

EMORY NURSING

CURRENT ISSUE

ABOUT

ARCHIVES

CONTACT

ADVERTISE

SUBSCRIBE

ALUMNI

Fall 2019 >> Features

SHARE

Learning about things larger than you

As a School of Nursing doctoral student, Amy Blumling provides much-needed care to a special patient population at Emory Healthcare. Twice monthly, she and other providers see patients at the eXtraordinaryY Clinic, the Southeast's only clinic for children with sex chromosome disorders.

By Pam Auchmutey



Emma St. Germain (right) talks with pediatric nurse practitioner **Sharron Close** (middle) and PhD nursing student **Amy Blumling** (left).

Boys and girls born with these disorders, also called X and Y variations, often face a tough road. As they grow, they may experience physical, developmental, and behavioral symptoms, ranging from mild to severe. Some go on to hold jobs and raise families as adults, while others struggle and require lifelong physical and mental health care.

"There are a lot of stigmas and misconceptions associated with these disorders," says Blumling. "People assume they have something to do with your biological gender or your sexual orientation. They have nothing to do with either."

Sex chromosome disorders develop prior to birth. Parents don't pass them onto their children. The disorders occur randomly when children are born with too many or too few of the chromosomes that determine their sex, normally XY for boys and XX for girls.

Humans typically are born with 46 chromosomes in 23 pairs. In a few births per thousand, some infants are born with a single sex chromosome (45X) and some with three or more (such as 47XXX, or trisomy X, in females and 47XXY, or Klinefelter syndrome, in males). Children may grow to be unusually tall, have an atypical body shape, and have hormonal problems during puberty that affect adult fertility. Learning may be difficult in school. They may feel anxious and insecure, speak and act inappropriately, and have difficulty making friends, all of which can lead to depression, whether they are children, teens, or adults.

Until recently, families of children with X and Y variations had few places to turn for help. Such disorders have largely gone undiagnosed or been misdiagnosed because their symptoms are similar to conditions such as autism or learning disorders. Even with

FEATURES ▼



Next Generation PhD

Meet the new nurse scientist



In a class of their own

The Robert Wood Johnson Foundation's Future of Nursing Scholars



Nursing's PhD Sleuth at CDC

At the Epidemic Intelligence Service (EIS) annual conference in Atlanta this past spring, Nancy McClung 15PhD BSN BA RN was front and center as she briefed the media about a CDC study that showed a marked decrease in the cancer-causing human papillomavirus (HPV) among females in the United States.



Learning about things larger than you

As a School of Nursing doctoral student, Amy Blumling 09OX 12BSN 18G provides much-needed care to a special patient population at Emory Healthcare. Twice monthly, she and other providers see patients at the eXtraordinaryY Clinic, the Southeast's only clinic for children with sex chromosome disorders.

BRIEFS ▶

a correct diagnosis, providers have been hard pressed to help patients because of the dearth of awareness and research on how best to manage sex chromosome disorders.

When Blumling entered the PhD program at the School of Nursing, she initially planned to resume studying the low vaccination rate for HPV (human papillomavirus) among children and the high rate of cervical cancer among women in rural Georgia.

Instead, she refocused her research on sex chromosome disorders after going to a conference held by the Association for X & Y Chromosome Variations or AXYS. Blumling attended the conference with Professor [Sharron Close](#) PhD MS CPNP-PC -FAAN, a School of Nursing faculty member known for her advocacy and pioneering research on children and adults with X and Y variations.

"By the end of the day, I was in tears," Blumling recalls. "I was so moved by the stories of the people who spoke and how desperate they were for help. They face so many issues, especially related to stigma and access to care."

In 2016, Close and [Amy Talboy](#) MD, a developmental pediatrician in Emory's Department of Human Genetics, established the [eXtraordinarY Clinic](#) for patients ranging from infancy to their early 20s. A genetic counselor, nurse navigator, neuropsychologist, pediatric endocrinologist, and adult urologist staff the clinic with Close and Talboy. Blumling is responsible for taking patient histories, performing physical and developmental assessments on babies and young children, and coordinating care with clinic specialists and community resources.

"We help get people connected," Blumling says. "They don't need to see the same providers all the time. But we know from the trajectory of their disorder that they need to start seeing certain specialists, such as an endocrinologist or a gynecologist during puberty."

"Parents really appreciate coming to the clinic," she adds. "It's a beacon of hope for them because they find people who really understand what they're going through. It gives them hope that their children can get the care and services they need."

For her dissertation, Blumling studies males age 18 and older with the rare sex chromosome disorder 48XXYY. Her 22 study participants live in the United States, Canada, the United Kingdom, the European Union, Brazil, and Australia. Through surveys and interviews, Blumling will determine how physical function and psychosocial health affect their quality of life: Are they suffering and if so, how much? What are their biggest issues? What makes their lives better? What makes them worse?

"I want to define what this population looks like and identify their needs and what works best for them," Blumling says. "That's why I love research. It's about learning things that are larger than you."

Related Stories

["Extraordinary Care"](#) (Fall 2018)

["eXtraordinarY: New clinic for X and Y chromosome variations"](#) (11/15/2017)

[Email the editor](#)