PARENTAL GENOME UNIFICATION AT THE STARTING POINT OF THE EMBRYO IS HIGHLY ERROR PRONE

Cytogenetic analysis of spontaneous abortions revealed, in the 1970s, that half of them were due to aneuploidy, and this was thought to be the tip of the iceberg. For example, autosomal trisomies lacked the expected counterpart, the monosomies. Since then, the many stages from gametes to the early development of the embryo have been investigated. In this regard, an important seminal work was the one published in 2009 by the group of J. Vermeesch (Nature Medicine). They showed that chromosomal instability is common in human embryos at the cleavage stage and, more importantly, that normal development of the embryo can occur by selection against abnormal blastomeres. This conclusion has been supported and clarified by the recent work of Coorens et al. (Nature, 2021) who found that abnormal cells can be segregated into the placenta.

An article by Cavazza et al. (Cell, 2021) now reveals that another important stage, underlying the development of the embryo, is involved in the generation of aneuploidy and chromosomal abnormalities in general.

After fertilization, the parental genomes polarize towards each other and the chromosomes of the two pronuclei cluster at the pronuclear interface, ready for unification. This process, the paper points out, is highly error-prone.