Title

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Journal

Pediatric Research, 41(Suppl 4)

ISSN

0031-3998

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Publication Date

1997-04-01

DOI

10.1203/00006450-199704001-00631

Published: 01 April 1997

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Pediatric Research volume 41, page104 (1997)

A 15 year old Chilean male, the product of a consanguineous uncle-niece marriage, has type IA GH deficiency and xeroderma pigmentosum (XP), both rare autosomal recessive disorders. Type IA growth hormone (GH) deficiency which is due to a 6.7 or 7.6 kb GH gene deletion is associated with production of antibodies in response to growth hormone therapy. XP causes freckling, xerosis and a variety of skin cancers. The underlying defect is abnormal DNA repair. Eight complementation groups A through G exist for XP. The chromosomal location of most of these genes has been identified (except for the `variant' type). None of these genes are known to map to the the GH locus on chromosome 17q22-q24. The XP in our patient is associated with skin freckling, actinic keratosis and basal cell carcinoma of the face. Fibroblast DNA repair studies following exposure to mitomycin C and ultraviolet C are compatible with`variant' XP. Molecular studies of the growth hormone gene locus show a novel deletion extending 5′ from the GH gene cluster and including the GH gene. GH deficiency and XP appear to be co-segregating in this family. One male sibling with GH deficiency died at the age of 4 years. He was not old enough to have developed XP and none of the remaining 6 brothers have GH deficiency or XP. We postulate that the unique gene deletion causing GH deficiency in this family, is also the location of a previously unmapped`variant' XP gene.

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Kimonis, V., Leschek, E., Schultz, R. et al. A Novel Gene Deletion Associated With Xeroderma Pigmentosum And Type Ia Growth Hormone Deficiency. 611. Pediatr Res 41, 104 (1997). https://doi.org/10.1203/00006450-199704001-00631

DOI
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