

Determination of IGHV gene SHM status

Date of result: 05/09/2016

Date of sample collection: 12/08/2016

Patient name: ***

Diagnosis: CLL

Tissue type: blood

Molecule type: genomic DNA

Utilized methodology

PCR amplification of IGHV-IGHD-IGHJ gene rearrangements with leader primers.

Genescan analysis

Bidirectional Sanger sequencing

Immunoinformatics analysis: IMGT V-Quest

Result: a productive IGHV3-23*01/IGHD4-17*01/IGHJ4*02 gene was detected. The rearranged IGHV gene had 96.2% nucleotide identity with the germline sequence of the IGHV3-23*01 gene.

Interpretation: following the 98% germline identity cut-off value which is used for discriminating CLL cases into the IG-mutated or IG-unmutated category, this case belongs to the IG-mutated category which is generally associated with favorable prognosis.

Signatures

[Report of an unmutated case]

Name of the Hospital/Lab

Determination of IGHV gene SHM status

Date of result: 05/09/2016

Date of sample collection: 12/08/2016

Patient name: ***

Diagnosis: CLL

Tissue type: blood

Molecule type: genomic DNA

Utilized methodology

PCR amplification of IGHV-IGHD-IGHJ gene rearrangements with leader primers.

Genescan analysis

Bidirectional Sanger sequencing

Immunoinformatics analysis: IMGT V-Quest

Result: a productive IGHV3-49*01/IGHD3-9*01/IGHJ4*02 gene was detected. The rearranged IGHV gene had 100% nucleotide identity with the germline sequence of the IGHV3-49*01 gene.

Interpretation: following the 98% germline identity cut-off value which is used for discriminating CLL cases into the IG-mutated or IG-unmutated category, this case belongs to the IG-unmutated category which is generally associated with adverse prognosis.

Signatures

[Report of a subset#2 case]

Name of the Hospital/Lab

Determination of IGHV gene SHM status

Date of result: 05/09/2016

Date of sample collection: 12/08/2016

Patient name: ***

Diagnosis: CLL

Tissue type: blood

Molecule type: genomic DNA

Utilized methodology

PCR amplification of IGHV-IGHD-IGHJ gene rearrangements with leader primers.

Genescan analysis

Bidirectional Sanger sequencing

Immunoinformatics analysis: IMGT V-Quest, ARResT/AssignSubsets tool

Result: a productive IGHV3-21*01/IGHD: not determined/IGHJ6*02 gene was detected. The rearranged IGHV gene had 96.8% nucleotide identity with the germline sequence of the IGHV3-21*01 gene.

Interpretation: following the 98% germline identity cut-off value which is used for discriminating CLL cases into the IG-mutated or IG-unmutated category, this case belongs to the IG-mutated category. However, this particular rearrangement belongs to stereotyped subset #2 which is associated with adverse prognosis regardless of the somatic hypermutation status (Baliakas et al. Blood 2015).

Signatures