

When someone is raised female and the genes say XY

September 12 2009, By SETH BORENSTEIN , AP Science Writer

(AP) -- It's the birth defect people don't talk about. A baby is born not completely male or female. The old term was hermaphrodite, then intersex. Now it's called "disorders of sexual development." Sometimes the person with the problem doesn't even know it and finds out in an all too public way.

That's been the painful plight of a few female athletes through history. And apparently that's the situation for South African track star Caster Semenya.

Two Australian newspapers reported Friday that [gender](#) tests show the world champion athlete has no ovaries or uterus and internal testes that produce large amounts of testosterone. The international sports federation that ordered the tests wouldn't confirm the reports.

Experts say Semenya should be allowed to race as a woman and they cringe at how her case is exploding publicly in the news media. They worry about psychological scars. Two years ago, a star female track athlete who tested male attempted suicide.

Unless she took some illicit substance, Semenya is a female with a [birth defect](#), simple as that, said Dr. Myron Genel, a professor emeritus of pediatrics at Yale University. He was part of a special panel of experts convened by the International Association of Athletics Federations in 1990 that helped end much, but not all, genetic gender testing.

"It's no different in a sense than a youngster who is born with a hole in the heart," Genel said. "These are in fact birth defects in an area that a lot of people are uncomfortable with."

Semenya is hardly alone. Estimates vary, but about 1 percent of people are born with abnormal sex organs, experts say. These people may have the physical characteristics of both genders or a chromosomal disorder or simply ambiguous features.

Sometimes a sexual development problem is all too obvious when a baby is born. Other times, the disorder in girls may not be noticed until puberty, when she doesn't start her period. And still other times, especially with the androgen insensitivity syndrome experts think Semenza might have, it remains hidden until she tries to have a baby - or in the case of an athlete, until she's given a genetic test.

Genetic testing of women over five Olympics found genetic gender issues in 27 out of 11,373 women tested, according to a 2000 Journal of the American Medical Association article. However, none were men deliberately posing as women, as competitors fear.

Dr. Louis Elsas, chairman of biochemistry at the University of Miami and a member of the IAAF panel with Genel, said he had hoped the genetic gender testing issue was over after the 1996 Olympics, when most major sports abandoned regular testing. He recalled having to talk to a female athlete and reveal that she had XY chromosomes and that she'd be infertile. It's something that shouldn't splash onto television, newspapers and the Internet, he said.

"It's a severe emotional trauma," Elsas said.

The concern that women with XY chromosomes have a competitive advantage "is malarkey. We don't segregate athletes by height," said

Genel, speaking from an international endocrinology conference in New York that has sessions on intersex issues.

Dr. Joe Leigh Simpson, past president of the American College of Medical Genetics and a member of the IAAF panel, agreed: "Any elite athlete ... has a competitive advantage, or otherwise they wouldn't be an elite athlete."

Simpson, associate dean at Florida International University, said the issue should be simply whether men are masquerading as women. Semenya is clearly a woman, he said.

Nearly all the disorders are caused by genetic mutations, Simpson said. And they usually happen in the first eight weeks of pregnancy, he said.

There are many types of sexual development disorders, all of them rare, but they add up, the experts said. Depending on the particular disorder and individual condition, treatment could involve surgery or hormone therapy or both. The issue is often not just what sex to assign the child, but when to assign it. It used to be that doctors pushed surgery on babies; now many times they wait. Sometimes they wait until the patient is old enough to help make a decision.

David Sandberg, a pediatric psychologist at Michigan, said he advises families to go slowly when deciding whether to raise their child as a boy or girl or whether to have surgery. Treatment varies depending on the disorder, but has become more conservative over the years, he said.

But that's when the problem is noticeable. When it comes to some athletes like Semenya, it's not even known until tests reveal it.

Maria Martinez-Patino knows the issue firsthand. A world-class athlete, she was raised and looked like a normal female and even received the

needed "certificate of femininity" to participate in the 1983 World Track and Field Championships in Helsinki, Finland.

In 1985 at the World University Games in Kobe, Japan, her test came back with an XY and she was not allowed to compete. Martinez-Patino had androgen insensitivity, meaning she didn't respond to testosterone. That meant she also didn't have a competitive advantage from having an XY chromosome.

"I sat in the stands that day watching my teammates, wondering how my body differed from theirs," she wrote in the medical journal *The Lancet* in 2005. "I spent the rest of that week in my room, feeling a sadness that I could not share."

On the Net:

American Academy of Pediatrics consensus statement on intersex disorders:

<http://pediatrics.aappublications.org/cgi/reprint/118/2/e488>

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