KMT2C, also known as Lysine (K)-specific methyltransferase 2C, is a gene that encodes a protein involved in the regulation of gene expression through histone methylation. Histones are proteins that help package and organize DNA in the cell nucleus. KMT2C is a member of the lysine methyltransferase family and plays a crucial role in epigenetic modifications of histones.

Key points about KMT2C:

Gene Function: KMT2C encodes a protein that acts as a histone methyltransferase. Specifically, it adds methyl groups to lysine residues on histone H3. These modifications influence the accessibility of DNA to the cellular machinery that reads and interprets the genetic code. Thus, KMT2C is involved in the epigenetic regulation of gene expression.

Histone Methylation: Methylation of histones can result in either activation or repression of gene expression, depending on the specific lysine residue and the number of methyl groups added.

Role in Development and Disease: Mutations or dysregulation of KMT2C have been associated with various developmental disorders and diseases. For example, mutations in KMT2C have been implicated in some cases of intellectual disability, autism spectrum disorders, and congenital heart defects.

Cancer: KMT2C mutations have also been identified in certain types of cancer, including colorectal cancer and acute lymphoblastic leukemia (ALL). Dysregulation of KMT2C can lead to aberrant gene expression and contribute to cancer development.

Role in Epigenetics: KMT2C is part of the epigenetic machinery that helps establish and maintain proper patterns of gene expression during development and in response to environmental cues. Epigenetic modifications are reversible and can be influenced by various factors, making them a focus of research in both health and disease.

Multifunctional Protein: The KMT2C protein is multifunctional and participates in a complex network of interactions with other proteins involved in epigenetic regulation. Its role in maintaining normal cellular function is critical.

Research into KMT2C and its functions continues to provide insights into the complex world of gene regulation and epigenetics. Understanding how this gene contributes to both normal development and disease is a focus of ongoing scientific investigation.