

Don't Overlook Diagnosis of Müllerian Agenesis

BY ROBERT FINN
San Francisco Bureau

PORTLAND, ORE. — Some cases of apparent imperforate hymen are actually müllerian agenesis, and that possibility should be in the differential diagnosis, David M. Lee, M.D., said at a conference sponsored by the North Pacific Pediatric Society.

He described one case of a 12-year-old girl who had three hymenectomies for what turned out to be müllerian agenesis.

Another case, that of a 15-year-old girl, is perhaps more typical. Her chief complaint was primary amenorrhea and cyclic pain. She had normal breast and pubic hair development. The original ultrasound examination was interpreted as showing a "small uterus," deviated to the right, and normal ovaries. She had one attempted hymenectomy.

"In defense of the first physician, it does look like an imperforate hymen," said Dr. Lee of Oregon Health & Science Univer-

sity, Portland. In fact, she had müllerian agenesis and was missing the uterus and the upper vaginal system.

In addition to müllerian agenesis, several other conditions should be in the differential, he said. These include vaginal atresia, longitudinal septi, transverse septum, cervical agenesis, androgen insensitivity syndrome, and intersex conditions.

The work-up should include serum testosterone—which differentiates between müllerian agenesis and androgen in-

sensitivity—and imaging, either ultrasound or MRI.

The incidence of müllerian agenesis is quoted in the published literature as 1 per 5,000 women, but Dr. Lee's clinical impression, based on the number of referrals he receives, is that the actual incidence is higher.

Women with müllerian agenesis have normal 46XX chromosomes, normal external genitalia, normal ovarian function, and normal thelarche and pubarche. The upper vagina is absent, and the lower vagina may be of variable length. They have normal fallopian tubes and uterine horns.

Assisted reproductive technology can help these women have children. This typically involves an egg aspiration, artificial insemination, and a surrogate who carries the embryo to term.

Vaginal dilatation with soft vaginal dilators of progressively larger sizes provides effective treatment for about 90% of women.

The remaining 10% will require surgery to create a neovagina. Dr. Lee uses epidural anesthesia for this surgery. He takes a skin graft from the buttocks or lateral thigh, somewhat thicker than a split-thickness skin graft, but not as thick as a full-thickness graft. He places the graft on an inflatable vaginal stent, and inserts that into the newly dissected neovaginal space, suturing the stent into place.

After about a week of immobilization to allow the graft to take, the patient is brought back to the operating room to have the stent and any necrotic remnants of the graft removed.

"You end up with a neovagina that's usually a normal 10-12 cm length, normally functioning, and it's a very successful procedure," Dr. Lee said.

Dr. Lee is also developing a laparoscopic procedure. "I mobilize peritoneum from the inside of the abdomen laparoscopically, and then pull that peritoneum down, and use peritoneum rather than a skin graft," he said. "I think that's going to be eventually the most effective surgical treatment." ■

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