



Program Description

The **Nemours eXtraordinary Kids Program**, located at the newly expanded Nemours/Alfred I. duPont Hospital for Children in Wilmington, Del., offers comprehensive care for children with X and Y chromosome variations. Families meet with providers who are experts at treating children with conditions such as Klinefelter syndrome, Turner syndrome and XYY syndrome. Consistent with Nemours' family-centered approach, the treatment team provides a thorough evaluation and specific recommendations to best support each child's medical management, development, academic, social and emotional needs. Our program was inspired by the goal of helping children and families to identify needs and resources and to coordinate care at a single institution. We strive to help all children reach their full potential.

This program generously funded in part by Sandy and Kevin Schindler.

Nemours eXtraordinary Kids Team

Judith Ross, MD, pediatric endocrinologist, is the program director. Highly experienced and renowned in the field, Dr. Ross has devoted the greater part of her career to the study and treatment of X and Y chromosome variations. She provides overall guidance concerning physical features, hormonal/endocrine issues and treatment, neurodevelopmental and genetic aspects, and any related medical concerns. To support Dr. Ross, the program includes this strong team of specialists:

Karen Kowal, PA, is the clinic coordinator. She is a physician assistant with more than 25 years of experience working with families of children with X or Y chromosome variations.

Developmental pediatrics evaluates and monitors development and manages medication for ADHD, anxiety and other behavior disorders.

Genetics, including **Karen Gripp, MD**, division chief, and **Sara Mora, MS**, certified genetic counselor, provides prenatal and genetic counseling, testing, diagnosis and family support.

Psychology and neuropsychology, including **Stephanie Chopko, PhD**, licensed child psychologist assist with behavioral and emotional concerns, cognitive functioning, social skills, learning disabilities and school strategies.

Psychiatry, including **Josephine Elia, MD**, child/adolescent psychiatrist, provides psychiatric diagnosis and treatment including attention deficit disorders, and recommendations for medication, therapy and home/school strategies.

Physical and occupational therapists address fine and gross motor skill deficits as well as daily living skills.

The speech pathologist addresses difficulties with speech, language and expression.

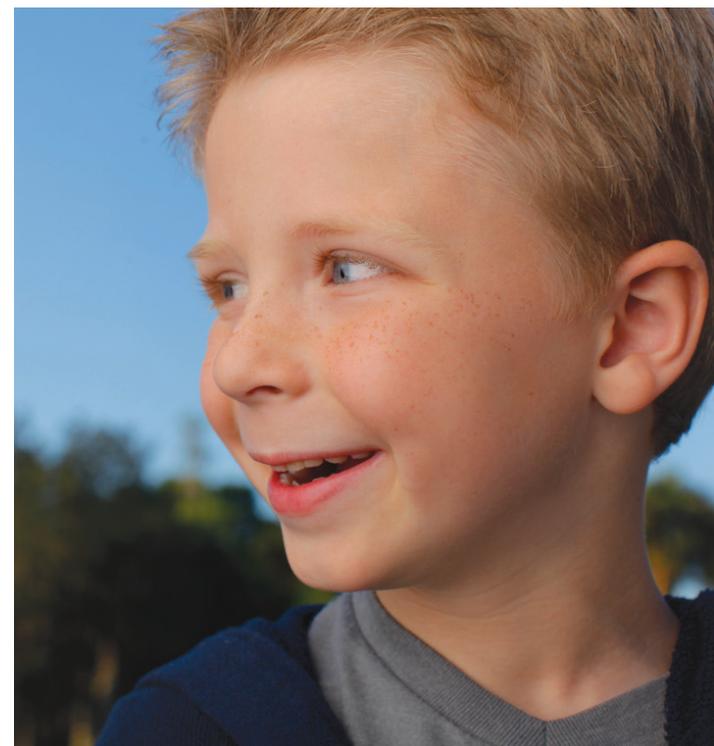
The social worker identifies resources, assists in managing complex cases and offers transition planning.

The nursing navigator coordinates needed services and facilitates appointments.

