FANCF (Fanconi anemia, complementation group F)

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Identity

Other names: FAF
HGNC (Hugo): FANCF
Location: 11p15

DNA/RNA

Description
1 exon; 1124 bp open reading frame.

Protein

Description
374 amino acids; 42 kDa.

Expression
Weak.

Localisation
Predominantly nuclear.

Function
Part of the FA complex with FANCA, FANCC, FANCE, 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 (ubiquinated) 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-ubiquinated form.

Homology

ROM (prokaryote).

Implicated in

Fanconi anaemia (FA)

FANCF is implicated in the FA complementation group F; it represents about 2-3% of FA cases.

Disease

Fanconi anaemia is a chromosome instability syndrome/cancer prone disease (at risk of leukaemia and squamous cell carcinoma).

Prognosis

Fanconi anaemia's prognosis is poor; mean survival is 20 years: patients die of bone marrow failure (infections, haemorrhages), leukaemia, or solid cancer. It has recently been shown that significant phenotypic differences were found between the various complementation groups. Patients from the rare groups...
FA-D, FA-E, and FA-F had somatic abnormalities more frequently.

**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).

**References**


This article should be referenced as such: