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- DNMT1 is associated with more than 300 diseases (figure A). Its expression is down-regulated in multiple types of carcinoma, such as non-small cell lung carcinoma, lung adenocarcinoma (LUAD) and breast carcinoma (figure B). DNA hypomethylation is a hallmark of these cancers.
- Multiple pathogenic mutations, e.g., Lys521del, Tyr540Asp and Asn545del (figure C), have been identified in the RFD domain (figure D), which is the DNA replication fork binding domain of DNMT1 protein. These mutations have direct impacts on the DNA methylation, and are thought to be related to disease progression.





