Gene Section

Mini Review

CLDN4 (claudin-4)
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Identity
Hugo: CLDN4
Other names: CPETR; CPETR1; WBSCR8 (Williams-Beuren syndrome chromosome region 8 protein); hCPE-R, CPE-R (Clostridium perfringens enterotoxin receptor)
Location: 7q11.23

DNA/RNA
Description
Intronless; one exon spanning 1.68 kb.

Transcription
One transcript of 1.68 kb with 630 bp of coding sequence.

Protein
Description
The CLDN4 protein contains 209 amino acids and has a molecular weight of 22.1 kDa with four putative transmembrane segments. It directly interacts with TJP1/ZO-1, TJP2/ZO-2 and TJP3/ZO-3.

Expression
Claudin-4 is expressed in many fetal and adult tissues, predominantly in lung, intestine and kidney. Overexpressed in pancreatic, breast, ovarian, and prostate cancer.

Localisation
Integral membrane protein. Tight junction component.

Function
CLDN4 plays a major role in tight junction-specific obliteration of the intercellular space.

Homology
Belongs to the claudin family.

Implicated in
Williams-Beuren syndrome
Disease
Williams-Beuren syndrome (WBS) includes supravalvular aortic stenosis (SVAS), multiple peripheral pulmonary arterial stenoses, elfin face, mental and statural deficiency, characteristic dental malformation, and infantile hypercalcemia. It is associated with an autosomal dominant contiguous gene deletion involving genes from chromosome band 7q11.23, including CLDN4, elastin and LIM-kinase1. Haploinsufficiency for CLDN4 may be the cause of certain cardiovascular and musculo-skeletal abnormalities observed in the context of this disease.

Gastric cancer
Oncogenesis
Downregulated in gastric cancer. Absence of CLDN4 may play a role in the disruption of cell-to-cell adhesion in diffuse type gastric cancer and in a loss of differentiation.

Pancreatic cancer
Oncogenesis
Overexpressed in pancreatic cancer. Overexpression is predominantly observed in well-differentiated tumors with decreased metastatic potencial.
Breast cancer
Oncogenesis

Ovarian cancer
Oncogenesis
CLDN4 is upregulated in ovarian tumors and cell lines and may represent a novel marker for this disease.

Squamous cell carcinoma and Bowen's disease
Oncogenesis
Expression of claudin-4 is associated with keratinization in SCC and BD.

Prostate cancer
Oncogenesis
Overexpressed in prostate cancer epithelium. Significance unclear.

References


This article should be referenced as such: