In 1990, Arlene (MD ’84, Res ’90) and Mark (MD ’84, Res ’90) Baratz’s 6-year-old daughter, Katie, had a surgical procedure to repair a hernia in her groin. Hernias are common, and are treated fairly easily with surgery. What’s far less common is for a young girl’s hernia to be caused by a partially descended testis—which is exactly what Katie’s surgeon found on his operating table.

How can a girl have testes? Good question. Male and female gonads and genitals actually develop from indifferent precursors, and every embryo has two sets of ducts, the Müllerian and Wolffian, that can develop into internal reproductive structures. A number of genes, especially the SRY gene located on the Y chromosome, direct gonadal and genital development. The presence of the SRY gene on a Y chromosome typically signals the indifferent gonads to develop into testes, which produce the hormones known as androgens. The androgens in turn (principally, testosterone) are responsible for the development of indifferent genitalia into male external structures. At the same time, the testis secretes the hormone Müllerian-inhibiting substance (MIS), which prevents the Müllerian ducts from developing.
into female internal structures. The absence of a Y chromosome with SRY, on the other hand, results in ovarian development. Without testosterone from the testis, female external structures develop, rather than male ones. (In addition, prenatal hormone exposure likely influences areas in the brain associated with hormone secretion in adolescence and adulthood.) This is a highly oversimplified explanation of what typically happens. Along the way, any number of hormonal signals can go astray; that’s where things become anything but straightforward—in the delivery room and beyond.

Katie, for example, has complete androgen insensitivity syndrome (CAIS): a condition in which a Y chromosome triggers the development of testes, but for various reasons, the body doesn’t respond to the androgens produced there. The result is that the fetus with XY (typically male) chromosomes develops external female structures. The baby will look like a girl—and, typically, like Katie, grow up to identify as a girl—but will have internal testes, fallopian tubes, and a womb.

When they learned of Katie’s testes, all that Arlene (a radiologist) and Mark Baratz (a Pitt orthopaedist) knew of was what they had been taught in medical school. “They were all called ‘types of hermaphroditism’ at that time,” Arlene Baratz says (“male pseudo-hermaphrodite” being the term used to describe Katie’s biology). “And we were told that when you diagnosed someone with one of these conditions, you did whatever you could to normalize their body. . . . And you had to do it early, so they wouldn’t know about it, and then you never told them.”

Before the era of informed consent, physicians often made decisions about gender assignment in cases of what are now known as “disorders of sex development” (DSD), which sometimes involved irreversible surgeries on ambiguous genitalia or other tissue, without involving the families. The thought was that the parents might reject the child. Today, parents are, of course, involved in all decision making, notes Selma Feldman Witchel (MD ’78, Fel ’83), cochair of the DSD Committee at Children’s Hospital of Pittsburgh of UPMC. The DSD Committee is a multidisciplinary team with representation from urology, genetics, pathology, and endocrinology; Arlene Baratz is the family/patient representative. Witchel, who is a Pitt associate professor of pediatrics, says the team gathers information from the family, physical examinations, hormone levels, imaging, and genetic testing to offer the best recommendations reflecting which gender a child will most likely identify with. At Children’s, members of the DSD team are also involved in the care of gender-questioning children, adolescents, and young adults. “We are very aware that binary concepts of sex and gender are inadequate, and we need to be open to how individuals see themselves,” says Witchel.

The Baratzes opted to leave in the testes, and let Katie eventually decide whether she wanted them. And they decided to be open with their daughter about CAIS, but it took some years to fully explain.

“I think I told her [in the early conversations] that most women have a nest inside their bodies where a baby can grow,” Arlene Baratz says, in a 2011 documentary, The Truth of My Sex. And when Katie asked if she had that nest, too, her mother told her no, “but you will be a mommy, and you’ll have a very lovely family, and it will be very exciting, because you’ll be adopting your children.”

“It really kind of changed my world,” recalls Katie (now Katie Baratz Dalke) in the documentary. “I remember even as, like, a 9-year-old feeling sad whenever I saw pregnant women.”

As Katie reached her teen years, her parents explained the details of her anatomy and genetics. Katie felt betrayed; a secret had been kept from her. “My doctors all knew about it . . . strangers, like people who just happened to be in the office when I was there, knew more about me than I did.”

Talking about sex biology is not something our society is particularly good at, and diverse sex biology (or identity) even less so. The binary model is deeply embedded in our collective psyche. When a new baby is born, or even beforehand, what’s the first thing everybody wants to know? Is it a boy or a girl?

Yet estimates of how many individuals present with DSD range from one in a hundred (according to the Intersex Society of North America) to one in 4,500 (per the National Institute of Child Health and Human Development). It depends on the variations you include. A 2006 world “Consensus Statement on Management of Intersex Disorders” includes Klinefelter syndrome—that results when boys have more than one X chromosome; the incidence of Klinefelter alone is one in 800.

“There’s a real need to find a common language,” Arlene Baratz says.

The world consensus statement, published in a period of increasing acceptance for sex variability, did away with the hermaphrodite-based terminology that the Baratzes first learned. Yet the current DSD lexicon is not embraced by everyone.

“I think clinicians often don’t realize how the terms that they use can be hurtful to patients,” says Arlene Baratz. “Just the term ‘disorders’ is very controversial in the community. People feel like, Well my body is different; why is that a disorder? It’s pathologizing.”

Patients can become disengaged, or demanding, about the terminology used to define them or their conditions, Arlene Baratz notes. Clinicians react. “The whole interaction ends up making people feel like they don’t want to go for medical care,” she says. “It has led to health disparities.”

To combat some of this, and to be the social support for others that she wishes she’d had as a young mom, Arlene Baratz is a board member and medical and family adviser to the AIS-DSD Support Group and moderator of the AIS-DSD Parents Group. In 2014, she helped draft a policy that got the community “a seat at the table” (from study inception to goals and design) for two currently National Institutes of Health–funded DSD research projects. Arlene Baratz is also a founding member of Accord Alliance (which promotes integrated, comprehensive care in the DSD community) and sits on the board of Advocates for Informed Choice (which advocates for children born with DSD in legal and other spheres). In February, she gave a talk at the medical school, “DSD 101: Dealing with Reproductive Diversity.”

With Katie, who is now a psychiatrist at the University of Pennsylvania, Arlene Baratz cochairs the committee on patient perspectives as part of an ongoing effort to update the 2006 world consensus statement, the earliest papers from which are expected to be published by summer’s end. A separate nomenclature committee has acknowledged the need to replace “DSD.” Baratz is hopeful: a number of patients sit on that committee.

“When I look back, I can see that [change] is happening,” she says. “It always seems very slow while you’re doing it, but it’s such a different world from when my family was young, and I’m very grateful for that.”