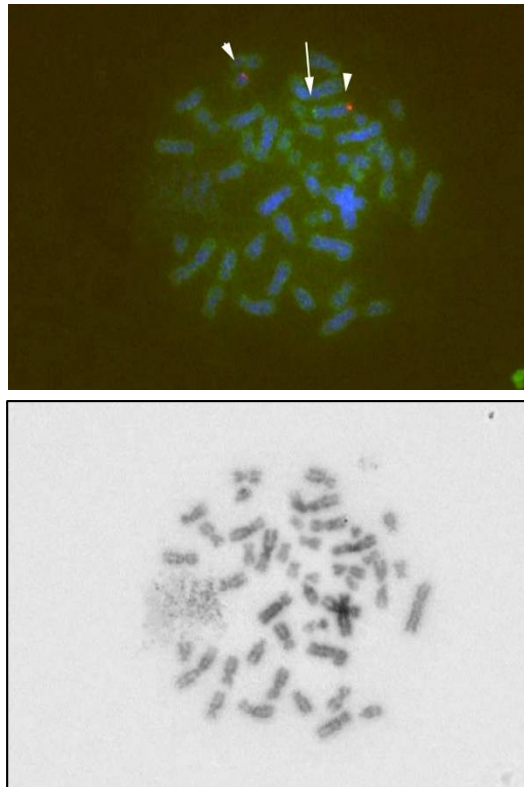
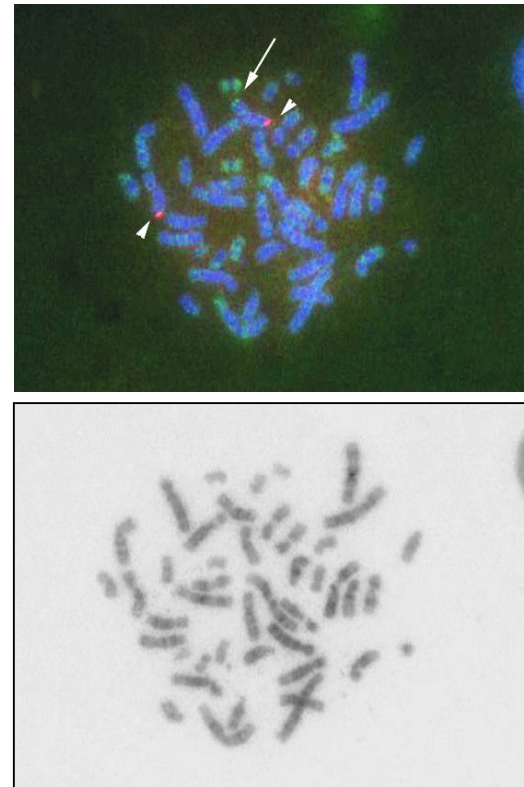


Case 1 (A-I-1)



The daughter of case 1 (A-II-1)



Supplementary fig. 1. Fluorescent *in situ* hybridization of family A. White arrows depict *SHOX* (green signals), and white arrowheads indicate the Xq/Yq telomere region (red signals). The *SHOX*-containing PAR1 deletion was located on the Y chromosome of case 1 (A-I-1) and on the X chromosome of his daughter (A-II-1). The lower panel shows the results of 4',6-diamidino-2-phenylindole, dihydrochloride (DAPI) staining.