Translocation t(8;14)(q24;q32) as a clue for the diagnosis of B cell prolymphocytic leukemia

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Clinical:

Age and sex
64 years old male patient.

Previous history
No preleukemia. No previous malignancy. No inborn condition of note.

Organomegaly
No hepatomegaly, no splenomegaly, enlarged lymph nodes (occurrence of an organomegaly on January 2011 with diaphragmatic lymph nodes and tonsil infiltration), no central nervous system involvement.

Blood

WBC : 14.6X 10^9/l
HB : 14.2g/dl
Platelets : 212X 10^9/l
Blasts : with 65% polymorphonuclear neutrophils, 27% lymphocytes and 8% monocytes. 45% of the lymphocytes present a typical morphology of prolymphocytes with enlarged and basophilic cytoplasm and presence of a unique, prominent nucleolus.

Cyto-Pathology Classification

Cytology
Prolymphocytic leukaemia

Immunophenotype
CD19+, CD5+(high),CD23+(low), CD22+(high), CD79b+(high), FMC7+, surface Ig Lambda (high). Matutes score = 2 (CD10-, CD20+ high, CD43-, CD81+ high)

Rearranged Ig Tcr: not done.

Pathology
Large infiltration of the tonsil by a monomorphic B-cell lymphoma with a diffuse growth pattern composed of small to medium-sized cells positive for CD5, compatible with the diagnosis of mantle cell lymphoma, but without expression of cycline D1.

Electron microscopy: not done.

Diagnosis
B cell prolymphocytic leukaemia

Survival

Date of diagnosis: 02-2009 (flow cytometry and FISH analysis) First conventionnal cytogenetic analysis performed on 01-2011.

Treatment: none to date
Complete remission : none
Treatment related death : no
Relapse : N/A
Status: Alive. Last follow up: 02-2011.

Karyotype

Sample: Blood sample
Culture time: 72 with DSP30+IL2
Banding: RHG

Karyotype at Relapse
46,XY,t(3;17)(q26;q12),t(8;14)(q24;q32)[20]
Other molecular cytogenetics results

FISH analysis using MYC break apart (Abbott-5J9101) and IgH break apart (Abbott-5J7301) probes on blood sample: confirmation of MYC and IgH rearrangements on respectively 89 and 90% of the cells. FISH analysis on tonsil sample: confirmation of MYC and IgH rearrangements on respectively 80 and 73% of the cells.

Other Molecular Studies

Results:

Complementary FISH analysis on initial blood sample (02-2009): - using MYC Break apart probe: presence of MYC rearrangement in 94% of the cells; - using CLL probe set (Abbott-8L5320): no deletion 17p, no deletion 11q, no trisomy12, no deletion 13q.

A) Monomorphic lymphoid proliferation with a diffuse growth pattern composed of small to medium-sized cell with slightly irregular nuclear contours (HES-stained section, x400 magnifications).
B) The lymphoma cells, intermingled with small reactive T cells, strongly express CD5 (immunohistochemistry on paraffin-embedded section, x400 magnifications).

Conventional RHG karyotype with the presence of a t(8;14)(q24;q32) [indicated by red arrows] associated with a t(3;17)(q26;q12).
FISH metaphase observed on blood sample using MYC break apart probe and showing a split of the fusion signal resulting in the 5'MYC signal located on the der(8) and the 3'MYC signal located on the der(14).
Interphasic nuclei of tonsil sample observed by hybridization in situ with MYC break apart probe showing a split of the fusion signal in 1 red and 1 green signals demonstrating the presence of MYC rearrangement.

Comments

MYC rearrangement in chronic lymphocytic disorder is a very rare event (<1% of CLL) (Lu et al., 2010). The presence of the t(8;14) translocation is preferentially associated with increased prolymphocytes (Huh et al., 2008; Merchant et al., 2003), and, indeed, is described as a recurrent abnormality in B cell prolymphocytic leukaemia (Merchant et al., 2003; Kuriakose et al., 2004; Crisostomo et al., 2007). In the case reported here, two years after the description of 14q32-IgH rearrangement on blood sample, the discovery of a t(8;14) by conventional karyotype was an important clue for the orientation of the diagnosis after reviewing cytologic and immunophenotypic data. In addition, these data lead to reconsider also the initial histopathologic hypothesis of mantle cell lymphoma all the more as no t(11;14) translocation as well as non hyperexpression of cycline D1 were present. Identification of a B cell prolymphocytic leukaemia is essential because of the potential rapid evolution with rising of leucocytes count and the poor response to CLL therapies (Swerdlow et al., 2008).

References


Crisostomo RH, Fernandez JA, Caceres W. Complex karyotype including chromosomal translocation (8;14) (q24;q32) in one case with B-cell prolymphocytic leukemia. Leuk Res. 2007 May;31(5):699-701


This article should be referenced as such: