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Solid Tumour Section

Short Communication

Breast: carcinoma with t(1;3)(p36;q13) PRDM16/BBX

Jean-Loup Huret

Genetics, Dept Medical Information, University of Poitiers, CHU Poitiers Hospital, F-86021 Poitiers, France. jean-loup.huret@chu-poitiers.fr

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Abstract

Review on t(1;3)(p36;q13) PRDM16/BBX in breast cancer, with data on the genes involved.

Keywords

chromosome 1; chromosome 3; t(1;3)(p36;q13); PRDM16; BBX; breast carcinoma

Clinics and pathology

Only one case to date, without further data; it is described as a breast adenocarcinoma, without location/morphology details (ductal carcinoma? lobular carcinoma?) (Yoshihara et al 2015).

Epidemiology

Invasive breast carcinoma is the most common cancer in women, accounting for 22% of all female cancers. One million women worldwide are diagnosed with breast cancer every year (Carcangiu et al. 2005). The most common histologic type of invasive breast carcinoma is designated as ductal carcinoma (80% of all cases), with many morphologic variants. Invasive lobular carcinoma is the second major type (5-10%) of breast cancer (Carcangiu et al. 2005).

Genes involved and proteins

PRDM16 (PR domain containing 16)

Location

1p36.32

DNA / RNA

11 splice variants

Protein

1276 amino acids and smaller proteins. Contains a N-term PR domain; 7 Zinc fingers, a proline-rich domain, and 3 Zinc fingers in the C-term. Binds DNA. Transcription activator; PRDM16 has an intrinsic histone methyltransferase activity. PRDM16 forms a transcriptional complex with CEBPB. PRDM16 plays a downstream regulatory role in mediating TGFB signaling (Bjork et al., 2010). PRDM16 induces brown fat determination and differentiation. PRDM16 is expressed selectively in the earliest stem and progenitor hematopoietic cells, and is required for the maintenance of the hematopoietic stem cell pool during development. PRDM16 is also required for survival, cell-cycle regulation and self-renewal in neural stem cells (Chuikov et al., 2010; Kajimura et al., 2010; Aguilo et al., 2011; Chi and Cohen, 2016).

BBX (BBX, HMG-box containing)

Location

3q13.12

Protein

941 amino acids; high mobility group (HMG)-box protein. These proteins possess a DNA-binding domain. They are involved in transcription, replication and DNA repair. BBX belongs to a novel subfamily of HMG-Box superfamily together with CIC (Chen et al., 2014). BBX proteins control growth and developmental processes of plants, including seedling photomorphogenesis, and

photoperiodic regulation of flowering (Gangappa and Botto, 2014), but their function in animals is poorly known. Bbx is highly expressed in the cortex and hippocampus, and may be associated with central nervous system in zebrafish embryos (Chen et al., 2014). Homozygous Bbx mutants rats exhibit intrauterine growth retardation and male infertility, suggesting that loss of BBX induces apoptosis and results in spermiogenesis defects (Wang et al., 2016).

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