Parents and family are integral members of the care team. Visits include feedback sessions with the family, discussion of concerns, team impressions and recommendations, and a comprehensive written report.

Appointments

To schedule an appointment with the **eXtraordinary Kids Clinic Program**, please call the clinic coordinator, Karen Kowal, physician assistant, at **(215) 955-1648** or fax (215) 955-1744, or kkowal@nemours.org.



Nemours eXtraordinary Kids Clinic Program

Nemours Alfred I. duPont
Hospital for Children

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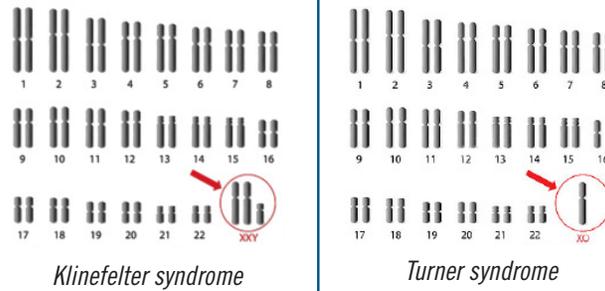
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About X and Y Chromosome Variations

X and Y chromosome variations are common but frequently undiagnosed genetic conditions that differ from the typical chromosome pairings of XX for females and XY for males. Due to the addition of one or more X or Y chromosomes to the normal complement of 46, or the absence of an X chromosome (45,X) the resulting change in total number of chromosomes may impact a child's development and physical characteristics. X and Y chromosome variations are usually identified in childhood by a simple blood test called a chromosome karyotype or through prenatal testing.

These conditions are associated with symptoms that vary from very mild to severe and may include physical, hormonal, neurocognitive, psychiatric and social issues such as delayed pubertal development, speech delays, low muscle tone, attention deficits and social immaturity. There are many misconceptions about people with X and Y chromosome variations. They are often mistakenly assumed to be of lower intelligence, disabled or emotionally unstable. Contrary to these beliefs, most people with X and Y variations are bright, talented, physically active and emotionally healthy. Most symptoms can be addressed successfully with appropriate treatments such as medications and speech and occupational/physical therapy.



Boys and men with *Klinefelter syndrome* (XXY) are usually above average in height and are infertile or have low fertility. Girls and women with *Turner syndrome* (45,X) have short stature and often have fertility problems as well. The X and Y chromosome disorders (also 47,XXY; 47,XXX) are often associated with neurodevelopmental problems such as speech/language delays and mild to moderate learning disabilities, including dyslexia and attention problems. Intellectual disability is not common but may affect a small proportion of patients. Some, but not the majority of people with X and Y chromosome variations, are on the autism spectrum. Most children and adults with these conditions look no different from the population in general.

There are thought to be about 500,000 people in the U.S. with X or Y chromosome variations but only 25 percent are actually diagnosed in their lifetime. At this time, up to 20 percent of diagnoses are made through prenatal diagnosis due to advanced maternal age. Experts predict that within the next five years, screening for X and Y chromosome disorders will be included among the standard newborn screening tests done during pregnancy or in the newborn period. With proper diagnosis and care, children with X and Y chromosome variations can succeed and achieve academically, physically and socially, and lead independent, productive lives.

Heightened awareness of X and Y chromosome variations, as well as advocacy, will help to:

- Work towards a national screening standard for newborns.
- Improve early treatment.
- Provide educational support.
- Work towards the goal of higher education and employment.
- Advance care and research in the field.

First Visit

Before your first visit to the program, we like to gather as much information as possible to help guide your child's evaluation. We will ask you to complete a questionnaire and provide previous test and assessment results, as well as school and educational records. Once we have reviewed the information, we will contact you to schedule an appointment.

During your initial visit, which could take several hours or up to several days, we will discuss your child's specific needs in the following areas. We may also conduct or schedule additional testing.

- physical development
- medical treatment
- motor skills
- speech and language
- social skills
- behavior
- academics

Based on our findings and conversations with family members, we will develop a treatment plan designed to meet your child's needs, provide recommendations, make referrals as appropriate and answer your questions and concerns.