WHY KLINEFELTER INDIVIDUALS ARE AZOOSPERMIC?

Klinefelter patients are azoospermic because their fetal germ cells (FGC) are arrested at an early stage. An article published recently in Nature (1) shows that the arrest is linked to the lack of X inactivation, resulting in an imbalance in gene expression. Genes related to pluripotency, the WNT pathway and the TGF- β pathway, are upregulated, while genes involved in FGC differentiation are downregulated.

1. https://www.nature.com/articles/s41586-024-08104-6