

ORIGINAL ARTICLE

Prognostic Factors in Philadelphia Chromosome-Positive Acute Myeloid Leukemia Using Fluorescence in Situ Hybridization

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SUMMARY

Background: Philadelphia chromosome-positive acute myeloid leukemia (Ph+ AML) is a rare leukemia subtype first classified by the World Health Organization in 2016. The incidence of Ph+ AML is approximately 0.5 - 3%, and its prognosis is poor. Ph+ AML with additional chromosomal abnormalities in children has rarely been reported, and its treatment and prognosis remain uncertain.

Methods: We retrospectively analyzed 649 patients with AML from 2006 - 2021. Six (0.9%) patients with Ph+ AML were identified and treated with conventional chemotherapy. The clinical features and prognoses were retrospectively analyzed.

Results: Six cases of AML with a Ph chromosome were reported. One of the six individuals exhibited a biphenotypic immunophenotype, one exhibited a simple myeloid immunophenotype, and the other four exhibited myeloid and lymphoid expression.

Karyotypic analysis (R banding) was performed in six cases, four of which were classical Ph chromosomal abnormalities, two of which had additional abnormalities outside the Ph chromosome. Fluorescence *in situ* hybridization (FISH) analysis using the BCR/ABL fusion gene distinguished that the BCR major breakpoint break in three cases was type P210 and the BCR minor breakpoint break in three cases was type P190. The complete remission rate of the six patients in this study using conventional chemotherapy was 60%, with a median survival time of 7.5 months.

Conclusions: In summary, Ph+ AML is a heterogeneous disease often associated with additional chromosomal abnormalities. Ph+ AML is seen with a lymphoid immunophenotype and alterations in associated genes such as the IGH gene. Adults were predominantly P210 and two cases in children were both P190. Conventional treatments are less effective, and there are no standard treatment regimens.

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Supplementary Data

Table S1. List of 22 types of FISH probes.

No.	Manufacturer	Probe Name	Target	Dye Color	Use in Figure
1	Beijing Jipuujia Medical Technology Co., Ltd.	CSP 16/CSP 22	16p11-q11/22p11-q11	R/G	Fig. 2G; Fig. 5D
2	Beijing Jipuujia Medical Technology Co., Ltd.	GLP EWSR1	22q12	R/G	Fig. 2F; Fig. 5E
3	Beijing Jipuujia Medical Technology Co., Ltd.	GLP RUNX1/ GLP RUNXIT1	21q22/8q22	G/R	
4	Beijing Jipuujia Medical Technology Co., Ltd.	GLP BCR/ GLP ABL(EB)	22q11/9q34	G/R	Fig. 2B, C; Fig. 5A
5	Beijing Jipuujia Medical Technology Co., Ltd.	GLP BCR/ GLP ABL (DB)	22q11/9q34	G/R	Fig. 2D; Fig. 5B
6	Beijing Jipuujia Medical Technology Co., Ltd.	GLP ASS	9q34	R	Fig. 2E; Fig. 5C
7	Beijing Jipuujia Medical Technology Co., Ltd.	GLP C-MYC	8q24	G/R	
8	Beijing Jipuujia Medical Technology Co., Ltd.	GLP MLL	11q23	G /R	
9	Beijing Jipuujia Medical Technology Co., Ltd.	GLP P16/CSP 17	9q21/17p11-q11	R/G	Fig. 2H; Fig. 5I
10	Beijing Jipuujia Medical Technology Co., Ltd.	GLP CBF3	16q22	G/R	
11	Beijing Jipuujia Medical Technology Co., Ltd.	GLP p53	17P13.1	R	
12	Guangzhou LBP Medicine Science & Technology Co., Ltd.	GLP CSF1R/ GLP D5S23, D5S721	5q33/5p	R/G	
13	Beijing Jipuujia Medical Technology Co., Ltd.	CSP 18/CSP X/ CSP Y	18p11.1-q11.1/Xp11.1- q11.1/Yp11.1-q11.1	A/G/O	
14	Beijing Jipuujia Medical Technology Co., Ltd.	GLP ETV6/ GLP RNUX1	12p13/21q22	R/ G	
15	Beijing Jipuujia Medical Technology Co., Ltd.	GLP TUPLE1/ GLP ARSA	22q11/22q13	R/G	Fig. 5F
16	Beijing Jipuujia Medical Technology Co., Ltd.	GLP 1P21/ GLP 1q36	1p21/1q36	R/G	Fig. 5G
17	Beijing Jipuujia Medical Technology Co., Ltd.	GLP TCF3/ GLP PBX1	19p13/1q23	G/R	Fig. 5H
18	Beijing Jipuujia Medical Technology Co., Ltd.	GLP IGH	14q32	R/G	Fig. 3
19	Guangzhou LBP Medicine Science & Technology Co., Ltd.	GLP D7S522/ CSP 7	7q/7p11-q11	R/G	
20	Beijing Jipuujia Medical Technology Co., Ltd.	GLP D20S108/ CSP8	20q12/8p11-q11	R/G	
21	Beijing Jipuujia Medical Technology Co., Ltd.	GLP IGH/ GLP CCND1	14q32/11q13	R/G	
22	Beijing Jipuujia Medical Technology Co., Ltd.	GLP PML/ GLP RARA	15q22/17q21	G/ R	