ATRX

The ATRX gene codes for ATRX (alpha-thalassemia/mental retardation syndrome, nondeletion type, X-linked) protein which is involved in silencing certain gene sites in humans.

Stating that ATRX is normal typically means that there are no detectable mutations, abnormalities, or disruptions in the ATRX gene. This is a positive finding, as normal ATRX function is important for maintaining genomic stability and proper cellular functioning. Abnormalities in the ATRX gene have been associated with certain genetic disorders and cancers, so a normal ATRX status is generally considered favorable.

Function

Transcriptional regulator ATRX contains an ATPase / helicase domain, and thus it belongs to the SWI/SNF family of chromatin remodeling proteins. This protein is found to undergo cell cycle-dependent phosphorylation, which regulates its nuclear matrix and chromatin association, and suggests its involvement in the gene regulation at interphase and chromosomal segregation in mitosis.

ATRX loss

ATRX loss

ATRX mutation

ATRX gene mutation