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Session: Plenary Session
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STUDIES OF THE FREQUENCY OF ASSISTED REPRODUCTIVE TECHNOLOGY (ART) BIRTHS AND TWINNING IN PRADER-WILLI SYNDROME

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Purpose of Study: To determine the association between ART and Prader-Willi Syndrome(PWS) by evaluating the frequencies of ART-births in three distinct molecular groups, compared with ART-births in the USA. To evaluate the natural frequency of twinning with and without ART in the PWS group.

Methods Used: Data was collected from surveys administered by:

1) The Prader-Willi Syndrome Association of the USA (PWSAUSA)
2) The sponsored Rare Diseases Clinical Research Network (RDCRN) Natural history PWS and morbid obesity study
3) Review of medical records from PWS patients seen by the Genetics Division at UCIMC

Summary of Results: Total number of PWS patients was 1,888. Total frequency of ART was 2.3% (44/1,888), 95% confidence interval (CI) 1.62%–2.98%. There was no statistical significant difference in the frequency of ART-conceived PWS patients. chi squared = 1.024 2 df p = 0.599. However the difference in frequencies of the genetic subtypes in the ART-conceived patients and naturally conceived patients was statistically significant (p = 0.019). By comparison ART conceived patients were more likely to have UPD and imprinting center defects. This study also demonstrated that there was no increased frequency of natural twinning without ART in the PWS population above the U.S. population.

Conclusions: Studies have concluded that the effects of ART procedures may be restricted to imprinting disorders in which the maternal allele is hypomethylated or in which an imprinting defect accounts for a significant proportion of affected cases. This study shows a significant increase in UPD in PWS from ART-births suggesting an association with ART and UPD. At this time, the mechanisms causing this association have not been fully established. Growth factor genes are implicated in the growth disturbance of the fetus and the placenta. Certainly advanced maternal age is a cause of increased risk for trisomy. One of the mechanisms for maternal disomy is trisomic rescue. Women of increased age have a higher occurrence of maternal uniparental disomy and also have a higher likelihood of pursuing ART.