SPERM GENOMES

The first human aneuploidy, the trisomy of chromosome 21 was described by Lejuene in 1959. Subsequent research showed that about 50% of spontaneous abortions have a chromosome anomaly. Most of these were trisomies; monosomies, theoretically expected to equal trisomies, were absent (with the exception of a special chromosome, the X). The obvious hypothesis was that monosomies were incompatible with normal development of the early embryo. Researchers were eager to investigate the chromosome constitution of embryos at very early stages. The group of J.Vermeesch has successfully analyzed human cleavage-stage embryos, disclosing their high chromosome instability. Aneuploidy in sperm cells has been studied in the past using different approaches (sperm–hamster oocyte fusion system; FISH).

Since then, technology has made it possible to sequence the genome of single cells. Using an ingenious procedure Bell et al. (Nature), have fully sequenced 31,128 human sperm and their 20 donors. They found 787 whole-chromosome aneuploidies and 133 chromosome arm-scale gains and losses (2.5% and 0.4% of cells, respectively). They also obtained interesting data on correlation between crossovers and non-disjunction.

In evaluating these results in the wider context of human reproduction, one has to remember that female meiosis, where the rate of aneuploidy increases with maternal age, is more prone to non-disjunction. As mentioned above, chromosome gain and loss frequently occur also after fertilization, in cleavage-stage embryos