中南大学学报(医学版) 2012, 37(10) 983-989 DOI: 10.3969/j.issn.1672-7347.2012.10.003 ISSN: 1672-7347 CN: 43-1427/R

论著

荧光原位杂交技术检测多发性骨髓瘤染色体异常

曹鹏飞[1], 陈方平[1], 程晔[2]

1. 中南大学 湘雅医院血液科, 长沙 410008;
2. 中南大学 湘雅医学院检验系, 长沙 410078

摘要:

目的:探讨荧光原位杂交(FISH) 技术检测多发性骨髓瘤患者染色体异常的临床价值。方法:运用FISH 技术, 采用5种特异性DNA 探针检测20 例多发性骨髓瘤(MM) 患者染色体异常, 并与常规细胞遗传学分析结果比较。结果:FISH 显示20 例MM 患者中18 例出现染色体异常, 总异常检出率为90%, 其中14q32 易位65%(13/20), del(13q14) 阳性率为55% (11/20), 1q21 异常25% (5/20), p53 阳性率15% (3/20)。常规染色体分析异常检出率仅为15% (3/20)。结论:del(13q14) 和14q32 易位是MM 患者最常见的染色体异常。 FISH 比传统的染色体检查更敏感可靠, 并可作为MM 预后评估的一项指标。

关键词: 骨髓瘤 原位杂交技术 染色体畸变

Detection of chromosomal aberrations in multiple myelomawith fluorescence in situ hybridization

CAO Pengfei[1], CHEN Fangping[1], CHEN Ye[2]

1. Department of Hematology, Xiangya Hospital, Central South University, Changsha 410008;
2. Department of Clinical Laboratory, Xiangya School Medicine, Central South University, Changsha 410078, China

Abstract:

Objective: To detect of chromosomal abnormalities in multiple myeloma (MM) patients withfluorescence in situ hybridization (FISH).
Methods: FISH was performed in 20 MM patients using 5 specific DNA probes. The difference inchromosomal abnormalities was compared by FISH and other routine cytogenetic tests.
Results: Eighteen of the 20 patients showed chromosomal abnormalities (90%). The positiverates of t(14q32), del(13q14), dup(1q21), and p53 gene were 65% (13 in 20), 55% (11 in 20),25% (5 in 20), and 15%(3 in 20), respectively. The abnormal rate of the conventional chromosomeexamination was 15% only.
Conclusion: FISH is more sensitive than traditional chromosomal tests and can be used as an indexin prognostic evaluation for MM. Del(13q14) and t(14q32) are the most common chromosomalabnormalities in MM patients.

Keywords: myeloma in situ hybridization chromosomal aberration

收稿日期 2011-11-29 修回日期 网络版发布日期

DOI: 10.3969/j.issn.1672-7347.2012.10.003

基金项目:

卫生部科研课题 (WKJ2007-3-001)。

通讯作者: 陈方平,Email:xychenfp@2118.cn

作者简介: 曹鹏飞,硕士,主管技师,主要从事血液学检验及肿瘤基础研究。

作者Email: xychenfp@2118.cn

参考文献:

1. 孙燕, 石远凯. 临床肿瘤内科手册［M］.5版. 北京: 人民卫生出版社, 2007: 453.SUN Yan, SHI Yuankai. Handbook of clinical tumor department ofInternal Medicine［M］.5th ed. Beijing: People's Medical PublishingHouse, 2007: 453.
2. 张艳, 江滨, 黄晓军, 等. 多发性骨髓瘤的细胞遗传学研究［J］.中国实验血液学杂志, 2007, 15(1): 76-

