PLACENTAL MOSAICISM

Chromosomal mosaicism present in the placenta but absent in the fetus is a well-known phenomenon. In a recent article in Nature, Coorens et al. sequenced 86 bulk placental samples and 106 microdissected placental samples in order to reconstruct the phylogeny of placental cells. The task was achieved by monitoring the somatic mutations that cells accumulate during their replication. The authors point out that the number of these mutations, including copy number variations, in the placenta is unmatched in any other normal tissue. One of the most interesting findings is the discovery of bottlenecks that allow normalization of zygotic aneuploidy and segregation of abnormal cells into the placenta. This was very evident in a case of chromosome 10 trisomy, where the extra chromosome was contributed by the mother: trisomy 10 was detected in a bulk sample; the embryo, the umbilical cord and a portion of the placenta contained only two chromosomes 10 but both of maternal origin.