Leukaemia Section
Short Communication

t(5;12)(q33;p13) ATF7IP/PDGFRB

Kenichiro Kobayashi
Department of Pediatric Hematology and Oncology Research Institute, National Center for Child Health and Development, 2-10-1 Okura Setagaya-ku Tokyo,157-8535, Japan. kobayashi-kn@ncchd.go.jp

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Abstract
Ph-like ALL is characterized by several chromosomal translocations involving activating cytokine receptor or tyrosine kinase such as CRLF2, ABL1, JAK2, and PDGFRB (Robert K.G et al, 2014). Recent increasing evidences suggest that patients with Ph-like ALL bearing PDGFRB translocation are potentiated to respond to tyrosine kinase inhibitors. Thus, this translocation should be included within the molecular companion diagnostics to facilitate tailor-made cancer therapy.

Keywords
Ph-like acute lymphoblastic leukemia, tyrosine kinase inhibitor (TKI), PDGFRB

Clinics and pathology

Disease
Ph-like acute lymphoblastic leukemia

Clinics
The patient is an 8-year-old male with B-ALL. Initial cytogenetics analysis showed a 45, XY, -7, add (12) (p13). RNA sequence analysis identified a novel translocation of ATF7IP/PDGFRB (Kobayashi K.et al, 2013). He showed good response to standard risk ALL therapy, but he relapsed in the continuation of the maintenance chemotherapy at 26 months after the diagnosis. He received 3 course of salvage therapies following by stem cell transplantation. Second generation dasatinib was commenced with the minimum residual disease (MRD) at day 60 post-transplant. The therapeutic response was prompt, with the disappearance of genomic-PCR based on MRD within 3 months, and he has maintained complete molecular remission for 12 months (Kobayashi K.et al, 2014).

Prognosis
As was shown in Ph-like ALL bearing PDGFRB translocation, i.e. EBF1/PDGFRB, t(5;12)(q33;p13) ATF7IP/PDGFRB translocation seems response to TKI.

Cytogenetics

Cytogenetics morphological
Bandng cytogenetics revealed 45, XY, -7, add (12) (p13). The mRNA sequence analysis identified an in-frame transcript fusing exon 13 of ATF7IP with exon 11 of PDGFRB, i.e. t(5;12)(q33;p13).

Genes involved and proteins

PDGFRB
Location
5q33
Protein
PDGFRB is a frequent target of chromosomal translocation in a broad spectrum of hematological malignancies.

ATF7IP
Location
12p13
Protein
ATF7IP acts as transcriptional regulators and is frequently overexpressed in cancer cells modulating
telomerase TERT and TERC gene expression (Liu, L. et al, 2009).

**Result of the chromosomal anomaly**

**Hybrid gene**

**Description**

5' ATF7IP-3' PDGFRB

**Fusion protein**

**Description**

Forced expression of ATF7IP/PDGFRB, not wild-type PDGFRB, conferred growth factor independence to murine Ba/F3 cells, indicating that coiled-coil domain from 5' ATF7IP- would favour subsequent constitutive activation of the PDGFRB tyrosine kinase domain.

**References**


*This article should be referenced as such:*