t(3;12)(q27;p13)

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Disease

Non Hodgkin lymphomas (NHL)

Epidemiology

One case to date, a 78-year-old female patient with a multifocal lymphoma, CD20+ diffuse large B-cell lymphoma (DLBCL) type, presenting as a primary central nervous system lymphoma (PCNSL) (Montesinos-Rongen et al., 2003). PCNSL are extra nodal NHL localized to -and remaining in- the central nervous system.

Genes involved and proteins

BCL6

Location
3q27

Protein
706 amino acids; composed of a NH2-term BTB/POZ domain (amino acids 1-130 (32-99 according to Swiss-Prot) which mediates homodimerization and protein-protein interactions with other corepressors (including HDAC1 and NCOR2/SMRT) to constitute a large repressing complex, another transcription repression domain (191-386), PEST sequences (300-417) with a KKYK motif (375-379), and six zinc finger at the C-term (518-541, 546-568, 574-596, 602-624, 630-652, 658-681), responsible for sequence specific DNA binding. Transcription repressor; recognizes the consensus sequence: TTCCT(A/C)GAA (Albagli-Curiel, 2003).

GAPDH

Location
12p13.3

Protein
335 amino acids; possess a nucleotide binding site for NAD+, and sites for glyceraldehyde 3-phosphate binding; catalyzes the phosphorylation and oxidation of glyceraldehyde-3-phosphate to 1,3-biphosphoglycerate (interconversion), using NAD+ as electron acceptor. Role in endocytosis and in nuclear membrane assembly. Associates with microtubules and RAB2, which stimulates the recruitment of dynein, to regulate microtubule motility and cargo transport. Also binds mRNA and t-RNA; may participate in tRNA export and mRNA stability. Role in the cell cycle, in DNA repair, and in apoptosis associated with oxidative stress (reviews in Sirover, 1999; Hara and Snyder, 2006; Hara et al., 2006; Colell et al., 2009).

Result of the chromosomal anomaly

Hybrid gene

Description
Breakpoint in the intron 2 of GAPDH; leads to the juxtaposition of the GAPDH promotor region with the 2 first exons and the entire BCL6, inducing deregulated expression of BCL6.

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