78.ZHANG Yan, JIANG Bin, HUANG Xiaojun, et al. The cell geneticsstudy of Multiple myeloma［J］.Journal of Experimental Hematology,2007, 15(1): 76-78.
3. Sawyer JR, Waldron JA, Jagannath S, et al. Cytogenetics ndings in 200patients with multiple myeloma［J］.Cancer Genet Cytogenet, 1995,82(1): 41-49.
4. Schmidt-Wolf IG, Glasmacher A, Hahn-Ast C, et al. Chromosomalaberrations in 130 patients with multiple myeloma studied byinterphase FISH: diagnostic and prognostic relevance［J］.CancerGenet Cytogenet, 2006, 167(1): 20-25.
5. 张之南, 沈悌. 血液病诊断及疗效标准［M］.2 版. 北京: 科学技术出版社, 1998: 373-380.ZHANG Zhinan, SHEN Ti. Standard of diagnosis and treatment ofHematology［M］.2nd ed. Beijing: Science and Technology Press, 1998:373-380.
6. Walker BA, Wardell CP, Chiecchio L, et al. Aberrant global methylationpatterns affect the molecular pathogenesis and prognosis of multiplemyeloma［J］.Blood, 2011, 117(2): 553-562.
7. Calasanz MJ, Cigudosa JC, Odero MD, et al. Cytogenetic analysis of 280 patients with multiple myeloma and related disorders: primarybreakpoints and clinical correlations［J］.Genes Chromosomes Cancer, 1997, 18(2): 84-93.
8. Pinkel D, Straume T, Gray JW. Cytogenetic analysis using quantitative,high-sensitivity, fluorescence hybridization［J］.Proc Natl Acad SciUSA, 1986, 83 (9): 2934-2938.
9. Harrison CJ, Mazzullo H, Cheung KL, et al. Cytogenetics of multiplemyeloma: interpretation of uorescence in situ hybridization results［J］.Br J Haematol, 2003, 120(6): 944-952.
10. Steward AK, Fonseca R. Review of molecular diagnostics in multiplemyeloma［J］.Expert Rev Mol Diagn, 2007, 7(4): 453-459.
11. Terpos E, Eleutherakis-Papaiakovou V, Dimopoulos MA. Clinicalimplications of chromosomal abnormalities in multiple myeloma［J］.Leuk Lymphoma, 2006, 47(5): 803-814.
12. Avet-Loiseau H, A al M, Moreau P, et al. Genetic abnormalities andsurvival in multiple myeloma: the experience of the IntergroupeFrancophone du Myeloma［J］.Blood, 2007, 109(8): 3489-3495.
13. Dewald GW, Therneau T, Larson D, et al. Relationship of patientsurvival and chromosome anomalies detected in metaphase and /orinterphase cells at diagnosis of myeloma［J］.Blood, 2005, 106(10):3553-3558.
14. Nilsson T, Lenhoff S, Rylander L, et al. High frequencies ofchromosomal aberration in multiple myeloma and monoclonalgammaopathy of undertermined significance in dirct chromosomepreparation［J］.Br J Haematol, 2004, 126(4): 487-494.
15. Zojer N, Konigsberg R, Ackermann J, et al. Deletion of 13q14 remainsan independent adverse prognostic variable in multiple myelomadespite its frequent detection by interphase fluorescence in situhybridization［J］.Blood, 2000, 95(1); 1925-1930.
16. Chiecchio L, Protheroe RK, Ibrahim AH, et al. Deletion ofchromosome 13 detected by conventional cytogenetics is a criticalprognostic factor in myeloma［J］.Leukemia, 2006, 20(9): 1610-1619.
17. Schop RF, Jalal SM, Van Wier SA, et al. Deletions of 17p13.1 and 13q14 are uncommon in Waldenstrom macroglobulinemia clonal cellscells and mostly seen at the time of disease progression［J］.CancerGenet Cytodenet, 2002, 132(1): 55-60.
18. Chang H, Sloan S, Li D, et al. Multiple myeloma involving centralnervous system: high frequency of chromosome 17p13.1(p53)deletions［J］.Br J Haematol, 2004, 127(3): 280-284.
19. Reece D, Song KW, Fu T, et al. Influence of cytogenetics in patientswith relapsed or refractory multiple myeloma treated with lenalidomideplus dexamethasone: adverse e ect of deletion 17p13［J］.Blood, 2009, 114(3): 522525.
20. Smadja NV, Leroux D, Soulier J, et al. Further cytogeneticcharacterization of multiple myeloma confirms that 14q32translocations are a very rare event in hyperdiploid cases［J］.GenesChromosomes Cancer, 2003, 38(3): 234-239.

6．Feng-huang Zhan, Bart Barlogie, John Shaughnessy Jr.基因表达谱鉴定高危多发性骨髓瘤[J]. 中南大学学报(医学版), 2007,32(02): 191-203
7．申建凯*，董丽华，齐慧，张广森 .血清血管内皮生长因子和白细胞介素-6在多发性骨髓瘤中的临床意义[J]. 中南大学学报(医学版), 0,(): 68-71
8．申建凯*，董丽华，齐慧，张广森 .血清血管内皮生长因子和白细胞介素-6在多发性骨髓瘤中的临床意义[J]. 中南大学学报(医学版), 2005,30(1): 68-71
9．刘竞， 李昕， 桂嵘， 蒋铁斌， 王二华.PDCD5蛋白对地塞米松诱导的多发性
骨髓瘤细胞凋亡的影响及机制初探[J]. 中南大学学报(医学版), 2010,35(7): 725-
10．贾海涛1， 葛峰1， 卢心鹏1， 曾慧兰2， 李丽萍1， 陈智鹏1， 卢春花1.蛋白酶体抑制剂PS-341诱导骨髓瘤细胞凋亡的
蛋白质组学研究[J]. 中南大学学报(医学版), 2010,35(8): 784-
11．何群， 赵谢兰， 贺艳娟， 谭达人.硼替佐米联合地塞米松治疗多发性骨髓瘤的临床分析[J]. 中南大学学报(医学版), 2010,35(8): 864-
12．杨俊杰； 张广森； 陈新瑞； 裴敏飞； 韩照平； 申建凯；.72例多发性骨髓瘤单克隆蛋白分析[J]. 中南大学学报(医学版), 2001,26(2): 152-
13．胡硕； 邓豪余； 李新辉； 段华新； 梁昌华；.SPECT骨显像对多发性骨髓瘤诊断的应用[J]. 中南大学学报(医学版), 2002,27(2): 176-
14．沈比先； 周顺科； 张可建；.老年人非外伤性多椎体压缩的MRI分析[J]. 中南大学学报(医学版), 2001,26(3): 235-
15．王建军, 洪丽萍, 潘艺, 刘水平, 吴坤陆, 汤立军.MYETS1基因的原核表达及其在多发性骨髓瘤细胞系中的缺失分析[J]. 中南大学学报(医学版), 2012,37(1): 27-31
