Atlas of Genetics and Cytogenetics in Oncology and Haematology

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# Gene Section
## Mini Review

# CLDN4 (claudin-4)

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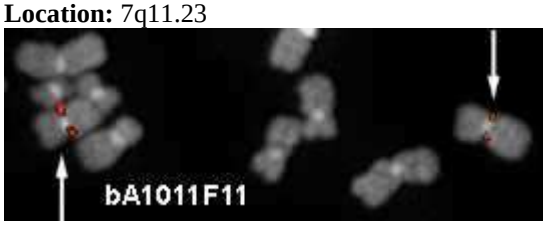
## Identity

**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




Probe(s) - Courtesy Mariano Rocchi, Resources for Molecular Cytogenetics.

## 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 syndrom

**Disease**

Williams-Beuren syndrom (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**

Overexpressed in breast cancer and Paget's disease. Significance unclear.

### 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 ephitelium. Significance unclear.

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