O-56 Monday, October 19, 2009 6:00 PM

REFINING ESTIMATES OF NATURAL FECUNDABILITY IN OLDER, REPRODUCTIVE AGE WOMEN. C. K. Tanner, A. Z. Steiner. Obstetrics and Gynecology, University of North Carolina School of Medicine, Chapel Hill, NC.

OBJECTIVE: Natural fecundability (probability of conception per cycle) is most accurately determined through prospective, time-to-pregnancy studies. Previous studies have examined fecundability in women aged 35-40 as a group. We hypothesized that significant heterogeneity would exist within this group. The objective of this study was to refine fecundability estimates for older, reproductive age women.

DESIGN: Prospective time-to-pregnancy cohort study of women aged 30-45.

MATERIALS AND METHODS: Women with no known history of infertility, who identified themselves as trying to conceive for 3 months or less completed a questionnaire at first menses following enrollment (N=131). Women who conceived prior to the study visit completed a similar questionnaire upon notification of pregnancy (N=45). 548 cycles were analyzed. A statistically significant decrease in fertility was observed throughout the study period. Differences in natural fecundability exist within the 35-40 year old group; with a notable decline in fertility observed at 38 years of age.

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A NOVEL MUTATION OF HOXA13 IN A FAMILY WITH HAND-FOOT-GENITAL SYNDROME AND THE ROLE OF POLYALANINE EXPANSIONS IN THE SPECTRUM OF MULLERIAN FUSION ANOMALIES. E. M. Jorgensen, J. I. Ruman, L. Doherty, H. S. Taylor. Obstetrics, Gynecology and Reproductive Sciences, Yale University School of Medicine, New Haven, CT; Obstetrics, Gynecology and Reproductive Science, Mount Sinai School of Medicine, New York, NY.

OBJECTIVE: Hand-foot-genital syndrome (HFGS) is a rare congenital disease characterized by genitourinary defects, urotergal septa, malformations of the distal limbs, and mutations of HOXA13. The severity of HFGS has been shown to be proportional to the length of polyalanine insertions, so other urotergal anomalies could potentially fall into this spectrum, perhaps harboring small polyalanine expansions. The purpose of this study is to characterize the genetic basis of true HFGS versus that of isolated, non-HFGS-related urotergal septa. Additionally, we report a novel mutation found in a family with HFGS.

DESIGN: Case-control study.

MATERIALS AND METHODS: Genomic DNA was extracted from biopsies of septa (non-HFGS patients) or blood samples (HFGS family). The entire HOXA13 gene was amplified in seven overlapping sections using PCR, and the products were submitted for DNA sequence analysis. To check for heterozygosity and to allow sequence analysis of each allele separately, PCR products were further cloned into plasmid vectors using TA cloning. RESULTS: A screen of subjects with idiopathic, non-HFGS-related uterine (n=12) or urotergal (n=5) septa showed no mutations of HOXA13. Affected members (n=3) of a family with clinically confirmed HFGS were heterozygous for a novel mutation of HOXA13. The mutation is a 30-nucleotide insertion beginning at base 269 that expands the third polyalanine tract by 10 alanine residues. An unaffected family member did not show any HOXA13 mutations.

CONCLUSIONS: Without the characteristic hand and foot symptoms, idiopathic septa appear to have a genetic basis that differs from that of true HFGS. An isolated urotergal septum seems to be a distinct condition rather than a mild form of HFGS. When patients present with septa, it is not necessary to subject them to x-rays of the distal limbs or to sequence analysis of HOXA13 unless they show clear signs of the other sequelae characteristic of true HFGS.

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O-58 Monday, October 19, 2009 4:30 PM

FREQUENCY OF OOCYTES AND PRIMORDIAL FOLLICLES IN GONADECTOMY TISSUE SAMPLES FROM PATIENTS WITH GONADAL DYSGENESIS. K. Marquard, D. Cao, P. C. Huettner, A. Rabinov, E. Junghem, V. Ratts. OBGYN-Division of Reproductive Endocrinology and Infertility/Pathology and Immunology, Washington University School of Medicine, St.Louis, MO.

OBJECTIVE: Gonadal dysgenesis with 46XY or 45X/46XY karyotype usually requires gonadectomy due to risk of malignancy. However, little is known about the reproductive potential of the excised tissue, and future techniques to isolate gametes may someday be beneficial. Thus, our objective was to determine whether spermatogonia/oocytes exist in post-gonadectomy specimens from patients with gonadal dysgenesis.

DESIGN: Retrospective Case Series

MATERIALS AND METHODS: Patients who underwent gonadectomy for gonadal dysgenesis/undescended testes at our institution from 1998-2008 were identified. Data collected from medical records included age, operative and physical exam findings, and peripheral karyotype. Microscopic examination of gonadal tissue on hematoxylin & eosin (H&E) slides was...