

CCDC88C (coiled-coil domain containing 88C) is a gene that encodes a protein involved in various cellular processes, including cell signaling and organization. The protein contains coiled-coil domains, which facilitate protein-protein interactions and are critical for cellular structure and function.

Role in Congenital Hydrocephalus Mutations in the CCDC88C gene have been linked to congenital hydrocephalus, a condition characterized by abnormal accumulation of cerebrospinal fluid (CSF) in the brain's ventricles. The association between CCDC88C mutations and congenital hydrocephalus highlights several key aspects:

Cellular Function: CCDC88C is involved in cellular processes that are crucial for normal brain development. It plays a role in neurogenesis and the organization of cell structures, which are essential for the proper functioning of the central nervous system.

Developmental Impact: Disruptions in the CCDC88C gene can affect the development of the brain's ventricular system and the ependymal cells that line the ventricles. Proper function of these cells is necessary for the circulation and absorption of CSF. Mutations may impair these functions, leading to the accumulation of CSF and resultant hydrocephalus.

Genetic and Phenotypic Correlations: Mutations in CCDC88C can contribute to a range of developmental abnormalities, including hydrocephalus. The precise mechanisms by which these mutations lead to hydrocephalus are still under investigation but are believed to involve disruptions in cellular signaling pathways and structural organization within the brain.

Summary CCDC88C is an important gene in the pathogenesis of congenital hydrocephalus, with its mutations affecting cellular processes critical for brain development and CSF dynamics. Research into this gene helps to further our understanding of the molecular mechanisms underlying hydrocephalus and may offer potential targets for therapeutic interventions.

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