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HA-1(H) Genotype Positive

National Cancer Institute

Source

National Cancer Institute. *HA-1(H) Genotype Positive*. NCI Thesaurus. Code C157564.

A genetic finding indicating that a subject expresses the HA-1(H) genotype and has a homozygous or heterozygous nucleotide substitution at position 416 of the ARHGAP45 gene where the guanine has been mutated to adenine, which encodes a variant of Rho GTPase-activating protein 45 where the arginine at position 139 has been replaced by histidine. The HA-1(H) form of ARHGAP45 protein can be processed into a nonapeptide that can be presented by HLA-A*0201 and other HLA-A isoforms, while the peptide produced by processing the protein resulting from transcription of the ARHGAP45 gene with guanine at position 416, which will have arginine at amino acid residue 139 (HA-1(R)) cannot bind HLA-A.