Current Practices and Perspectives of Genetic Counselors and Reproductive Endocrinologists Regarding Transfer of Mosaic Embryos

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With the recent transition in testing methodology used for preimplantation genetic testing for aneuploidy (PGT-A) from array comparative genomic hybridization to next generation sequencing, mosaic embryos are being identified more readily. Given the limited clinical guidance and information regarding outcomes after the transfer of mosaic embryos (TME), a mosaic test result can present challenging scenarios for providers and patients. The current landscape of this area of reproductive medicine must be described before a consensus can be determined and areas for improvement can be identified. This cross-sectional descriptive study aimed to define the current practices regarding TME as reported by prenatal and/or infertility genetic counselors (GCs) and reproductive endocrinologists (REs). In addition, it aimed to determine GCs’ and REs’ perspectives on patient education, informed consent, decision making and clinical guidance with regard to the TME. An invitation to participate in the electronic survey was distributed to GCs through the National Society of Genetic Counselors listserv and to REs via an email from the principal investigator. A total of 223 responses were analyzed consisting of 194 GCs and 29 REs. Data analysis showed that infertility GCs practices and perspectives were more consistent with REs than non-infertility GCs. However, regardless of specialty, responses showed little to no consensus among providers regarding their perspectives on this topic. Overall, respondents reported feeling more comfortable with pre-test PGT-A counseling compared to counseling about TME. Furthermore, a majority of respondents indicated that additional consensus and/or guidance is needed for several topics related to TME, such as when to discuss the possibility of mosaic embryos with patients, when the decision should be made whether or not to transfer mosaic embryos and prioritization when multiple mosaic embryos are available. These results support the urgent need for additional consensus and guidance regarding best practices when mosaic embryos are identified.

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