Gene Section
Mini Review

FANCA (Fanconi anaemia A)
Hans Joenje

Department of Clinical Genetics and Human Genetics VU University Medical Center Van der Boechorststraat 7, NL-1081 BT Amsterdam, The Netherlands (HJ)

Published in Atlas Database: December 2001
Online updated version: http://AtlasGeneticsOncology.org/Genes/FA1ID102.html
DOI: 10.4267/2042/37829
This article is an update of: Huret JL. FA1 (Fanconi anaemia 1). Atlas Genet Cytogenet Oncol Haematol. 1998;2(3):81-82.
This work is licensed under a Creative Commons Attribution-Noncommercial-No Derivative Works 2.0 France Licence.

© 2002 Atlas of Genetics and Cytogenetics in Oncology and Haematology

Identity
Other names: FACA; FAA; FA1
HGNC (Hugo): FANCA
Location: 16q24.3

DNA/RNA
Description
43 exons spanning 80 kb.
Transcription
5.5 kb mRNA.

Protein
Description
1455 amino acids; 163 kDa; 2 nuclear localisation signals (NLS) consensus sequences in N-terminus and a potential leucine zipper near C-term, none proven to functional as such.
Expression
Wide: brain, placenta, testis, tonsils (mRNA); in mice: protein expression predominant in lymphoid organs, testis, ovary.
Localisation
Mainly nuclear.
Function
Binds to the protein encoded by FANCC (Fanconi anaemia complementation group C), as well as some of the other FA proteins (FANCE, FANCF, FANCG).

Homology
No known homology or functional motifs.

Mutations

Germinal
Various nucleotide substitutions, deletions, or insertions have been described. Over 90% of the mutations are private, with about 30% being relatively large deletions. Founder mutations have been described in South Africa.

Implicated in
Fanconi anaemia

Note
FANCA is implicated in the FA complementation group A.
Disease
Fanconi anaemia is a chromosome instability syndrome/cancer prone disease (at risk of leukaemia and squamous cell carcinoma).
Prognosis
Poor; mean survival is 20 years; patients die of bone marrow failure (infections, haemorrhages), leukaemia, or solid cancer.
Cytogenetics
Spontaneously enhanced chromatid-type aberrations (breaks, gaps, interchanges; increased rate of breaks compared to control, when induced by specific clastogens known as DNA cross-linking agents (e.g. mitomycin C, diepoxybutane).


Garcia-Higuera I, Kuang Y, Denham J, D’Andrea AD. The Fanconi anemia proteins FAA and FAC stabilize each other and promote the nuclear accumulation of the Fanconi anemia complex. Blood. 2000 Nov 1;96(9):3224-30


Wong JC, Alon N, Norga K, Kruyt FA, Yuussoufian H, Buchwald M. Cloning and analysis of the mouse Fanconi anemia group A cDNA and an overlapping pentazinc finger cDNA. Genomics. 2000 Aug 1;67(3):273-83


McMahon LW, Sangemann J, Goodman SR, Kumaresh K, Lambert MW. Human alpha spectrin II and the Fanca, FANCC, and FANCG proteins bind to DNA containing psoralen...


This article should be referenced as such: