KARYOTYPING HELPS TO FIND A NOVEL GENE FOR MALE FERTILITY

Several hundred genes are mutable to cause male infertility, and many novel genes remain to be identified. Karyotyping can help to find these, as shown in the January 2, 2020 issue of the American Journal of Human Genetics by a group of researchers from Boston, Manchester, Münster and Stanford (Schilit et al.). They studied a male patient with unexplained severe oligospermia who had a balanced translocation t(20;22)(q13.3;q11.2). By analysing the expression of all genes in the TADs that were disrupted by this translocation, they found a 20-fold overexpression of the *SYCP2* gene on the chromosome 20 segment in the der(20)t(20;22). 4C-Seq analysis indicated that this was caused by adoption of an enhancer in the segment derived from chromosome 22. *SYCP2* encodes synaptonemal complex protein 2, and its overexpression in a budding yeast model caused meiotic arrest by disruption of the synaptonemal complex. Together with the identification of exonic deletions in *SYCP2* in three other infertile males, one of which also caused meiotic arrest, this study adds *SYCP2* to the growing list of male fertility genes. In this case, karyotyping was essential to find the causative gene.