Disorders of Sexual Development in Adult Women

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Question 1:

In your discussion of androgen insensitivity syndrome, there are multiple recommendations to refer to experts, including psychologists, endocrinologists, pelvic floor physical therapists, as well as others. Beyond clinical awareness so that the gynecologist can assist with initial diagnosis, is there truly a role for the generalist gynecologist in the care team of these patients? If so, what is that role beyond serving as a referral center?

Response from Drs. Gomez-Lobo and Oelschlager:

This is an excellent question. We believe that a well-informed general gynecologist can provide general gynecologic care to a woman with androgen insensitivity syndrome, including counseling about sexual function, counseling about prevention of sexually transmitted infections (STIs), screening for STIs, screening for bone health and breast cancer, and providing and monitoring estrogen therapy. As providers of women’s health, general gynecologists are especially equipped to provide general care for these women. If gynecologists are inexperienced with guiding patients through vaginal dilator therapy, or counseling, or performing gonadectomy, consulting with an experienced specialist is encouraged.

Question 2:

On a related note, what is the gynecologist’s role in managing the gonads of a patient with androgen insensitivity syndrome? Should these patients be referred to a urologist for gonadectomy?

Response from Drs. Gomez-Lobo and Oelschlager:

The gonads in these patients are typically found intra-abdominally. An experienced gynecologic surgeon can perform the gonadectomy with urology backup or provide a referral to a urologist. If the gonads are in the inguinal canal or labia, a herniorrhaphy is typically required and the gynecologist should collaborate with a surgeon experienced in herniorrhaphy.
Question 3:

As pediatric and adolescent gynecologists, how do you counsel your patients with androgen insensitivity syndrome about gonadectomy?

Response from Drs. Gomez-Lobo and Oelschlager:

We are careful to discuss the fact that testes (gonads) in women with XY chromosomes have been noted to be associated with increased risk of tumors and cancer. These cancers tend to be treatable. The risk in women with androgen insensitivity syndrome appears to be low but increase over time. The current standard of care is to remove the gonads after puberty. The gonads produce testosterone, which is aromatized to estrogen, which is important for bone and heart health. There are limited data to accurately evaluate the absolute risk and benefits of removing the testes. We also do not know the best imaging modality to monitor the gonads to accurately identify a tumor early.

Question 4:

How do you counsel patients with Turner syndrome regarding their future fertility? Do you recommend routine referral to a reproductive endocrinologist and infertility specialist?

Response from Drs. Gomez-Lobo and Oelschlager:

If a patient with Turner syndrome has had spontaneous puberty and is having menses, we recommend that they see a reproductive endocrinologist to discuss possible oocyte cryopreservation. If a patient with Turner syndrome required exogenous estrogen to induce puberty, then we discuss the option of donor egg pregnancy. With all Turner patients we discuss the possibility of aortic dissection during pregnancy and the fact that a normal cardiac evaluation before pregnancy does not preclude this calamitous event.

Question 5:

Given the increased incidence of mental health and psychiatric disease in patients with disorders of sexual development, what sort of collaboration with psychiatrists and other mental health professionals do you maintain in your practice to provide the best all-around care for these patients?

Response from Drs. Gomez-Lobo and Oelschlager:

Both of our pediatric clinical sites have multidisciplinary disorders of sexual development clinics with psychologists, psychiatrists, and social workers. For our adult patients we have developed relationships with adult psychologists and psychiatrists in the community for referral.

Question 6:

Particularly for patients with androgen insensitivity syndrome, the paucity of data and the heterogeneity of disease limits our care and counseling. Is there any ongoing research to assist with our knowledge of this complex syndrome?

Response from Drs. Gomez-Lobo and Oelschlager:

The annual AIS-DSD (Androgen Insensitivity Syndrome-Disorder of Sex Development) Support Group conference features research specific to androgen insensitivity syndrome but also for other DSD conditions. For example, research presented this year addressed attitudes towards DSD nomenclature. The National Institutes of Health (NIH) recently had a request for proposals for research in DSD, where multiple proposals were submitted specific to issues with androgen insensitivity syndrome.
Question 7:

More generally, is there any innovative and breakthrough research that is in the pipeline to assist with our care of patients with disorders of sexual development?

Response from Drs. Gomez-Lobo and Oelschlager:

The Disorders of Sex Development Translational Research Network (DSD-TRN), a national network of multidisciplinary DSD teams and patient advocacy groups, is focused on examining the pathophysiology of DSD, identifying novel diagnostic techniques, standardizing examination and evaluations of patients with DSD, evaluating quality of life outcomes, building a scalable registry to facilitate new diagnostic and treatment protocols, and identifying best practices. The DSD-TRN has partnered with the University of California, Los Angeles Clinical Genomics Center to offer a DSD genetic panel through whole exome sequencing, which currently tests for well over 100 genes involved in sex differentiation. This allows for accurate determination of genetic causes of DSD conditions that are not identified through karyotype or single gene testing.