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Physician-scientists seek solutions to reproductive problems related to chromosomal variations

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Approximately one in every 500 to 650 baby boys is born with an extra X chromosome, a variation in their genetic code that until a few years ago was thought to result in infertility in all cases. However, this is no longer the case. A recent conference hosted by NewYork-Presbyterian Hospital/Weill Cornell Medical Center and advocacy organization KS&A focused on raising awareness of the condition and the recent availability of treatments for both children and adults.

As recently as 10 years ago, all men born with an extra X chromosome -- a condition whose classic symptoms are known as Klinefelter syndrome -- were thought to be infertile. Now, new research at NewYork-Presbyterian/Weill Cornell, led by Dr. Peter Schlegel, has pioneered a surgical approach -- a combination of TESE (testicular sperm extraction) and IVF (in vitro fertilization) -- that enables these men to father healthy children approximately 40 percent of the time it is employed.

But this is just the beginning. "Our Department has five scientists who are leading research into Klinefelter's and other chromosomal variations. We are very pleased to join forces with KS&A to raise awareness for these conditions," says Dr. Schlegel, chairman of the Department of Urology and professor of urology and reproductive medicine at Weill Cornell Medical College, and urologist-in-chief at NewYork-Presbyterian/Weill Cornell.

KS&A presented Dr. Schlegel with a "Lifetime Achievement Award" at its recent meeting to honor his infertility treatment advances -- which are rewriting the textbook on Klinefelter syndrome.

"Our current research in the laboratory focuses on understanding the mechanism by which the presence of an additional X chromosome affects sperm production and testosterone synthesis in males with Klinefelter syndrome. These critical and unique studies will allow us to provide improved treatment and management recommendations based on solid understanding of underlying pathophysiology," says Dr. Darius A. Paduch, assistant professor of urology and reproductive medicine at Weill Cornell Medical College and assistant attending urologist at NewYork-Presbyterian/Weill Cornell.

Dr. Paduch, who leads NewYork-Presbyterian/Weill Cornell's translational research into the molecular biology and genetics of 47XXY and Klinefelter syndrome, has assembled a diverse group of scientists from Cornell-Ithaca and Rockefeller University to create a center of excellence in research on Klinefelter syndrome. The Center will be one of the first nationally comprehensive resources for patients of all ages dedicated to variations in the X and Y chromosomes.

The unique meeting brought together national expertise in molecular biology and genetics of reproduction and molecular endocrinology. Each of the scientists who participated in the conference -- Weill Cornell's Dr. Matthew Hardy, an adjunct professor of urology, and Cornell-Ithaca's Dr. Paula Cohen, associate professor of genetics, as well as Dr. Alex Travis, assistant professor of reproductive biology -- offer unique expertise in basic science, which, combined with access to patients seen in the Department of Urology, will allow for multidisciplinary collaborations focused on basic scientific research to improve patient care.

"We are very pleased to join with NewYork-Presbyterian/Weill Cornell, whose important research is a cornerstone for a growing body of knowledge that will help transform the lives of individuals who have X and Y chromosome variations," says Robert Shelton, chairman of the Board of Directors of KS&A. "There is a high likelihood that research into these chromosomal variations will make substantial contributions to knowledge about breast cancer, learning disabilities and other seemingly unrelated phenomena as more is discovered concerning the relevance of genes located on the X and Y chromosomes."

Most individuals have a total of 46 chromosomes. Commonly, men have one X and one Y chromosome; and women have two X chromosomes. But this is not true for everyone. The most common variation is 47XXY in boys and Trisomy X in girls. Without proper interventions, boys born with an extra X chromosome are at a significantly heightened risk of developing the signs and symptoms of Klinefelter syndrome as adults. Unlike other genetic syndromes such as Down's or Fragile X, comparatively little is known about X and Y chromosome variations. NewYork-Presbyterian/Weill Cornell and KS&A are working diligently to change that.

Source: New York- Presbyterian Hospital