Atlas of Genetics and Cytogenetics in Oncology and Haematology

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Gene Section
Mini Review

FANCE (Fanconi anemia, complementation group E)

Jean-Loup Huret

Genetics, Dept Medical Information, UMR 8125 CNRS, University of Poitiers, CHU Poitiers Hospital, F-86021 Poitiers, France (JLH)

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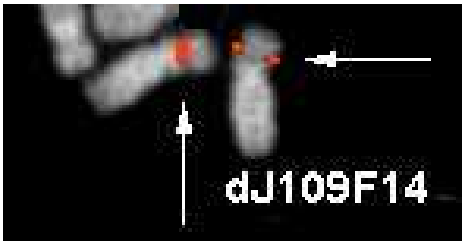
Identity

Other names: FACE; FAE

HGNC (Hugo): FANCE

Location: 6p21

Local order: located between the 60S ribosomal protein RPL10Aand a ZNF127 like protein.



Probe(s) - Courtesy Mariano Rocchi, Resources for Molecular Cytogenetics.

DNA/RNA

Description

The gene spans 15 kb and contains 10 exons; 1611 bp open reading frame.

Protein

Description

536 amino acids, 60 kDa; contains two potential nuclear localization signals.

Function

Part of the FA complex with FANCA, FANCC, 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 (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

No known homology.

Implicated in

Fanconi anaemia (FA)

FANCE is implicated in the FA complementation group E; it represents about 2% of FA cases.

Disease

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

Prognosis

Fanconi anaemia's prognosis is poor; mean survival is 20 years (depending on mutation, treatment): patients die of bone marrow failure (infections, haemorrhages), leukaemia, or androgen therapy related liver tumours.
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

Spontaneous, chromatid/chromosome breaks; increased rate of breaks compared to control, when induced by breaking agent.

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