

혈액종양질환별 FISH 검사안내

혈액종양 질환의 진단과 예후 예측, 치료 후 추적관찰을 하기위해 FISH 검사의 중요성이 매우 높아졌습니다. FISH검사는 세포분열이 일어나지 않고 있는 세포에서도 검사가 가능하므로, 많은 수의 세포를 관찰할 수 있어 검사결과가 더욱 정확하고 예민하며 또한 분열세포를 얻지 못한 경우에도 매우 유용합니다. 많은 기관에서 FISH검사를 혈액종양질환의 진단시 스크리닝과 추적 관찰 표지자로 이용하고 있습니다.

FISH 검사의 적응증

- ① 혈액종양 질환 진단시 FISH panel test로 염색체 이상을 스크리닝 : 환자 진단과 예후 판정
 - ② 치료 후 환자의 추적관찰 : FISH검사로 반정량적인 모니터링 가능
 - ③ 염색체 검사 상 정상적인 핵형이나 No mitotic cell인 경우에도 염색체 이상 유무 검출 가능
- ▶ 혈액종양의 진단별로 가장 흔히 관찰되며, 기존의 핵형분석으로는 발견이 어렵거나, 예후에 중요한 염색체 이상을 중심으로 혈액종양질환별 panel test를 구성하였습니다.

Disease	Fish probe	Abnormality	Comment
AML panel	AML1/ETO	t(8;21)	Most common in AML
	PML/RARA	t(15;17)	Diagnostic for AML, M3:APL
	MLL	11q23	AML, Not detectable by karyotyping in 50%
	CBFB	Inv(16);t(16;16)	Diagnostic for AML, M4eo
ALL panel	BCR/ABL	t(9;22)	Unfavourable prognosis
	MLL	11q23	Unfavourable prognosis
	p16	9p21	Common genetic abnormality in ALL
	TEL/AML1	t(12;21)	Most common in pediatric ALL, good prognosis
MDS panel	EGR	5q31	Associated with Good risk
	7q22/7q35	Monosomy7 7q deletion	Associated with Poor risk
	CEP8	Trisomy 8	Most common in MDS and MPD
	X/Y	Y loss	Associated with Good risk
Multiple Myeloma panel	RB1	13q14	Poor prognosis
	IGH	14q32	Common in multiple myeloma
Lymphoma panel	IGH	14q32	IgH rearrangements:t(8;14), t(11;14), t(14;18)
	p16	9p21	Common in T-cell lymphoma

- **검체** : 각 질환 panel 별 골수 5 mL (전용용기 사용, Heparin tube)
- **검사/소요일** : 월~금/4일
- **검사수가** : 문의요망
- **검사문의** : 031) 260-9640 (세포유전학팀), 031) 260-9261 (고객지원부)

혈액종양질환별 FISH 검사안내

▶ WHO Classification of acute myeloid leukemia (AML)

Acute myeloid leukemia with recurrent genetic abnormalities

- Acute myeloid leukemia with t(8;21)(q22;q22);(AML1/ETO)
- Acute myeloid leukemia with abnormal bone marrow eosinophils inv(16)(p13q22) or t(16;16)(p13;q22);(CBFB/MYH11)
- Acute promyelocytic leukemia (AML with t(15;17)(q22;q12)(PML/RARA) and variant
- Acute myeloid leukemia with 11q23 (MLL) abnormalities

▶ Prognostic implication of genetic alteration in childhood precursor B lymphoblastic leukaemia.

Cytogenetic finding	Genetic alteration	Frequency	Prognosis
t(9;22)(q34;q11.2)	BCR/ABL	3~4%	Unfavourable
t(4;11)(q21;q23) ¹	AF4/MLL	2~3%	Unfavourable
t(1;19)(q23;p13.3)	PBX/E2A	6%(25% of pre-B-ALL)	Unfavourable
t(12;21)(p13;q22)	TEL/AML1	16~29%	Favourable
Hyperdiploid>50		20~25%	Favourable
Hypodiploid		5%	Unfavourable

¹prototype 11q23 translocation in precursor B ALL;other translocations may the MLL gene

▶ International prognostic scoring system for MDS

Score	0	0.5	1	1.5	2
Prognostic variables % blast	<5	5~10	-	11~20	20~30
karyotype**	good	intermediate	poor		
Cytopenisis	0~1	2~3			

**Karyotype : Good = normal, -Y, del(5q);

Poor = complex(>3 abnormalities)or chromosome 7 abnormalities

Intermediate = other abnormalities

■ 참고문헌

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