ATRX gene mutation

ATRX gene mutations are strongly coupled to IDH & TP53 mutations, and occur in a number of human cancers (in the CNS: in secondary Glioblastoma and its precursors, grade II & grade III gliomas). Uncommon in oligodendrogliomas and secondary Glioblastomas.

Testing for these is performed by polymerase chain reaction (PCR).

In general, ATRX & TP53 mutations are mutually exclusive of 1p/19q co-deletion, and as such may be used as confirmatory markers to distinguish astrocytomas from oligodendrogliomas.

Indications: ATRX is a confirmatory test along with IDH1 mutation. ATRX & TP53 can be detected by immunohistochemical (IHC) stains or by Fluorescence in situ hybridization (FISH) and may be done in some hospitals or may be sent out to specialty labs; results typically take ≈ 2–3 days. The cost of IHC is on the order of $100–150 U.S., FISH is about $200–250 U.S.

Transcriptional regulator ATRX also known as ATP-dependent helicase ATRX, X-linked helicase II, or X-linked nuclear protein (XNP) is a protein that in humans is encoded by the ATRX gene.

Clinical significance

Mutations of the ATRX gene are associated with an X-linked mental retardation (XLMR) syndrome most often accompanied by alpha-thalassemia (ATRX) syndrome. These mutations have been shown to cause diverse changes in the pattern of DNA methylation, which may provide a link between chromatin remodeling, DNA methylation, and gene expression in developmental processes. Multiple alternatively spliced transcript variants encoding distinct isoforms have been reported. Female carriers may demonstrate skewed X chromosome inactivation.

Interactions

ATRX has been shown to interact with EZH2.