THE X-CHROMOSOME TELOMERE-TO-TELOMERE, NO GAPS

The initial human genome reference map was full of gaps, that is, "holes" in the sequence that were impossible to clone in bacteria. In subsequent years most, but not all, gaps were filled (see hg38); the centromeres remained as gaps. Centromeres are full of repetitive sequences and are highly polymorphic in the population. Then new technologies that were able to yield ultra-long (greater than 100 kb) reads became available. The fully haploid cell line CHM13hTERT also proved to be very useful in patching the gaps. This Nature paper reports the first human chromosome, the X chromosome, the sequence of which extends from pter to qter with no gaps.