**Date:** November 7, 2026 15:10-15:20
**Venue:** Crystal Court II, Hopewell Hotel
**Session:** Parallel Session 2A: Women's Health and Reproductive Genomics

Abstract No.: 45

Women's Health and Reproductive Genomics

# Karyomapping-Based Preimplantation Genetic Testing for Structural Rearrangements: Subtype-Stratified Embryo Classification, Carrier Status Resolution, and Pregnancy Outcomes

First Author: Patthorn JAIKLA

Co-Author: Wanwisa Suksalak | Wipawee Tangsee | Chutimon Pomsuwan | Nadsarin Tochoowong | Sujin Chanchamroen | Wiwat Quangkananurug

Submitted_by Institution: Next Generation Genomic Co., Ltd., Bangkok, Thailand

*Objective:*

Preimplantation genetic testing for structural rearrangements (PGT-SR) is most commonly performed by next-generation sequencing, which detects chromosomal imbalance but cannot distinguish balanced-carrier embryos from those that have not inherited the rearrangement. Karyomapping resolves carrier status alongside imbalance detection through genome-wide single nucleotide polymorphism (SNP) haplotyping and linkage analysis, in a single workflow applicable across rearrangement subtypes. Cohort-level clinical data remain limited. We report embryo classification and pregnancy outcomes from all consecutive PGT-SR cycles performed at a single reproductive genetics center.

*Methods:*

A Retrospective cohort study of all PGT-SR cycles performed from 2020 to 2026. Karyomapping was performed using the HumanKaryomap-12 and KaryoMap v2 BeadChip, with haplotype linkage analysis conducted using BlueFuse Multi and the kMap software (Illumina), respectively. Couples carrying balanced reciprocal translocations, Robertsonian translocations, or pericentric inversions were included. Trophectoderm biopsy at the blastocyst stage was followed by vitrification and frozen embryo transfer. Each embryo was classified for rearrangement status as unaffected, balanced carrier, or unbalanced, with concurrent aneuploidy screening. Outcomes were clinical pregnancy and live birth per transfer cycle.

*Results:*

Ninety-seven blastocysts from 9 retrieval cycles in 7 couples (4 reciprocal translocations, 2 Robertsonian translocations, 1 pericentric inversion; mean maternal age 33.0 years, range 27–48) were analysed. Unbalanced embryos accounted for 52.6% (51/97), varying markedly by subtype: 66.7% (48/72) in reciprocal translocations, 14.3% (3/21) in Robertsonian translocations, and 0% (0/4) in the inversion carrier. Among 46 embryos balanced for the rearrangement, carrier status was determined, with an overall unaffected-to-carrier ratio of 1.4:1 (27:19), similar to that observed in reciprocal translocation carriers (14:10), and 1:1 (9:9) in Robertsonian carriers. Overall, 35.1% (34/97) of all embryos analysed were transferable, comprising embryos with unaffected or carrier status after exclusion of concurrent aneuploidy. Nine embryos were transferred over 7 cycles (5 single, 2 double), achieving a clinical pregnancy rate of 71.4% (5/7; 95% CI 29–96%). All five pregnancies resulted in live birth, with no pregnancy loss.

*Conclusions:*

Karyomapping-based PGT-SR yielded a conclusive result for every blastocyst tested while simultaneously resolving balanced-carrier status, with observed unaffected-to-carrier ratios matching theoretical expectation. Unbalanced rates differed substantially across rearrangement subtypes, information directly applicable to pre-cycle counselling. Because carrier assignment relies on haplotype phasing, cycles yielding no unbalanced embryos — as occurred in the inversion carrier — require an external confirmed-carrier reference sample. All pregnancies achieved in this cohort progressed to live birth without loss.

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