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Reproductive decisions made by balanced chromosome translocation carriers

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I. Introduction

Balanced chromosome translocation carriers are at increased risk for pregnancy loss and at risk to have offspring with chromosome abnormalities. When translocation carriers are identified, various options are available to these patients including: preimplantation genetic testing for structural rearrangements (PGT-SR), prenatal diagnosis, and gamete donation. The decision for further testing may depend on multiple factors including personal preference and resource availability.

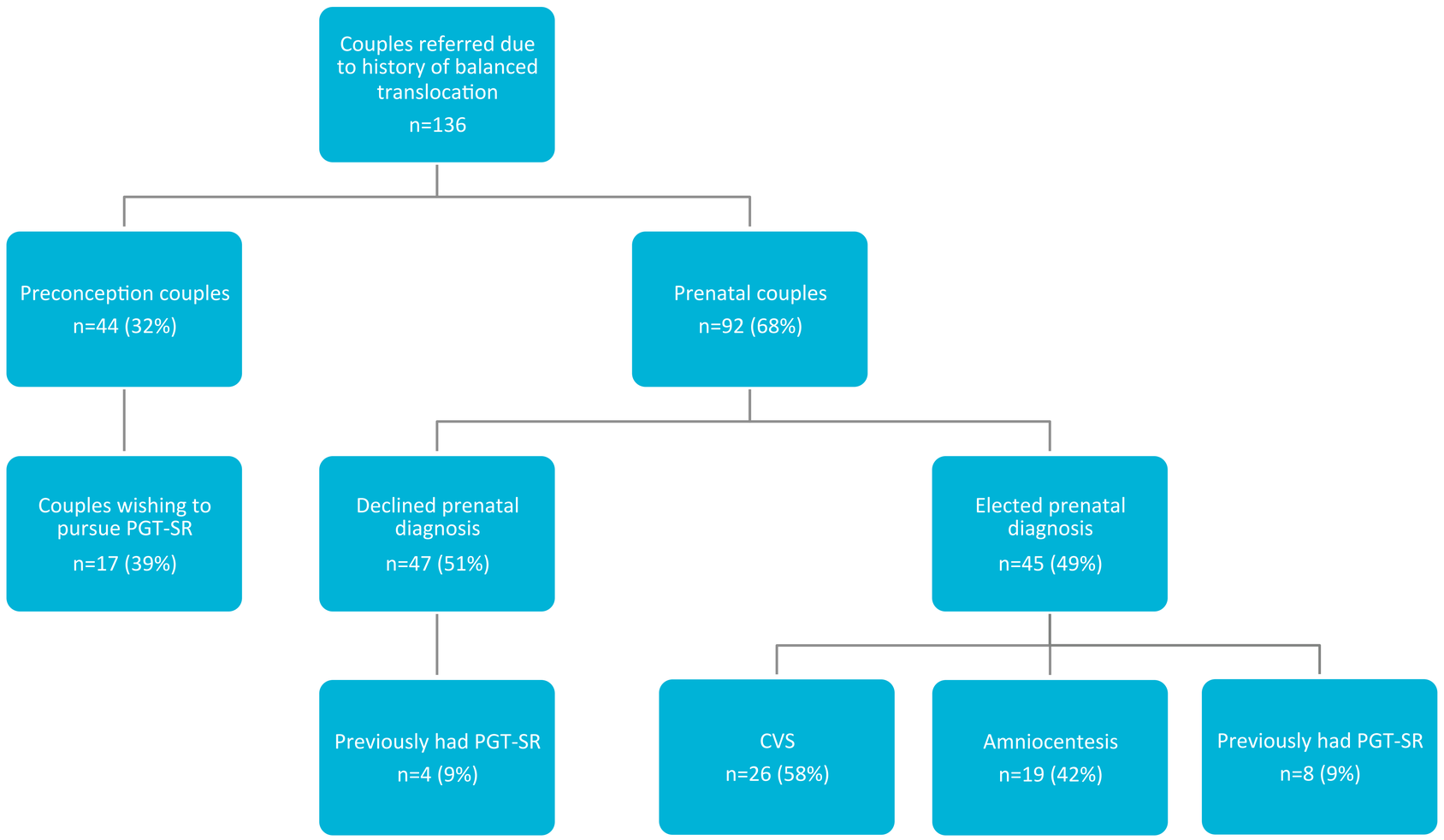
II. Methods

We undertook a retrospective chart review of patients referred for genetic counseling who reported a personal history in either genetic parent of a balanced translocation. During a 20 month time period in 2018 – 2019, a total of 136 patients reported this history. Of the 136 couples, 44 (32%) were preconceptual and 92 (68%) were already pregnant. The mother was the carrier in 74% of the cases and the father in 26% of the cases. All patients had both preconceptual and pregnancy options discussed for their current or future pregnancies as well as a review of any PGT-SR testing previously performed. This study was intended to assess the decisions made by translocation carriers seen for genetic counseling.

III. Results

Of those couples seen preconceptually, 17 (39%) indicated that they wished to pursue PGT-SR. Of those seen prenatally, 8 (9%) had PGT-SR previously performed. Of the patients who had PGT-SR previously performed, half of them chose to pursue prenatal diagnosis with 3 having chorionic villus sampling (CVS) and 1 having amniocentesis. In the total cohort of prenatal patients, regardless of whether they had PGT-SR, 49% elected prenatal diagnosis and 51% declined. In the group of patients who pursued prenatal diagnosis (n=45), slightly more than half of those, 58% had CVS compared to 42% who had amniocentesis.

Figure 1. Summary of Study Population



IV. Conclusions

Patients who are referred for genetic counseling with a known history of balanced chromosome translocations have greater risks and multiple options to evaluate these risks. The decisions regarding prenatal diagnosis were split almost evenly between patients who chose to pursue prenatal diagnosis and those who declined. The reasons for their decisions were not assessed in the current study. Not surprisingly given the availability of PGT-SR and financial cost of the testing, most pregnant patients did not have PGT prior to pregnancy. This is in contrast with the higher percentage of preconception patients who indicated they wanted to pursue PGT-SR prior to a future pregnancy. Regardless of the testing options chosen, this study did not show a clear trend in testing decisions for patients who are balanced translocation carriers. Further studies are necessary to determine trends and patient perceptions toward preconception and prenatal diagnosis.

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