

Summary:

This gene encodes an inosine triphosphate pyrophosphohydrolase. The encoded protein hydrolyzes inosine triphosphate and deoxyinosine triphosphate to the monophosphate nucleotide and diphosphate. This protein, which is a member of the HAM1 NTPase protein family, is found in the cytoplasm and acts as a homodimer. Defects in the encoded protein can result in inosine triphosphate pyrophosphorylase deficiency which causes an accumulation of ITP in red blood cells. Alternate splicing results in multiple transcript variants. [provided by RefSeq, Jun 2012]

Locus ID:

3704

MW:

15.2