t(3;9)(q27;p24)

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Clinics and pathology

Disease
Non Hodgkin lymphoma (NHL).

Epidemiology
Only one case to date, a gastric lymphoma (Chen et al., 2006).

Prognosis
No data.

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).

DMRT1
Location: 9p24
Protein
373 amino acids; contains a DM (Doublesex/Mab-3) domain (amino acids 72-118) and a Proline/Serine-rich region (aa 322-347) according to SwissProt; binds DNA. Transcription factor; sex determination gene of major role in the platypus and in birds; expressed at a higher level in the embryonic testis than in the embryonic ovary in vertebrates. Postnatally, DMRT1 is only expressed in undifferentiated spermatogonia and in Sertoli cells, and silenced in granulosa cells by repression of DMRT1 promoter by FOXL2 (Lei et al., 2009). Required for testicular development in vertebrates.

Result of the chromosomal anomaly

Hybrid gene

Description
5’ DMRT1 - 3’ BCL6.

References


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