

Gene Section

Short Communication

CFAP70 (cilia and flagella associated protein 70)

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Abstract

Review on CFAP70, a poorly known gene and protein, with data on DNA, on the protein encoded, and where the gene is implicated.

Keywords

CFAP70; Tetratricopeptide repeats; Cancer; Fusion gene; Axoneme; Cilia; Dynein

Identity

Other names: TTC18

HGNC (Hugo): CFAP70

Location : 10q22.1; on reverse strand

DNA/RNA

Transcription

Various transcripts (see below).

Protein

Description

Various isoforms have been described:

According to Vega: 3 protein coding transcripts: CFAP70-001: 3691 base pairs (bp), coding for a protein of 590 amino acids (aa); CFAP70-006: 1710bp, 498 aa; CFAP70-002: 1403bp, 361 aa
According to Uniprot: Isoform 1 (Q5T0N1-1), canonical sequence: 1,121 aa, 125,721 Da; Isoform 2 (Q5T0N1-2): 1,091 aa, 122,269 Da; Isoform 3

(Q5T0N1-3): 119 aa, 12,999 Da; Isoform 4 (Q5T0N1-6): 195 aa, 22,338 Da; Isoform 5 (Q5T0N1-7): 225 aa, 25,790 Da.

According to Ensembl: Protein ENSP00000310829: the transcript has 28 exons, 3703bp, 1121 aa, 33 domains; Protein ENSP00000347781: the transcript has 28 exons, 3691bp, 590 aa, 32 domains; Protein ENSP00000409527; the transcript has 12 exons, 1710bp, 498 aa, 23 domains; Protein ENSP00000343650: the transcript has 12 exons, 1403bp, 361 aa, 13 domains; Protein ENSP00000378334; the transcript has 9 exons, 1222bp, 179 aa, 13 domains.

Domains in the canonical sequence (Q5T0N1-1 in UniProt): 1121 aa.

There are 8 tetratricopeptide repeats (TRP): TPR 1, aa 635-668, TPR 2: aa 669-702, TPR 3 : aa 704-736, TPR 4: aa 929-962, TPR 5: aa 963-996, TPR 6: aa 1000-1033, TPR 7: aa 1035-1066 and TPR 8: aa 1068-1100.

The TPR sequence contains about 34 amino acids, with a consensus sequence of small and large hydrophobic amino acids; small hydrophobic amino acids are often found at positions 8, 20, and 27, and large ones at positions 4, 7, and 24. Three-dimensional structure shows that the TPR motif contains two antiparallel α -helical subdomains (termed helix A and helix B), equivalent in length. TPR adopts a helix-turn-helix arrangement. They self-associate via a "knobs and holes" mechanism (Das et al., 1998; D'Andrea and Regan 2003; Cerveny et al., 2013; SMART).

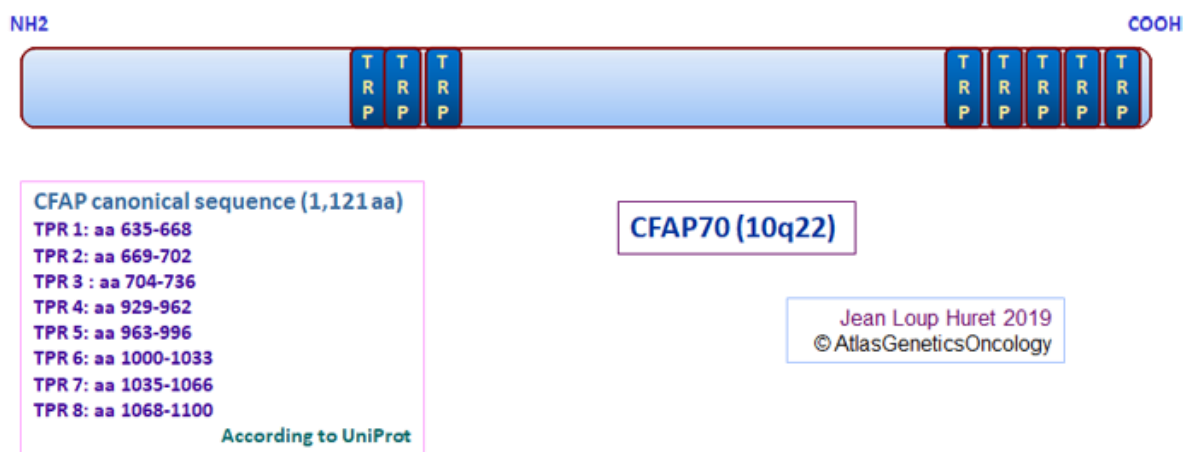


Figure 1 CFAP70 protein and domains.

Expression

CFAP70 is expressed in adrenal gland, bronchus, testis, sperm flagella and in cilia of glandular cells in fallopian tube, and endometrium, according to The Human Protein Atlas.

Localisation

CFAP70 localizes on the epithelial cilia and sperm flagella. It associates tightly to the ciliary axoneme (Shamoto et al., 2018).

Function

Tetratricopeptide repeats are found in at least 149 proteins (PFAM). Tetratricopeptide repeat containing proteins are involved in protein-protein interactions and the assembly of multiprotein complexes, cell cycle, transcription, mitochondrial protein transport, and neurogenesis.

CFAP70 binds to the axoneme of motile cilia and flagella in close proximity to the outer dynein arms and regulates outer dynein arms activity. It is the N-terminus of CFAP70 which localizes at the base of the outer dynein arms and not the tetratricopeptide repeats. Knockdown of CFAP70 causes a reduction in ciliary motility (Shamoto et al., 2018).

Implicated in

Top note

Low or high expression of CFAP70 is not a prognostic indicator in any cancer tested (The Human Protein Atlas).

Breast adenocarcinoma

The fusion gene DNAJC9/CFAP70 (10q22-10q22) has been found in breast adenocarcinoma (Gao et al. 2018).

The fusion gene CFAP70/ MCU (10q22-10q22) has been found in a breast cancer cell line (Klijin et al. 2015).

Uterus corpus adenocarcinoma

The fusion gene P4HA1/CFAP70 (10q22-10q22) has been found in uterus corpus adenocarcinoma (Hu et al. 2018).

Lung squamous cell carcinoma

The fusion gene ECD/CFAP70 (10q22-10q22) has been found in lung squamous cell carcinoma (Yoshihara et al. 2015).

Thyroid adenocarcinoma

The fusion gene MRPS16/CFAP70 (10q22-10q22) has been found in thyroid adenocarcinoma (Yoshihara et al. 2015).

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