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Gene Section
Mini Review

FANCA (Fanconi anaemia A)

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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).

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