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MOSAIC TURNER SYNDROME: PREIMPLANTATION GENETIC TESTING FOR ANEUPLOIDY OUTCOMES

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Title:
MOSAIC TURNER SYNDROME: PREIMPLANTATION GENETIC TESTING FOR ANEUPLOIDY OUTCOMES

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Preferred Presentation Type:
Oral or Poster

Study Type:
Retrospective Cohort Study (includes comparator groups)

Category - Subcategory(ies)s:
Genetics: Genetic Counseling
Genetics: Genetics General
Genetics: PGT

References:

1. Bernard V, et al. Spontaneous fertility and pregnancy outcomes amongst 480 women with Turner syndrome. Hum Reprod. 2016; 31(4):782-8.
2. Giles J, et al. Preimplantation genetic testing for aneuploidy in patients with partial X monosomy using their own oocytes: is this a suitable indication? Fertil Steril. 2020; 114(2):346-53.

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Trainee: Yes

Abstract Category:
All Other Categories

Applied for the In-Training Award for Research

Abstract Text:
OBJECTIVE: Phenotype and reproductive potential in patients with mosaic Turner Syndrome (TS) (45,X/46,XX) show inconsistent data regarding fertility rates and the risk of chromosomal aneuploidy in offspring [1,2]. Most research has focused on spontaneous pregnancy outcomes and pregnancy safety. Limited data exists on mosaic TS patients undergoing in vitro fertilization (IVF) with preimplantation genetic testing for aneuploidy (PGT-A). This study evaluates embryo ploidy in patients with mosaic TS.

MATERIALS AND METHODS: This multicenter retrospective cohort study included patients with mosaic TS who underwent IVF/PGT-A from 2016 to 2024. PGT-A was performed with next generation sequencing. Patients were characterized based on karyotype result, with >10% or ≤10% 45,X cells, as ≤10% mosaicism may in some cases represent a normal age-related X chromosome loss. Prior to considering pregnancy, patients underwent appropriate cardiac testing. Mosaic TS patients were matched 1:3 to controls with normal karyotypes, by age, anti-Mullerian hormone (AMH), and body mass index (BMI). The primary outcome was embryonic ploidy. Subgroup analyses evaluated patients with >10% and ≤10% mosaicism. Chi squared was used for comparative statistics. Logistic regression fitted with generalized estimating equations was used to adjust for confounders (age, AMH, BMI, treatment year, and IVF center) and estimate odds ratios (OR) with 95% CIs for the association between mosaic TS and ploidy.

RESULTS: A total of 33 IVF/PGT-A cycles from 21 patients with mosaic TS were included. Of these, 20 cycles were from 12 patients with ≤10% TS mosaicism (mean 8.2%, range 3.3-10%) and 13 cycles from 9 patients with >10% TS mosaicism (mean 16.5%, range 12-35%). No significant difference in aneuploidy was observed between mosaic TS patients and controls (37.9% vs 37.7%; OR 0.81 [0.54-1.23]). Among mosaic TS patients, those with >10% mosaicism had higher odds of aneuploidy compared to those with ≤10% mosaicism (41.3% vs 35.1%; OR 1.94 [1.13-3.31]). Patients with >10% TS mosaicism had significantly higher odds of aneuploidy compared to controls (41.3% vs 37.3%; OR 1.76 [1.10-2.83]). Similar odds of mosaicism (OR 0.84 [0.21-3.37]) and indeterminate result (OR 0.29 [0.03-2.65]) were observed between groups.

CONCLUSIONS: Patients with mosaic TS demonstrated a higher risk of embryonic aneuploidy when karyotype showed >10% mosaicism. This increased risk may be attributed to a greater proportion of germ cells originating from 45,X cell lines, which lack the 2nd X chromosome essential for proper pairing and segregation during meiosis. Consequently, patients with higher levels of mosaicism may be more susceptible to meiotic errors, leading to an increased incidence of embryonic aneuploidy. Clinicians should counsel patients with mosaic TS, particularly those with >10% mosaicism, on their heightened risk for chromosomal abnormalities and guide reproductive decision-making accordingly.

IMPACT STATEMENT: Patients with >10% mosaic Turner syndrome exhibit higher rates of embryonic aneuploidy compared to those with a normal karyotype or ≤10% mosaicism, suggesting that PGT-A outcomes are influenced by the degree of chromosomal mosaicism.

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Biographical Sketch Kate Devine, M.D. serves as US Fertility's Medical Director and Chief Research officer and endeavors to expand the knowledge base of our field on behalf of patients striving to achieve their dreams of family. Dr. Devine supports the faculty of the six USF-affiliated Reproductive Endocrinology and Infertility fellowships. Furthermore, she oversees a culture of excellence in the medical care of our patients across the USF Network. Dr. Devine, a board-certified Reproductive Endocrinologist, cares for patients in Washington, D.C., and has served as Research Director at Shady Grove Fertility for nearly a decade. She is Clinical Professor of Obstetrics and Gynecology at George Washington University and Associate Program Director for the Reproductive Endocrinology and Infertility Fellowship at the National Institutes of Health (NIH). Dr. Devine serves as Chair of the Society for Reproductive Endocrinology and Infertility (SREI) Research Committee, Chair of the Society for Assisted Reproductive Technology (SART) Quality Assurance Committee, and on the Editorial Board of Fertility and Sterility. She has authored more than 50 peer-reviewed publications, has received numerous research grants and awards, including the 2023 ASRM New Investigator Award, and has spoken nationally and internationally on the topic of her research, which focuses primarily

on endometrial receptivity and optimization of frozen embryo transfer protocols. She earned her BA from Columbia University and MD from Albert Einstein College of Medicine. She completed residency at New York University Medical Center and fellowship at the NIH.
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Biographical Sketch Phillip Romanski, M.D., M.Sc., is a Reproductive Endocrinology and Infertility physician at RMA of New York in Manhattan and is a faculty member at the National Institutes of Health. He is an expert in family-building including the evaluation and management of female and male infertility, third-party reproduction, and fertility preservation. Dr. Romanski completed his residency in Obstetrics and Gynecology at Harvard Medical School (Brigham and Women's Hospital/Massachusetts General Hospital) and his fellowship in Reproductive Endocrinology and Infertility at the Weill Cornell Medical Center/NewYork-Presbyterian Hospital. Dr. Romanski additionally serves as the Associate Research Director for US Fertility and has authored over 60 peer-reviewed research publications with a particular interest in patients with a history of unsuccessful treatment and patients with diminished ovarian reserve. In recognition of his research contributions, he has received multiple national awards and has subsequently been invited to speak at both national and international conferences to present his work.
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Organization Name	Relationship Type	Who has this Relationship?	
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Biographical Sketch Dr. Lucky Sekhon is a double board certified OBGYN and REI at RMA of New York. She is passionate about empowering people by educating them about their reproductive health and options for fertility preservation and family building.
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