RESEARCH – Nation's First CAH Center of Excellence joins in First-ever Multicenter Study of Surgical Outcomes in patients with Nontypical Genital Development

Females with classical CAH are born with a spectrum of urogenital development. Other conditions causing nontypical genitalia formation result from hormonal abnormalities or discordance between a child's genetic sex, gonadal sex, internal sex ducts and/or external genital sex. Surgical treatment approaches for managing many of these patients have received intense scrutiny by patient advocates, based on poor post-surgical cosmesis and sexual dysfunction observed in genitoplasty outcome studies. However, these findings are based on outdated surgical procedures.

Over the past two decades, our understanding of genital neurovascular anatomy along with improvements in surgical technique have provided surgeons specializing in this area with a more modern set of surgical strategies when compared to older techniques with the goal of improving post-surgical outcomes. By far, the largest group of patients to benefit from these more modern approaches is female patients with Classical Congenital Adrenal Hyperplasia. Female patients with CAH make up over 90% of children born with nontypical genitalia. Designated by the CARES foundation, the first Comprehensive Center for Congenital Adrenal Hyperplasia has been at the forefront of medical and surgical care for affected children. The Center is affiliated with New York Presbyterian Hospital's Phyllis and David Komansky Center for Children's Health at the Weill Cornell Medical College, and it is the main referral site for children with CAH worldwide.

As part of the Center's mission to promote clinical research in children with CAH, it is currently enrolling patients to evaluate outcomes of modern genitoplasty as part of a National Institutes of Health study. The NIH grant was awarded to Dr. Amy Wisniewski from The University of Oklahoma Health Sciences Center. This multi-site study comes at a critical point to resolve the debate around the surgical treatment approaches for affected children, and findings from this study may mark a significant change in clinical practice.

The aims of the study include:

1. To determine the effectiveness of modern genitoplasty in improving the cosmetic appearance and functional outcomes in young children born with a spectrum of urogenital development. Outcomes will be ascertained by surgeons who performed the procedures, parents and professionals with expertise in care of these patients.
2. To document urinary tract infections and surgical complications associated with not proceeding with genitoplasty as well as proceeding with current surgical techniques.
3. To examine specific parental adjustment outcomes: depression, anxiety, post-traumatic stress symptoms, perceived illness uncertainty, parent quality of life scores and decisional regret by parents following their child's surgery.

This multi-site study incorporates parents' ratings of surgical results along with their ratings of their own psychological outcomes and their satisfaction or regret with their medical decisions. This multi-disciplinary approach in clinical research is novel and therefore made it a fitting collaboration with our Center. The Center's specialized care and comprehensive team of endocrinologists, surgeons, nurse practitioners, genetic

counselors and psychologists serves as a critical site for this kind of ground-breaking research. Dr. Dix Poppas, Professor and Chief of Pediatric urology serves as Surgical Director and Dr. Maria Vogiatzi Chief of Pediatric Endocrinology serves as Medical Director for the Comprehensive Center of Excellence for CAH. Dr. Poppas is leading this research collaboration at Cornell.

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