



# Patient Report

## **Down There: Cervical Cancer in PJS**

A Report by  
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This is the most difficult patient-view report I've written. For those who don't know me or my reports, I am a PJS patient who manages this group. I am not a doctor, but as a well-read patient, I review different topics of interest to PJS folks like genetics, imaging, PJS-cancer links, history and PJS literature. Periodically I write new reports, like this one on cervical cancer in PJS women.

Each person has a different "need to know" about PJS, its manifestations and treatment. I have a high need to know about most PJS topics, but this one makes me nervous. It isn't just because it's about sex (a woman's cervix is the part that gets bumped during intercourse) and having babies (except in c-sections, babies exit the uterus through the cervix and vagina), but the rare cervical cancer associated with PJS is difficult to diagnose and treat.

Let's start with sex. These days books and articles on cervical cancer & pap smears emphasize that risk for cervical cancer comes from HPV (the human papilloma virus) which is sexually transmitted. There are many types of HPV and many thousands of women affected with it. Only a few types of the virus in a small percentage of women develop into cervical cancer. This type of cancer is a squamous cell cancer. It develops on the surface of the cervix and grows into the cervical body and uterus. Pap smears find most of these cancers early and they can be removed in an in-office procedure. About 80% of cervical cancers are squamous cell cancers.

Unfortunately, PJS-related cervical cancer (which I'll call MDA for minimal deviation adenocarcinoma of the uterine cervix, also called adenoma malignum), is a whole different creature. Adenocarcinomas of the cervix are 10-15% of the cervical cancers. MDA is 1-3% of those. It is rarer than rare. And hard to diagnose.

The recommendation that PJS women get annual pelvic ultrasound exams is to help detect this cancer as well as PJS-related ovarian tumors. Please note: many PJS experts think most PJS women have ovarian sex cord tumors with annular tubules (SCTAT), but these microscopic tumors are seldom visible by imaging or even at oophorectomy (removal of ovary). Similarly, many cases of MDA are diagnosed at or after hysterectomy (removal of the uterus or womb).

An annual pap smear, while necessary, isn't sufficient to test for adenoma malignum. The Johns Hopkins [PJS booklet](#) recommends for women: From age 18 -- annual gynecological exam, transvaginal ultrasound and a CA-125 blood test for ovarian cancer. Other guidelines are similar to this.

If you haven't had a pelvic ultrasound (many pregnant women have), it is easier than scopes, but worth preparing for. There are two parts to the exam. An hour before the exam you will be asked to drink a quart of liquid (and not to pee). At the exam, you remove clothing below the waist & climb onto a table. The ultrasound examiner squeezes some cool, slippery goop onto your belly and then rubs it with a handheld device, similar to a computer mouse, while you try not to laugh or pee. Once that part of the exam is

over, you can use the bathroom. Then comes the magic wand which is cloaked in a condom and lathered in more slippery goop. I've always had the choice of slipping it into my vagina myself, but don't know if that's a California custom. The examiner then manipulates the wand to produce images on a small TV screen. Most the images are in black and white, but it's possible to see color and even hear the pulse (very cool). Each part takes 10-20 minutes. When they are over, you go back to the bathroom to clean up. If there is a problem or question, a specialist may come in to get better images or talk with you. The specialist's report is sent to your doctor who will explain it to you (and give you a copy, if you ask).

I was told by a genetic counselor that PJS women need to see a gynecological oncologist (specialists in cancers of women's reproductive tracts). I have a wonderful gyn-onc who knows next to nothing about PJS (I taught her everything she knows). She sees many women with BRCA1 and BRCA2 --hereditary breast/ovarian cancer syndromes, but she is unfamiliar with SCTAT and MDA. She is used to looking for problems in different places than I am likely to have them.

MDA is difficult for many reasons. It is both highly aggressive and very well hidden. The few symptoms of MDA could easily be confused with other, more normal, problems. The main symptoms I've read about are a watery or mucoid discharge from the vagina, sometimes for years before diagnosis, and vaginal bleeding. Since SCTAT causes irregular vaginal bleeding in many PJS women, this isn't a fail-proof symptom. I think annual transvaginal ultrasound exams are necessary to diagnose MDA (which can look like a cyst). Some screening guidelines include deep biopsies or cervical brushing to reach those hard to find MDAs.

For parents worried about young daughters -- most reports of MDAs in PJS women are ages 20's to 40's. The age 18 screening guideline is about 5 years before the earliest reported age of onset. FYI, a rule of thumb for screening guidelines for hereditary cancer is to start screening 5 years before the earliest reported age of onset in a family member or others with the syndrome.

**The genetic connection.** There have been three studies exploring the link between MDAs & PJS <sup>1,2,3</sup>. The first found that the MDA genetic locus was near, but not at, the PJS genetic locus. The second found that though approximately 10% of all cases of MDA of the uterine cervix are found to occur in women with PJS, another tumor suppressor gene near the PJS locus is probably responsible for MDA & SCTAT in PJS women. The third and most recent report puts the MDA genetic locus back at the PJS genetic locus. It will be interesting to follow these investigations. If a genotype-phenotype connection could be found, then screening could be modified to fit a patient's genes.

By the way, I followed the genetic counselor's advice in 1999 and saw my gyn-onc who did a complete pelvic exam & endometrial biopsy and pronounced me fine. Then the pelvic ultrasound found a 13 cm ovarian tumor (that was missed on manual exam). I chose to have the ovary and my uterus removed, but kept part of one ovary. I was 43

years old and had had many years of bad pap smears including squamous carcinoma-in-situ of the cervix. The gyn-onc argued that I should have both ovaries & uterus removed to limit future cancer risk and to treat my breast cancer. I now have cysts on the remaining ovary and get frequent pelvic ultrasound exams. It seems likely that I'll have that removed, but it's been a good 3 1/2 years. So, we were both right.

I don't want to scare anyone, especially into unnecessary surgery. But I do hope that you will be motivated to have a pelvic ultrasound if you are age 18 or older. If your GP or gynecologist isn't willing to order the test, you are welcome to show him/her this letter and the screening guidelines from PJS specialists. I am also willing to give references to many articles to support the request.

Remember, each of us hopes to live many years with this rare syndrome. My advice in this letter is from a patient to other patients. I am not a doctor or specialist. May we all be as healthy as possible for as long as possible.

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<sup>1</sup>*A distinct region of chromosome 19p13.3 associated with the sporadic form of adenoma malignum of the uterine cervix.* Lee JY, Dong SM, Kim HS, Kim SY, Na EY, Shin MS, Lee SH, Park WS, Kim KM, Lee YS, Jang JJ, Yoo NJ. *Cancer research.* 1998 Mar 15;58(6):1140-3.  
PMID: [9515797](#).

<sup>2</sup>*Somatic mutations in the STK11/LKB1 gene are uncommon in rare gynecological tumor types associated with Peutz-Jegher's syndrome.* Connolly DC, Katabuchi H, Cliby WA, Cho KR. *American journal of pathology.* 2000 Jan;156(1):339-45.  
PMID: [10623683](#).  
Full text available for free.

<sup>3</sup>*Mutations in the STK11 Gene Characterize Minimal Deviation Adenocarcinoma of the Uterine Cervix.* Kuragaki C, Enomoto T, Ueno Y, Sun H, Fujita M, Nakashima R, Ueda Y, Wada H, Murata Y, Toki T, Konishi I, Fujii S. *Laboratory investigation; a journal of technical methods and pathology.* 2003 Jan;83(1):35-45.  
PMID: [12533684](#).

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