FA1 (Fanconi anaemia 1)

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Identity

Other names: FACA (Fanconi anaemia complementation group A); FAA
Location: 16q24.3

DNA/RNA

Description
43 exons at least; spans on 80 kb.

Transcription
5.5 mRNA.

Protein

Description
1455 amino acids; 163 kDa; 2 nuclear localisation signals in N-term and a potential leucine zipper near the C-term.

Expression
Wide: brain, placenta, testis, tonsils.

Localisation
Was thought to be nuclear; mostly cytoplasmic.

Function
Binds to the protein encoded by FACC (Fanconi anaemia complementation group C), the dimer being found in the cytoplasm and the nucleus.

Homology
No known homology.

Mutations

Germinial
Various nucleotide substitutions, deletions, or insertions have been described.

Implicated in

Fanconi anaemia; FA1 is implicated in the FA complementation group A

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: