Platelet derived growth factor receptor A

PDGFRA, i.e. platelet derived growth factor receptor A, also termed PDGFRα, i.e. platelet-derived growth factor receptor α, is a cell surface receptor located on the surface of a wide range of cell types. This receptor binds to certain isoforms of platelet-derived growth factors (PDGFs) and thereby becomes active in stimulating cell signaling pathways that elicit responses such as cellular growth and differentiation. The receptor is critical for the development of certain tissues and organs during embryogenesis and for the maintenance of these tissues and organs, particularly hematologic tissues, throughout life. Mutations in the gene which codes for PDGFRA, i.e. the PDGFRA gene, are associated with an array of clinically significant neoplasms.

This gene encodes a cell surface tyrosine kinase receptor for members of the platelet-derived growth factor family. These growth factors are mitogens for cells of mesenchymal origin. The identity of the growth factor bound to a receptor monomer determines whether the functional receptor is a homodimer or a heterodimer, composed of both platelet-derived growth factor receptor alpha and beta polypeptides. Studies in knockout mice, where homozygosity is lethal, indicate that the alpha form of the platelet-derived growth factor receptor is particularly important for kidney development since mice heterozygous for the receptor exhibit defective kidney phenotypes.

Solomon et al., from the Department of Pathology, University of California, San Francisco, USA. identifed a recurrent PDGFRA p.K385 hotspot mutation as the solitary pathogenic alteration in DNT-like and RGNT-like low-grade glioneuronal tumors of the septum pellucidum, which together with their stereotypic anatomic location, should help facilitate accurate diagnosis of this distinct neuroepithelial tumor entity for which they propose the name “myxoid glioneuronal tumor of the septum pellucidum and lateral ventricle, PDGFRA-mutant” [1].

1)
Solomon DA, Korshunov A, Sill M, Jones DTW, Kool M, Pfister SM, Fan X, Bannykh S, Hu J, Danielpour M, Li R, Johnston J, Cham E, Cooney T, Sun PP, Oberheim Bush NA, McDermott M, Van Ziffle J, Onodera C, Grenert JP, Bastian BC, Villanueva-Meyer JE, Pekmezci M, Bollen AW, Perry A. Myxoid glioneuronal tumor of the septum pellucidum and lateral ventricle is defined by a recurrent PDGFRA p.K385 mutation and DNT-like methylation profile. Acta Neuropathol. 2018 Jul 13. doi: 10.1007/s00401-018-1883-2. [Epub ahead of print] PubMed PMID: 30006677.