

Doctors miss major cause of infertility and obesity

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Gail Donnelly's classmates nicknamed her "Knobby" because she was so skinny all her bones seemed to poke out from under her skin. But when Donnelly turned 27, that once knobby frame disappeared under mysteriously ballooning weight. Her diet hadn't changed, she was still walking several miles a day, but she gained 50 pounds in just six months.

Her doctor thought the cause was ovarian cysts. It took ten years and two surgeries before a new doctor accurately diagnosed her with polycystic [ovary syndrome](#) (PCOS). It's a serious [metabolic disorder](#) and one of the major causes of hormonally related infertility, yet the disorder remains largely undiagnosed and unknown. About 5 million women in the U.S. are affected by it.

"Women are told they are too fat and aren't taken seriously for a long time," said Andrea Dunaif, M.D., the Charles F. Kettering Professor of Medicine at Northwestern University Feinberg School of Medicine and a physician at Northwestern Memorial Hospital. "They go to an average of four doctors before they are diagnosed. They have been to physicians who say 'there is nothing wrong with you, don't worry'."

Before she received the news about PCOS, Donnelly, an ordinarily happy person, had sunk into a deep depression and her boyfriend accused her of letting herself go.

Dunaif, a national expert, knows otherwise. The complex genetic disease has long-term health risks throughout a woman's lifespan, including

obesity and double the rate of metabolic syndrome, a constellation of risk factors for diabetes and heart disease.

Not only women are affected. Dunaif recently published a paper showing that brothers and fathers of women with PCOS also have a greater prevalence of [obesity](#) and [metabolic syndrome](#). "It's essential that women and men are diagnosed and treated for this," Dunaif said.

She recently was awarded a \$5 million, two-year grant from the National Institutes of Health to continue her ongoing research into the syndrome's genetic causes.

Symptoms of PCOS in women often show up in adolescence and may include irregular periods and excess hair on the face, chest or back - all caused by high levels of male hormones.

Dunaif blames the syndrome's low profile on its name. "It has the word ovary in the name and that has led people to think it's just 'female' troubles and nothing important," she said.

PCOS gets its name from the small ovarian cysts found in the first women studied, though not all [women](#) who suffer from PCOS have these cysts. Dunaif would like to rename the syndrome "Syndrome XX" to bring it into the spotlight.

After Dunaif began treating Donnelly with medication for insulin resistance - which had caused her rapid weight gain -- Donnelly's excess pounds dropped off and she was able to become pregnant. "If I had known about this sooner, my life would have been entirely different," Donnelly said.

She couldn't take the medication when she was pregnant or nursing, however, so her weight soared 80 pounds with each pregnancy. At one

point, she weighed 280 pounds. Of her struggle with PCOS, she said, "It's like having a battle with my body at all times."

Now a 40 year-old mother of three from the Chicago suburbs, Donnelly worries about her children inheriting the disease. She realizes many of her relatives -- who developed diabetes as adults -- likely had PCOS.

Provided by Northwestern University

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