SPERM MOSAICISM

It is well known that in humans new mutations arise more frequently in the father than in the mother (Goldman et al. Nat Genet 2016). A recent article in Trends in Genetics (Breuss et al., 2021) analyzes the different types of mutations found in spermatozoa (sperm mosaicism) and their mechanism of origin. The authors distinguish three types of mutations:
Type I: those that arise during meiosis (not age dependent)
Type II: those that arise in spermatogonia (age-dependent)
Type III: those that arise during paternal embryogenesis, and therefore contribute stably to sperm throughout life.
These mutations can contribute to disease but are also the main source of genetic variation in the population. Furthermore, their identification also has consequences for clinical practice.