

Fitting Together the Pieces

Caitlin L. Gong



Early diagnosis for Turner Syndrome has been one of the top priorities of the Turner Syndrome Foundation since its inception. A video was made last year featuring Dr. Henry Anhalt; Shaz Kramer, the mother of a 16 year old daughter with Turner Syndrome; and Sarah Kramer, her 16 year old daughter emphasizing the importance of early diagnosis. It can be viewed at <http://www.youtube.com/watch?v=1Q36VMahz2w>.

All too often girls and women with this condition are diagnosed at too late an age to receive the necessary growth hormone and estrogen treatments to develop normally as a female. Catrina Beeny's story emphasizes why early diagnosis is so crucial and that it should be more easily identifiable so that it is less of a random diagnosis.

Catrina Beeny of Kalamazoo County, Michigan, was diagnosed when she was 20 with Turner Syndrome after a chance meeting with a female cashier at her college bookstore with her mother. The cashier looked at her and asked her if she had Turner Syndrome. Catrina replied that she didn't think so and her mother went on to explain, "They looked at her when she was little but they said she didn't have it. Then when she was older and had her period, they were doubly convinced."

The cashier said that she had asked because she had a daughter with this condition and said, "You have the same stature and build as her. You have a lot of the physicality of a girl with Turner Syndrome. You have a low hair line, shorter fingers. Did you have any ear problems when you were little?"

Catrina was floored by this and mentioned that when she was growing up she had 15 ear surgeries. The cashier said that she might have Mosaic Turner Syndrome because she had minimal indications of the condition. She would be diagnosed by a blood test called a karyotype where pieces of a person's chromosome are examined under a microscope to find the exact location where a chromosomal abnormality is located.

Growing up she also had difficulties feeling accepted in elementary and middle school. She explained, "I always felt awkward and different. I wasn't treated badly but I didn't really feel connected to many classmates either. I think some of it had to do with the fact that I looked and acted different from the other girls in my class. I was shorter and a little bit awkward socially. High school was a different story. I met my two best friends, Abby and Kaitlin, who are still my best friends to this day."

Eventually Catrina took time off from school because of this information and for other personal reasons and worked on getting permission from her doctor to let her have a karyotype done. It took her and her mom six-seven months before they were able to get the blood test and the results showed that she had full Turner Syndrome and no second X chromosome. With Turner Syndrome, girls and women either are missing part of an X chromosome or the full X chromosome.

Learning that she had Turner Syndrome was a relief because she knew now where all of her medical problems and insecurities came from. If it hadn't been for that meeting with the cashier, she still wouldn't know she had it today. Now she can find support for her condition and become part of a larger community of other individuals who live with this complex condition that affects 1 in 2000 females.

She explained in detail how this late diagnosis impacted her when she said, "I would advise doctors to know that Turner Syndrome comes in many forms. Doctors, or at least the doctors I saw before I was diagnosed, did not know that a woman with Turners could have some symptoms and not others. They believed that all girls with the condition were exactly the same. It took six months for me to even convince my doctor to let me get the karyotype. The biggest thing for doctors to remember is that Turner Syndrome and other genetic disorders are not all or nothing. To have Turner Syndrome does not necessarily mean that you won't have periods; it just means it's more likely that you won't. The existence of gray areas for symptoms of Turner Syndrome needs to be understood or diagnoses will fall through the cracks by being later or never happening."

Having this condition did not have a negative impact on her life. She mentioned that she only has to take a pill for hypothyroidism. What the diagnosis truly did for her was allow her to have a better understanding of herself. She said, "I now understand my physical and emotional characteristics. In terms of connecting with individuals with Turner Syndrome, I have connected online with a few individuals that have Turner Syndrome but I have never met another girl or woman with Turner Syndrome in person."

Why does she feel early diagnosis of this condition is so important? She said, "Early diagnosis of Turner Syndrome and other conditions is so important because early diagnosis can save and improve lives. It can help doctors and patients become better prepared for medical situations and can prevent some medical situations from arising. For Turner Syndrome specifically, it can help to understand things such as skeletal, ear, and heart defects. Knowing about the potential of these issues can help to start preventative medicine and help to see more potential issues early on when they are more treatable. Also specifically for Turner Syndrome girls can go on growth hormones to grow taller but only if it is diagnosed when they are children."

Early diagnosis comes with increased awareness of the condition. February was recently established as Turner Syndrome Awareness Month in the state of New Jersey after years of advocacy work by the Turner Syndrome Foundation, a nationally recognized nonprofit organization dedicated to instilling a greater understanding of this condition in educators and medical providers. The mission now is to have February established as Turner Syndrome Awareness Month across the United States.

Why is this awareness month so important? Catrina explained, “An awareness month is so important because many people, including health care professionals, do not know about Turner Syndrome. Creating awareness will help to increase knowledge about this condition and therefore more help for those with it and affected by it. Early diagnosis can lead to prevention of other medical issues and catching medical issues when they are treatable. Also, by having an awareness month it helps to bring together those that are affected by Turner Syndrome to meet one another and share their stories. This helps boost morale for those who truly need it.”

The foundation’s end of the year giving campaign is now active. Consider supporting the efforts of the Turner Syndrome Foundation with a kind donation. Donations can be made online by visiting <https://app.etapestry.com/cart/TurnerSyndromeFoundation/default/item.php?ref=4448.0.2116649>, mailing a check to PO Box 726, Holmdel, NJ 07733, or by calling 1-800-594-4585.