

The Boston Globe

Health &
wellness

Women with Turner Syndrome tell their stories



JOANNE RATHE/GLOBE STAFF

Miriam Beit-Aharon, who has Turner syndrome, with her mother, Claudette Beit-Aharon. Claudette is the editor of a collection of coming-of-age stories by women, including her daughter, who have the syndrome, titled “Standing Tall With Turner Syndrome.”

By Alyssa Botelho | GLOBE STAFF APRIL 07, 2014

Most kids are thrown into puberty whether they like it or not. But Miriam Beit-Aharon, who has a rare genetic disorder, made a choice to enter womanhood.

Beit-Aharon has Turner syndrome, a disorder that stunts sexual development and causes infertility in about one in every 2,500 female births. She didn't start puberty until she began taking estrogen therapy in high school, a transition that the now 22-year-old wrote about in her application to the University of Massachusetts-Amherst, where she received her diploma last May.

Her application essay is now part of a collection of coming-of-age stories written by 18 women with the syndrome. Talking about Turner, Beit-Aharon says, means talking frankly about womanhood, sex, relationships, and raising kids — topics not all doctors discuss enough. The voices in the book “Standing Tall With Turner Syndrome” are meant to fill that silence.

“These girls want to know but they're afraid to ask,” said Claudette Beit-Aharon, Miriam's mother and the editor of the book.

Our biological sex is determined by a pair of chromosomes — gene-carrying bundles — inherited from our parents at the moment of conception. Boys get an X chromosome from mom and a Y chromosome from dad, while girls get an X from both. Those with Turner only have one functioning X chromosome. The second X chromosome is either abnormal or completely missing. More than 90 percent of women with Turner, which affects 70,000 in the US, lack normal ovarian function and have reduced fertility.

Miriam Beit-Aharon was born full-term and healthy, but she was small at 5½ pounds and couldn't seem to gain any weight.

“She was eating like a horse but she wasn't doing anything with it,” Claudette Beit-Aharon said. “And so the odyssey of trying to figure out what was wrong began.”

As many doctors' visits passed and her friends grew taller, Miriam fit into the same coat four winters in a row. At age 9 she was 3 feet 8 inches tall, the size of a 4-year-old. That's when a savvy nurse at a new pediatrician's office urged Claudette to take Miriam to a pediatric endocrinologist. There, her chromosomes were mapped and it was discovered that she had a missing X chromosome.

Miriam Beit-Aharon now has no obvious outward signs of having Turner, except perhaps for her trademark short stature — she is now 5 feet tall. But a closer look reveals low-set ears, which give her recurring ear infections, a double row of eyelashes, and slightly crooked elbow joints. Others with Turner suffer from heart defects, kidney problems, hearing loss, and high blood pressure, and many have loose “webbed” skin at their necks due to inflammation at birth.

The majority of women with Turner have typical intelligence, though up to 5 percent suffer from severe



learning disabilities. Some may have trouble with tasks that require spatial awareness. Some may also have trouble with math and understanding nonverbal cues.

Luckily, most symptoms associated with Turner can be remedied. Miriam Beit-Aharon began taking daily injections of growth hormone immediately after she was diagnosed, and later, an estrogen-progesterone combination used in birth control pills to begin menstruating (though that doesn't reverse infertility). Women with Turner who suffer from heart and kidney complications undergo the same treatment as those who don't have the syndrome.

Less easily fixed are the social complications, said Marsha Davenport, a pediatric endocrinologist at the University of North Carolina.

"These girls overcome major health problems incredibly well," she said, but the hurdles of dating, developing a healthy sex life, and building families can be more difficult to overcome. "The social difficulties really do seem to trump everything else."

These were the challenges that Claudette Beit-Aharon, a part-time English teacher, asked to be addressed when she sought essays from women with Turner living in the US, Canada, and the United Kingdom. Eighteen women, age 22 to 58, answered the call. One talked frankly about her first time having sex and the path toward satisfying intimacy. A few described the disappointment of relationships that didn't work out. Some wrote about the jealousy of not being able to conceive children, others about the joys of marriage and adoption. Many included the stories of their diagnoses and milestones in their treatment.

