

Gene Section

Short Communication

COX6C (cytochrome c oxidase subunit VIc)

Jean-Loup Huret, Sylvie Senon

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

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Identity

Other names: Cytochrome c oxidase polypeptide VIc

precursor

HGNC (Hugo): COX6C **Location:** 8q22.2

Location (base pair): 100,846,955

DNA/RNA

Description

The gene spans 15.5 kb; orientation: reverse strand; 4 exons; this cytochrome C oxidase subunit 6c is encoded by the nuclear DNA (some other subunits are encoded by the mitochondrial DNA).

Protein

Description

75 amino acids, 8.8 kDa; contains a transmembrane domain.

Expression

Wide.

Localisation

Mitochondria (inner membrane).

Function

Subunit of the cytochrome C oxidase (respiratory chain complex of the mitochondria); catalytic activity.

Implicated in

Uterine fibromyoma

Disease

The most common benign tumors of the female genital tract.

Hybrid/Mutated gene

HMGA2 HMGA2 exon 3 was found fused to COX6C exon 2 in one case without cytogenetic analysis; HMGA2 (previously named HMGIC), sitting in 12q15, is the most frequently involved gene in uterine leiomyoma.

Abnormal protein

The 3 N-term DNA binding domains (AT hooks) of HMGA2 is fused to the C-term of COX6C

References

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