



Fig. 4. Characterization of Y-chromosome structures by FISH. (A) Probe hybridization sites shown below a schematic of the reference Y chromosome. (B) Metaphase FISH assays on a control sample, AMC1574, and AMC0972 demonstrate the copy number and chromosome-arm location of hybridized probes. The resolution of metaphase FISH cannot distinguish signals of closely spaced hybridization sites. (C) Schematic of pseudoisoYp chromosome in AMC1574 formed by inter-sister-chromatid crossing-over between IR1 on Yp and IR1-b4 on Yq. Predicted FISH probe hybridization sites are shown. (D) Two-color interphase FISH demonstrates the rearrangement of AZFc amplicons in two cases with pericentric inversions. (E) Schematic of pericentric Y-chromosome inversion in AMC0972 and predicted FISH probe hybridization sites. See Supplementary Fig. S1 for two-step model: intrachromatid gr/rg inversion in AZFc followed by intrachromatid recombination between IR1 on Yp and IR1-b3 on Yq.