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The majority of women with Turner have typical intelligence, though

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of women with more severe forms of the disease can have major learning disabilities. Many girls have some trouble with math, spatial awareness skills such as following a map, and understanding nonverbal cues such as facial recognition

SOURCE: Turner Syndrome Society of the United States and the MGH Turner Syndrome clinic

One of the writers is Susan Lazar, a 53-year-old psychotherapist in Teaneck, N.J. Lazar had not met anyone else with Turner until she was in her late 40s, and had never written about her syndrome until Claudette Beit-Aharon's invitation. She decided to write about the hurdle of telling a romantic partner about having Turner.

"A few months after we started dating, me and my now husband were watching 'Raising Arizona,' a comedy about a couple who can't have kids, and I started to cry," Lazar said. With her husband's willingness to "find a way," she writes, they adopted two children, who are now 23 and 21.

She writes too, about what she calls "the whole sex thing."

"You're missing a chromosome and you can't get pregnant so you might feel less feminine, and that's a big deal," she said. "Psychologically there might be issues to work through, but physically our sex lives should be the same."

Joy Webster, a 48-year-old school librarian and English teacher in Quebec City, Canada, wrote about her life-long struggle to talk about Turner with her family.

"From such a young age I was taught to not talk about it, that nobody needs to know," she said. Her father, who died when she was 26, never knew she had the syndrome, and her brother did not know until just a few years ago. Her mother's long silence, Webster found out in 2010, was because she thought she had somehow caused her daughter's disorder.

"She felt this sense of shame for 45 years, and I didn't know," Webster said. Her mother's understanding that the disease is not inherited, Webster said, has been a first step in talking more openly about the syndrome.

Turner syndrome was first described in 1938 by University of Oklahoma endocrinologist Henry Turner, and the advent of chromosome-imaging techniques in 1959 led to an understanding of the missing X chromosome. But attention on the syndrome has faded as new technologies have allowed closer study of individual genes within chromosomes, said biologist David Page.

“I’ve watched in despair as Turner has fallen off the radar screen,” said Page, who is the director of the Whitehead Institute at MIT and an expert on the sex chromosomes. All proceeds from the book “Standing Tall With Turner Syndrome” will go toward reinvigorating research.

Two questions, Page said, should be given special priority. The first is how the second X chromosome comes to be lost in a fertilized egg as it begins to divide and grow.

The second question is how that X chromosome, once lost, yields the features of Turner. Normal females all carry two X chromosomes, but mysteriously, only one is turned on in each of their cells. And men, who only have one X chromosome, don’t suffer from Turner symptoms. The key, Page said, is to better understand the small fraction of genes that stay active on the “silenced” second X, and their equivalent genes on the Y chromosome.

Others are turning their attention to developing better therapies. Davenport, the pediatric endocrinologist, is leading a clinical trial to understand whether giving low amounts of growth hormone to girls with Turner immediately after birth, instead of in early childhood, can improve their early brain development. Geneticist Angela Lin is part of a team investigating whether in vitro fertilization (IVF) therapy is safe for women with Turner who want to get pregnant. She is a co-director of the Massachusetts General Hospital Turner Syndrome Clinic, where Miriam Beit-Aharon

is a patient.

Miriam Beit-Aharon says she hopes to have kids one day, either by IVF, or adoption, or both. But for now she is focused on her first job, which began last week. A longtime lover of ships and sailing, she is working as a deckhand on the A.J. Meerwald, a restored oyster schooner that runs maritime education programs for the public in southern New Jersey.

She is, her mother described with a laugh, “a short woman on a tall ship.”

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Correction: An earlier version of this story incorrectly stated that Miriam Beit-Aharon had difficulty passing her driving test. The Globe regrets the error.

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