# Lysine-Specific Methyltransferase 2C

## Alternative Names
KMT2C
Myeloid/Lymphoid or Mixed-Lineage Leukemia 3
MLL3
KIAA1506

## Record Category
Gene locus

## WHO-ICD
N/A to gene loci

## Incidence per 100,000 Live Births
N/A to gene loci

## OMIM Number
606833

## Mode of Inheritance
N/A to gene loci

## Gene Map Locus
7q36.1

## Description
The KMT2C gene encodes Lysine-Specific Methyltransferase 2C, a nuclear protein belonging to the myeloid/lymphoid or mixed-lineage leukemia (MLL) family. Similar to all other MLL proteins, KMT2C is activated by a minimized RBBP5–ASH2L heterodimer. The protein functions as an enzyme and is responsible for catalyzing the methylation of the Lysine-4 residue of histone H3. This methylation is necessary for epigenetic transcriptional activation. KMT2C may also function as the catalytic subunit of the MLL2/3 coactivator complex of nuclear receptors, which is involved in transcriptional coactivation.

There have been individual reports of KMT2C gene mutations in patients with Kleefstra Syndrome, a disorder characterized by severe mental retardation, obesity, hypotonia, seizures, microcephaly and facial dysmorphia. However, a direct causal link between this condition and KMT2C mutations has not yet been established.

## Molecular Genetics
The KMT2C gene, located on the long arm of chromosome 7, spans a length of 301.7 kb of DNA. Its coding sequence is spread across 65 exons and it encodes a 541.3 kDa protein product comprised of 4911 amino acids. Two additional isoforms of the KMT2C protein exist due to alternatively spliced transcript variants. The gene is widely expressed in the human body with highest expression seen in the testis, ovary, brain and liver. The heterozygous nonsense mutation R1481X has been tentatively linked to Kleefstra syndrome.

## Epidemiology in the Arab World
Saudi Arabia
Monies et al. (2017) studied the findings of 1000 diagnostic panels and exomes carried out at a next generation sequencing lab in Saudi Arabia. One male patient from a consanguineous family presented with speech and mental delay, learning disability, recurrent fever, seizures and Generalized Tonic-Clonic epilepsy. Using whole exome sequencing a heterozygous mutation (c.6589C>A, p.Q2197K) was identified in exon 36 of the patient's KMT2C gene. A mutation in the KMT2C gene had previously been tentatively linked to Kleefstra syndrome, and hence this result was seen to confirm this association.

## References
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## Related CTGA Records

Kleefstra Syndrome (OMIM 610253)

## External Links

http://www.genecards.org/cgi-bin/carddisp.pl?gene=KMT2C

## Contributors

Sayeeda Hana
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