulliparous control group. Informed consent was obtained from each individual participating in the study.

An MRI protocol was developed to demonstrate pelvic anatomy using high resolution, tri-planar, fast spin-echo weighted images through the pelvis, including the vagina, urethra and clitoris. Several anatomic features were analyzed to ascertain vaginal depth, vaginal size, phallic width, corporal body width, urethral length, pubic angle, and presence or absence of a uterus and/or gonads. MRI scans were interpreted by one of the authors (FC) and the measurements were recorded for comparison.

Sexual function was assessed using the Derogatis Sexual Function Inventory (DSFI) [6] and the Derogatis Interview for Sexual Functioning (DISF) [7]. The interviews were conducted by one of the two authors (ME, AA) and results and responses were reviewed and recorded for further comparison to determine statistically significant ($P < 0.01$) differences.

History and physicals were conducted by either of the same two authors conducting the interviews, and emphasized medical diagnoses, procedures and objective data. Physical exams were directed toward assessing height, weight, waist and body measurements, vaginal length and width, stretched clitoral length and anatomic width. One patient in the CAIS group had undergone vaginoplasty post pubertally and did not require regular dilations; another patient performed vaginal dilations intermittently. All CAIS patients were on estrogen therapy. All patients except one in the control group reported regular sexual activity.

Statistical analyses were performed using GraphPad Prism® 5 for Windows. Paired *t*-tests were performed when comparing objective physical findings, scores and measurements from the MRI exams, considering a *P*-value < 0.01 significant.

Results

The participant group included five women with CAIS and six women without. Controls were age-matched, nulliparous women who responded to the invitation. Patient age ranged from 22 to 58 years. One woman in the control group only underwent the MRI examination and declined to participate in the interview and physical examination.

The MRI results demonstrated a statistically significant difference in vaginal depth, but not width (Figs. 1 and 2). Median vaginal size was 1.68 cm^2 in CAIS women and 4.8 cm^2 in the control group ($P = 0.0081$). Respective

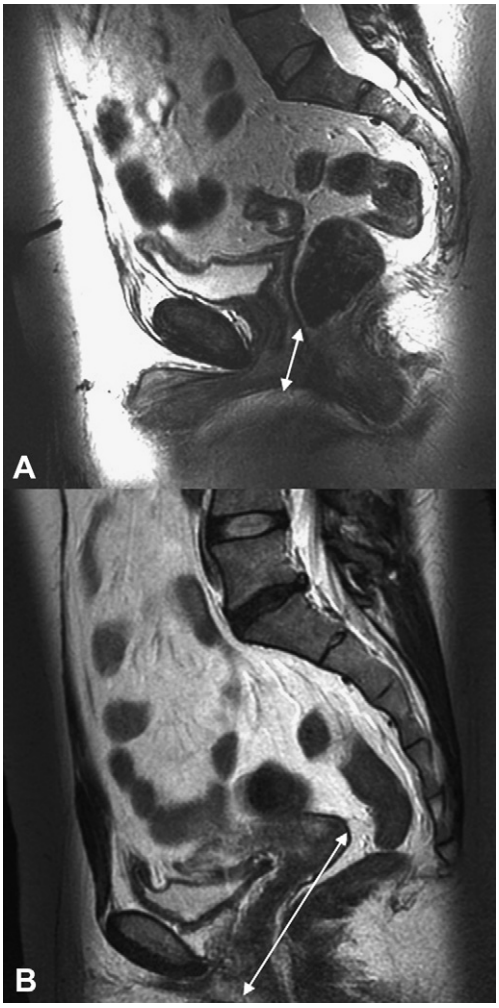

Figure 1 (A) CAIS vaginal depth 3.26 cm, (B) control vaginal depth 7.8 cm, as demonstrated on MRI.