

An abnormal terminal X-Y interchange accounts for most but not all cases of human XX maleness

Petit, Christine

de la Chapelle, Albert

Levilliers, Jacqueline

Castillo, Sylvia

Noël, Bernard

Weissenbach, Jean

To determine if human XX maleness results from an abnormal chromosomal X-Y interchange, we studied the inheritance of the paternal pseudoautosomal region in nine patients. Those six patients in whom Y-specific DNA was found (Y(+)) inherited the entire pseudoautosomal region from the paternal Y chromosome and lost that of the paternal X chromosome. Moreover, in three Y(+) cases, we observed the deletion of a paternal Xp locus tightly linked to the pseudoautosomal region. These results definitively show that an abnormal and terminal X-Y interchange during paternal meiosis causes Y(+)XX maleness. In contrast, no abnormal X-Y interchange was observed in any of the three Y(-) cases analyzed, suggesting that maleness can occur in the absence of any Y-specific DNA. © 1987.