Researchers Discover a 14th Gene Linked to FA

Mutations in the gene may cause breast, ovarian cancer susceptibility

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Nature Genetics 42, 406 - 409 (2010) Published online: 18 April 2010 | doi:10.1038/ng.570

Mutation of the RAD51C gene in a Fanconi anemia–like disorder

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Abstract

Fanconi anemia (FA) is a rare chromosomal-instability disorder associated with a variety of developmental abnormalities, bone marrow failure and predisposition to leukemia and other cancers1. We have identified a homozygous missense mutation in the RAD51C gene in a consanguineous family with multiple severe congenital abnormalities characteristic of FA. RAD51C is a member of the RAD51-like gene family involved in homologous recombination–mediated DNA repair. The mutation results in loss of RAD51 focus formation in response to DNA damage and in increased cellular sensitivity to the DNA interstrand cross-linking agent mitomycin C and the topoisomerase-1 inhibitor camptothecin. Thus, biallelic germline mutations in a RAD51 paralog are associated with an FA-like syndrome.