PDGFB

Platelet-derived growth factor subunit B is a protein that in humans is encoded by the PDGFB gene.

Function

The protein encoded by this gene is a member of the platelet-derived growth factor family. The four members of this family are mitogenic factors for cells of mesenchymal origin and are characterized by a motif of eight cysteines. This gene product can exist either as a homodimer (PDGF-BB) or as a heterodimer with the platelet-derived growth factor alpha (PDGFA) polypeptide (PDGF-AB), where the dimers are connected by disulfide bonds.

Clinical significance

Mutations in this gene are associated with meningioma. Reciprocal translocations between chromosomes 22 and 17, at sites where this gene and that for COL1A1 are located, are associated with a particular type of skin tumor called dermatofibrosarcoma protuberans resulting from unregulated expression of growth factor. Two splice variants have been identified for this gene.

Pericytes are perivascular cells along capillaries that are critical for the development of a functional vascular bed in the central nervous system and other organs. Pericyte functions in the adult brain are less well understood. Pericytes have been suggested to mediate functional hyperemia at the capillary level, regulate the blood-brain barrier and to give rise to scar tissue after spinal cord injury. Furthermore, pericyte loss has been suggested to precede cognitive decline in mouse models of Alzheimer's disease. Despite this observation, there is no convincing causality between pericyte loss and the pathogenesis of Alzheimer's disease. However, recent loss-of-function mutations in PDGFB and PDGFRB genes have implicated pericytes as the principle cell type affected in primary familiar brain calcification (PFBC), a neuropsychiatric disorder with dominant inheritance.

Zarb et al., reviewed the role of the PDGFB/PDGFRB signaling pathway in pericyte development and briefly discussed homeostatic functions of pericytes in the brain. They provided an overview of recent studies with mouse models of PFBC and discuss suggested pathogenic mechanisms for PFBC with special reference to pericytes [1].

1)
Zarb Y, Franzoso FD, Keller A. Pericytes in Primary Familial Brain Calcification. Adv Exp Med Biol. 2019;1147:247-264. doi: 10.1007/978-3-030-16908-4_11. PubMed PMID: 31147881.