Identity

Other names: EMSY; GL002; ENT0
HGNC (Hugo): C11ORF30
Location: 11q13.5
Local order: CCND1 is also at 11q13.

DNA/RNA

Description
The gene spans 107 kb; on direct strand.

Transcription
At least 11 transcripts; one transcript of 4kb and 21 exons encodes a protein of 1323 amino acids and 141 kDa.

Protein

Localisation
Nucleus.

Function
Binds BRCA2 at its N-term transactivation domain; may repress the transcriptional activity of BRCA2.

Implicated in

C11ORF30/EMSY was found amplified in 7 and 17 % of cases respectively of sporadic breast cancers and high grade ovarian cancers, but very rarely in other cancers tested

Note
11q13 is frequently amplified in sporadic breast cancers; CCND1, in particular, is amplified in about 20% of cases. However, C11ORF30/EMSY was found amplified in cases with normal CCND1: they are independant factors.

Prognosis
C11ORF30/EMSY amplification is associated with a worse prognosis in node-negative breast cancers, and has no prgnosis implication in node-positive breast cancers.

References


This article should be referenced as such: