



# Patient Report

## **Testicular Tumors & Gynecomastia In PJS Boys**

A Report by  
Stephanie Sugars

***Disclaimer: I'm not a doctor or medical professional, but a PJS patient. I am not offering medical advice of any type. In fact I'm completely unqualified to write this report as I have neither testicles nor education in male reproductive organs beyond that sixth grade sex education movie I saw in 1968. On the other hand, if I don't do it, who will? Hopefully Dr. Young in Boston is writing a wonderful article on the topic. Until then, here are my observations.***

This report is in two parts, the first in which I try to provide some overview and the second which is a list of all article titles I could find on the topic. I actually read only two articles from that list and most of what I've read on testicular tumors is more general than PJS specific. Those interested in the topic are encouraged to continue their own research by reading the abstracts available through PubMed at <http://www.ncbi.nlm.nih.gov/PubMed/>. I can supply titles of other articles on testicular tumors for those interested, though a visit to a medical library and examination of texts on urology, pediatric and adult endocrinology and oncology would be a good place to start. Use a medical dictionary to help with unfamiliar terms.

Most of the reported testicular tumors in PJS males occur in prepubertal boys. In fact, I could find only one reference to a testicular tumor that might have occurred in an adult.<sup>[1]</sup> The tumors present in a variety of ways. Sometimes they cause pain or can be felt (palpated). Other times the child might present with gynecomastia, swollen breasts, due to increased estrogen or aromatase production. Sometimes a child is tall or large for his age or experiences precocious (early) puberty. Or perhaps his bone age is advanced. Often the tumors are microscopic, but can be seen by testicular ultrasound. Other times, they can be diagnosed only by biopsy. They are rarely cancerous and I found no articles stating any PJS male ever died of a testicular tumor. Hormonal tests have showed a variety of results in different boys, so I wouldn't even guess whether there is a PJS-testicular tumor hormonal profile.

The tumors appear on pathology exam to resemble the SCTAT (sex cord tumors with annular tubules) that are very common in PJS females. The link between Sertoli cell tumors and SCTAT is unclear to me. Here's a quote from article 12:

"Only three other male patients with Peutz-Jeghers syndrome and gonadal tumors have been described previously (articles 13, 16, 17), all presented with gynecomastia between birth and six years of age. Thus, our patient represents the fourth such case reported. Three of these four boys had testicular enlargement and prepubertal secretion of gonadotropin, all had advanced bone age, and three had increased linear growth."

"The tumors in our patient were comprised of sex cords forming annular tubules...We consider them to be sex-cord tumors, although others might consider them to be large-cell calcifying Sertoli-cell tumors. The distinction may be somewhat arbitrary; both are more often bilateral, multifocal and microscopic than most other sex-cord neoplasms."

I should take a moment to explain Sertoli cell tumors. There are two main categories of testicular tumors - germ cell tumors (GCTs) and sex-cord stromal tumors. GCTs account for 95% of testicular tumors. Sertoli cell tumors are less than 1% of testicular tumors and are of two named types -- large cell calcifying, and sclerosing. The type associated with PJS is the large cell calcifying or LCCSCT. The germ cells are responsible for producing sperm and the Sertoli cells have three functions:

1. They support the germ cells;
2. They produce estrogens and some androgens; and
3. They form the basement membrane of the seminiferous tubules.

Several articles suggest that presence of testicular tumors in PJS prepubertal boys leads to a disruption of hormones, frequently an overproduction of estrogen, which can cause gynecomastia and the other problems like accelerated growth and bone age.

What to do about the hormonal disruption is open to debate. Many articles suggest removal of one or both testicles. Other articles discuss different hormonal treatments. One of our members mentioned the use of Tamoxifen, an anti-estrogen hormonal treatment usually used for the prevention and treatment of breast cancer in women. Aromatase inhibitors are now used for metastatic breast cancer and I wonder if they'd be used for those whose testicular tumors overproduce aromatase.

Oh yes, bilateral mastectomy, removal of the breast tissue, seems fairly common in cases with gynecomastia.

I've only read of three cases of malignant testicular tumors in PJS males. The first is described in article 11 -- the case of a boy with gastric cancer that metastasized to the testicle. The second and third are a father and son with Sertoli cell cancer mentioned in a table in an article by Dr. Boardman of Mayo Clinic.<sup>[1]</sup>

One other thing that I find fascinating is that LKB1, the PJS gene product, has widespread expression throughout the human body, but is predominant in the seminiferous tubules of the testis (spermatogonia and possibly Sertoli cells, rather than the permatids).<sup>[2]</sup> I don't know whether this is connected to risk of testicular tumors in PJS males, since the researchers didn't address that question.

It's a good thing that Dr. Young is writing an article about Sertoli cell tumors in PJS males. He worked with Dr. Scully in Boston, Massachusetts, for many years before Dr. Scully retired. Both have written extensively about rare reproductive tract tumors and Dr. Scully was the first to describe SCTAT and to associate it with PJS in 1970. Their work is pioneering and doctors from around the world consult them for help with their difficult cases. We need to learn more about these tumors and their effects on PJS males. At least six cases of possible testicular tumors have been reported in this group in just over three years. What we've seen in print, doesn't encompass everything.

Which reminds me. If you have a history of gynecomastia and/or testicular tumors, you might want to steel yourself before looking through the next e-mail. Perhaps you will find yourself in print, recognizable by doctors' names, dates and ages or location. It can be quite a shock to hear your story told by others.

This is my report as of today, July 15, 2003. Hopefully new and better articles on testicular tumors and gynecomastia in PJS boys will be published soon. Those who wish to help Dr. Young in his study, may contact him at Massachusetts General Hospital.

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I've sent him copies of these e-mails, so he's familiar with them.

Very best to all today and everyday,  
Stephanie Sugars

### **References:**

[<sup>1</sup>] Boardman LA, Couch FJ, Burgart LJ, Schwartz D, Berry R, McDonnell SK, Schaid DJ, Hartmann LC, Schroeder JJ, Stratakis CA, Thibodeau SN. Genetic heterogeneity in Peutz-Jeghers syndrome. *Hum Mutat.* 2000;16(1):23-30. PMID: [10874301](https://pubmed.ncbi.nlm.nih.gov/10874301/) [PubMed - indexed for MEDLINE]

[<sup>2</sup>] Rowan A, Churchman M, Jefferey R, Hanby A, Poulson R, Tomlinson I. In situ analysis of LKB1/STK11 mRNA expression in human normal tissues and tumours. *J Pathol.* 2000 Oct;192(2):203-6. PMID: [11004696](https://pubmed.ncbi.nlm.nih.gov/11004696/) [PubMed - indexed for MEDLINE]

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