

Case Report Section

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Translocation t(11;20)(p15;q11) detected in AML M0: A case report

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Clinics

Age and sex: 28 years old female patient.

Previous History:

- no preleukemia;
- no previous malignant disease;
- no inborn condition of note.

Organomegaly:

- no hepatomegaly;
- no splenomegaly;
- no enlarged lymph nodes;
- no central nervous system involvement.

Blood

Blasts: 52%

Cytopathology classification

Cytology: AML

Immunophenotype: Positive for CD13, CD33 and CD117

Rearranged Ig or Tcr: -

Pathology: MPO and SBB negative

Precise diagnosis: AML-M0 (FAB)

Survival

Date of diagnosis: 03-2007.

Treatment: 7+3 protocol (7 days of cytosine arabinoside continuous infusion + 3 doses of Daunorubicin), patient did not respond. Hence, she was treated with high dose of cytosine arabinoside together with etoposide; however, she didn't respond. In view of poor prognosis, she was discharged on 25/03/2007 and died on 26/03/2007.

Complete remission: none

Treatment related death: -

Relapse: -

Status: Dead

Survival: 1 month

Karyotype

Sample: Bone marrow

Culture time: 24/48h

Banding: G-banding

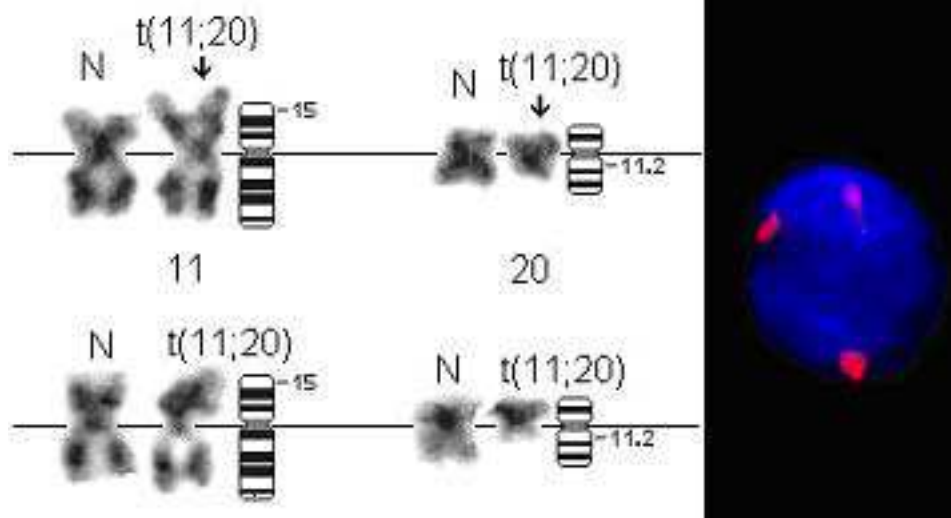
Results: 46,XX,t(11;20)(p15;q11) [100%, 24/24 cell].

Other molecular cytogenetic techniques: FISH with WCP probes for chromosome 11.

Other molecular cytogenetics results: Three spots of 11 in interphase cells. Due to low mitotic index metaphases could not be spotted. No more material could be tested for NUP98 and TOP1 fusion.

Comments

Chromosomal rearrangement between 11p15 and 20q11 has been reported in hematologic malignancies (Lam and Aplan, 2001), which results in fusion of the nucleoporin gene, NUP98 on chromosome 11p15 with topoisomerase 1, TOP1 gene located at 20q11. The NUP98 is a member of nuclear pore complex and involved in nuclear-cytosolic transport of RNA and protein complexes, and TOP1 is a protein whose c-terminus site of active tyrosine forms phosphodiester bond with 3'-strand of DNA and generates transient single strand DNA breaks (Wang 2002). In t(11;20), the rearrangement generates NUP98-TOP1 fusion protein from fusion of N-terminal portion of NUP98 (1-



G-banding shows $t(11;20)(p15;q11)$; Interphase FISH with three signals of chromosome 11.

514) with most of TOP1 gene (170-765), including core, linker and catalytic domains (Gurevich et al. 2004). It has been suggested that the NUP98-TOP1 has leukemogenic activities independent of topoisomerase activity. The $t(11;20)$ has also been described in patients with polycythemia vera, T-MDS and acute myeloid leukemia (Chen et al. 2003, Potenza et al. 2004, Panagopoulos et al. 2002). Our case aged 28 years with 60 kg weight has been reported with AML-M0 and probable NUP98 and TOP1 fusion. In addition, $del(3p)$, $inv(X)$ and $der(7)$ was observed in one single cell. Short survival of 2.5 - 30 months was reported after diagnosis (Sekikawa and Horiguchi-yamada, 2005). Our patient got the shortest survival of 23 days after diagnosis

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