CHROMOSOME INSTABILITY AT THE BLASTOCYST STAGE

The pioneering work by J. Vermeesch's group in 2009 (1) revealed that chromosome instability is common in human cleavage-stage embryos. The single cell analyses were conducted by microarray technology. Several confirmatory papers followed.
Handyside et al. (2024), in a study published on **bioRxiv** (2), performed a similar investigation across 342 cycles of Preimplantation Genetic Testing for Monogenic diseases (PGT-M), combining parental haplotyping with SNP intensity analysis. This cost-effective assay enabled the precise identification of meiotic and mitotic whole chromosome and segmental gains and losses, along with their parental origins.

The authors concluded: "Meiotic aneuploidies were predominantly whole chromosome aneuploidies of maternal origin and increased with maternal age. Mitotic aneuploidies (with normal parental haplotype patterns) were mainly segmental imbalances."

The tables included in the paper provide a rich source of data for researchers in this field.

In the Discussion, the authors address the challenges faced in PGT practice, making this study an engaging and essential read for those involved in preimplantation genetic testing.

1. https://www.nature.com/articles/nm.1924
2. https://www.biorxiv.org/content/10.1101/2024.11.17.623999v1