

FANCA (Fanconi anaemia complementation group

A) (updated: old version not available)

Identity

Other names **FACA**

FAA

FA1

Hugo [FANCA](#)

Location 16q24.3

DNA/RNA

Description 43 exons spanning 80 kb; 4365 bp open reading frame

Transcription 5.5 kb mRNA

Protein

Description 1455 amino acids; 163 kDa; 2 nuclear localisation signals (NLS) consensus sequences in N-terminus and a leucine zipper in 1069-1090, none proven to functional as such; FANCA is normally phosphorylated

Expression wide: brain, placenta, testis, tonsils (mRNA); in mice: protein expression predominant in lymphoid organs, testis, ovary.

Localisation both cytoplasmic and nuclear

Function part of the FA complex with [FANCC](#), [FANCE](#), [FANCF](#), and [FANCG](#); this complex is only found in the nucleus.

FANCA and FANCG form a complex in the cytoplasm, through a N-term FANCA (involving the nuclear localization signal) - FANCG interaction; FANCC join the complex; phosphorylation of FANCA would induce its translocation into the nucleus. This FA complex translocates into the nucleus, where FANCE and FANCF are present; FANCE and FANCF join the complex. The FA complex subsequently interacts with [FANCD2](#) by monoubiquitination of FANCD2 during S phase or following DNA damage. Activated (ubiquitinated) FANCD2, downstream in the FA pathway, will then interact with other proteins involved in DNA repair, possibly BRCA1; after DNA repair, FANCD2 return to the non-ubiquitinated form.

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

Entity	Fanconi anaemia (FA); FANCA is implicated in the FA complementation group A; it represents about 70% of FA cases
Disease	Fanconi anaemia is a chromosome instability syndrome/cancer prone disease (at risk of leukaemia and squamous cell carcinoma)
Prognosis	<p>Fanconi anaemia's prognosis is poor; mean survival is 20 years: patients die of bone marrow failure (infections, haemorrhages), leukaemia, or solid cancer.</p> <p>It has recently been shown that significant phenotypic differences were found between the various complementation groups. In FA group A, patients homozygous for null mutations had an earlier onset of anemia and a higher incidence of leukemia than those with mutations producing an altered protein. Patients homozygous for null mutations in FANCA are high-risk groups with a poor hematologic outcome and should be considered as candidates both for frequent monitoring and early therapeutic intervention.</p>
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).

External links

	Nomenclature
Hugo	FANCA
GDB	FANCA
Entrez Gene	FANCA 2175 Fanconi anemia, complementation group A
	Cards
Atlas	FA1ID102
GeneCards	FANCA
Ensembl	FANCA
CancerGene	FANCA
Genatlas	FANCA
GeneLynx	FANCA
eGenome	FANCA
euGene	2175
	Genomic and cartography
GoldenPath	FANCA - 16q24.3 chr16:88331460-88410555 - 16q24.3 (hg17-May_2004)
Ensembl	FANCA - 16q24.3 [CytoView]

[NCBI](#) [Genes Cyto](#) [Gene Seq](#) [Map View - NCBI]

[OMIM](#) [Disease map \[OMIM\]](#)

[HomoloGene](#) [FANCA](#)

Gene and transcription

[Genbank](#) [AC005360](#) [SRS] [AC005360](#) [ENTREZ]

[Genbank](#) [AC005565](#) [SRS] [AC005565](#) [ENTREZ]

[Genbank](#) [AC005567](#) [SRS] [AC005567](#) [ENTREZ]

[Genbank](#) [AF054569](#) [SRS] [AF054569](#) [ENTREZ]

[Genbank](#) [AJ131189](#) [SRS] [AJ131189](#) [ENTREZ]

[RefSeq](#) [NM_000135](#) [SRS] [NM_000135](#) [ENTREZ]

[RefSeq](#) [NT_086856](#) [SRS] [NT_086856](#) [ENTREZ]

[AceView](#) [FANCA](#) AceView - NCBI

[TRASER](#) [FANCA](#) Traser - Stanford

[Unigene](#) [Hs.284153](#) [SRS] [Hs.284153](#) [NCBI] [HS284153](#) [spliceNest]

Protein : pattern, domain, 3D structure

[SwissProt](#) [O15360](#) [SRS] [O15360](#) [EXPASY] [O15360](#) [INTERPRO]

[CluSTr](#) [O15360](#)

[Blocks](#) [O15360](#)

Polymorphism : SNP, mutations, diseases

[OMIM](#) [607139](#) [map]

[GENECLINICS](#) [607139](#)

[SNP](#) [FANCA](#) [dbSNP-NCBI]

[SNP](#) [NM_000135](#) [SNP-NCI]

[SNP](#) [FANCA](#) [GeneSNPs - Utah] [FANCA](#) [SNP - CSHL] [FANCA](#) [HGBASE - SRS]

General knowledge

[Family Browser](#) [FANCA](#) [UCSC Family Browser]

[SOURCE](#) [NM_000135](#)

[SMD](#) [Hs.284153](#)

[SAGE](#) [Hs.284153](#)

[Amigo](#) [process|DNA repair](#)

[Amigo](#) [component|cytoplasm](#)

[Amigo](#) [component|nucleus](#)

[Amigo](#) [process|protein complex assembly](#)

[BIOCARTA](#) [Role of BRCA1, BRCA2 and ATR in Cancer Susceptibility](#)

[BIOCARTA](#) [BRCA1-dependent Ub-ligase activity](#)

[PubGene](#) [FANCA](#)

Other databases

Other database [Fanconi Anemia Mutation Database](#)

Probes

[Probe](#) [FANCA Related clones \(RZPD - Berlin\)](#)

PubMed

[PubMed](#) [17 Pubmed reference\(s\) in LocusLink](#)

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Medline [97051928](#)

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