

UC Irvine

UC Irvine Previously Published Works

Title

FUNCTIONAL COMPLEMENTATION OF ATAXIA TELANGIECTASIA GROUP-D CELLS BY MICROCELL-MEDIATED CHROMOSOME TRANSFER OF A SINGLE REARRANGED HUMAN-CHROMOSOME AND LOCALIZATION OF THE AT-D GENE TO THE REGION 11Q22.3-23.1

Permalink

<https://escholarship.org/uc/item/6js5277z>

Journal

AMERICAN JOURNAL OF HUMAN GENETICS, 49(4)

ISSN

0002-9297

Authors

HENNING, KA
LAMBERT, C
SCHULTZ, RA
[et al.](#)

Publication Date

1991-10-01

Copyright Information

This work is made available under the terms of a Creative Commons Attribution License, available at <https://creativecommons.org/licenses/by/4.0/>

Peer reviewed

Functional complementation of ataxia telangiectasia group D cells by microcell-mediated chromosome transfer of a single rearranged human chromosome and localization of the AT-D gene to the region 11q22.3-23.1. K.A. Henning* (1), C. Lambert (2), R.A. Schultz (3), M. Smith (4), L.D. McDaniel (3), T. Donlon (2), E.J. Stanbridge (4), and E.C. Friedberg (1). (1) The University of Texas Southwestern Medical Center at Dallas, Dallas, Texas, USA, (2) Stanford University, Stanford, California, USA, (3) The University of Maryland, Baltimore, Maryland, USA, and (4) The University of California, Irvine, California, USA.

Using the technique of microcell-mediated chromosome transfer we have identified a single neo-tagged human chromosome which corrects the phenotypes of increased gamma-ray sensitivity, and sensitivity to streptonigrin and bleomycin in AT-D cells. Additionally, AT-D cells carrying this chromosome are corrected for radio-resistant DNA synthesis and G₂/M blockage after exposure to ionizing radiation. The complementing chromosome is a rearranged human chromosome 18 carrying translocated material from the long arm of chromosome 11. A normal chromosome 18 does not complement AT-D cells, however transfer of human chromosome 11 or 11q does. DNA hybridization using single copy chromosome 11-specific probes reveals the presence of material from the region 11q22.3-23.1 in the rearranged chromosome. Both the AT-A and the AT-C genes have been mapped close to this region, suggesting that there may be only a single wild-type AT gene or multiple closely-linked genes. Isolation and identification of this rearranged human chromosome containing a limited region of chromosome 11 offers a unique opportunity to define the genetic complexity of AT and to facilitate cloning of the AT-D gene by functional complementation.