FACC (Fanconi anaemia complementation group C)
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Identity

Other names: FAC
Location: in 9q22.3
Local order: next to PTCH and XPAC.

DNA/RNA

Description
14 exons; spans 80 kb.

Transcription
mRNA of 2.3, 3.2, and 4.6 kb (variable 3’ untranslated region, alternative splicing, exon skipping).

Protein

Description
558 amino acids; 63 kDa; alpha helical structure in C-term.

Expression
Wide, in particular in the bones.

Localisation
Cytoplasmic at any cell-cycle stage.

Function
Peak expression during the G2/M transition; binds to cdc2 (mitotic cyclin-dependent kinase); probably involved in basic aspect(s) of the cell protection against DNA damages: role in the cell cycle regulation and/or in DNA repair and/or in the prevention of cellular apoptosis; binds to FAA, the protein encoded by FA1 (Fanconi anaemia complementation group A), the dimer being found in the cytoplasm and the nucleus.

Homology
No known homology.

Mutations

Germinal
Mainly nucleotide substitutions, dispersed along the coding sequence.

Implicated in

Fanconi anaemia; FACC is implicated in the FA complementation group C

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

Prognosis
Poor; mean survival is 16 years: patients die of bone marrow failure (infections, haemorrhages), leukaemia, or androgen therapy related liver tumours.

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

References


This article should be referenced as such: