By Carly Heyman

Living With FXPOI

As a 25-year-old woman with fragile X-associated primary ovarian insufficiency (FXPOI), I am one of a relative few who know the cause of their ovarian insufficiency: I am a Fragile X carrier. My older brother Scott was diagnosed with fragile X syndrome when the gene was first identified in 1989.

Since I was first informed I was a carrier five years ago, I have known that getting pregnant for me will be a challenge. But my attitude is straightforward: All people have "stuff" in their lives—and this is mine.

"Primary ovarian insufficiency." What does that mean? Does it mean I can't get pregnant? Does it mean I should go through hormone stimulating therapy? Does it mean there will be no "mini me?" Naturally, as my friends and I become older, our conversation turns more to a future of marriage, jobs and family. But I am different from my peers. Getting pregnant will not be simple for me, and I may never have a child who shares my genes. I feel sad sometimes when my friends discuss pregnancy. However, when I was first diagnosed with FXPOI, having babies was not my major concern. Instead, I was incredibly relieved that at last we had a definitive diagnosis for the variety of symptoms that had been affecting me for years. Someone finally knew what was wrong with me, and I was not going crazy!

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My FXPOI symptoms began when I was 17 and accelerated over the next three years. I was plagued by night sweats, hot flashes, depression, anxiety, weight loss and poor concentration. You name it, I had it. The only people I could relate to were menopausal women over the age of 50—definitely not cool for a young girl about to enter college.

My family and I were so clueless about my condition that we even installed two extra air vents in my bedroom because I was convinced that my room was getting overheated at night. No one knew what was wrong with me, and I was losing faith in medicine and medical professionals.

The psych docs were pushing me toward medical solutions and the medical docs were pushing me toward the psych end of things. I was stuck in the middle, desperate for an answer and scared for my future. My mind, body and spirit were weakening.

Finally, after three devastating years I was diagnosed at the National Institutes of Health with primary ovarian insufficiency (the "fragile X-associated" was added later when scientists learned more about the condition). I was told my chances of getting pregnant were 5-10 percent, but at the time I did not care. I was anxious to feel like myself again, eager to stop taking useless medications and hopeful about returning to college as a "normal" healthy student. The pregnancy thing? I conveniently thought, "I'll deal with that later."

Since the onset of hormone replacement therapy, I have returned to my happy, healthy self, buoyed by physical and emotional well-being and satisfied with my knowledge of FXPOI.

Well, "later" has now arrived. So, where does that leave me? Honestly, I do not know. I have a loving, understanding and supportive boyfriend. If I adopt, great. If I stimulate eggs, terrific. If I use an egg donor, no problem. Who knows—my future husband could have an insufficient sperm count!

Since the onset of hormone replacement therapy, I have returned to my happy, healthy self, buoyed by physical and emotional well-being and satisfied with my knowledge of FXPOI. Growing up with a sibling with fragile X syndrome, I have learned to love, understand and accept people with special needs for who they are and what they can offer. Why not treat myself with the same respect?

There are basically two ways one can respond to a FXPOI diagnosis, or to any diagnosis, for that matter. Personally, I can sulk, feel bad for myself and squirm uncomfortably every time my girlfriends bring up the subject of pregnancy. Or, I can accept my medical disposition, enjoy the built-in birth control it provides, and become an advocate. That's the path I have chosen, and for me it has been the right one. Among other benefits, it has given me the opportunity to educate and support my family, friends, doctors and researchers.

I have also been able to help a younger cousin going through a similar scenario spare herself the terrible experience I

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endured before my diagnosis. I encourage all family and friends to be open and accepting of their conditions. I have found that the more you talk about a disorder and learn about it, the less scary and embarrassing it becomes.

I recognize that life is too short to punish oneself for conditions one cannot control. I choose to lead a life in which I am active in the areas I can control and accepting of the areas I cannot. I cannot control FXPOI—but I can control my attitude toward it.

I hope to share this attitude with others in hopes that they, too, can one day accept themselves for who they are, which goes so far beyond their disorder, disability or diagnosis. FXPOI is a part of my life, but it is not my life. ■

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