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临床研究与应用

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隆突性皮肤纤维肉瘤免疫表型和COL1A1/PDGFB融合基因的临床应用研究

叶新青^①, 邝晓聪^②, 韦常宏^①, 黄俊琪^①, 叶洪涛^③

作者单位: ①广西医科大学附属肿瘤医院病理科(南宁市 530021); ②广西医科大学基础医学院病理生理学教研室; ③广西玉林市第一人民医院病理科

Clinical study of molecular markers and COL1A1/PDGFB fusion gene in dermatofibrosarcoma protuberans

Xinqing YE¹, Xiaocong KUANG², Changhong WEI¹, Junqi HUANG¹, Hongtao YE³

1Department of Pathology, The Affiliated Tumor Hospital of Guangxi Medical University, Nanning 530021, China.

2Teaching and Research Section of Pathophysiology, College of Basic Medicine, Guangxi Medical University, Yulin 530021, China.

3Department of Pathology, Yulin No. 1 People's Hospital, Yulin 537000, China.

摘要

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摘要

目的: 探讨隆突性皮肤纤维肉瘤(dermatofibrosarcoma protuberans, DFSP)诊断中免疫表型和荧光原位杂交(fluorescence in situ hybridization, FISH)检测COL 1A1/PDGFB融合基因的应用价值。方法: 观察73例DFSP中免疫组织化学标记物vimentin、CD34、CD99、S100、desmin、SMA和FISH检测COL 1A1/PDGFB融合基因的表达。选取85例非DFSP作为免疫组织化学的对照组, 10例非DFSP作为FISH检测COL 1A1/PDGFB融合基因的对照组。结果: vimentin、CD34、CD99、S100、desmin、SMA在73例DFSP中阳性率分别是100%、91.78%、61.64%、0、0、6.85%, 在对照组中不同程度表达, 其中CD34的表达在鉴别诊断中有意义。COL 1A1/PDGFB融合基因在DFSP的阳性率为86.96%(60/69), 对照组均阴性。结论: 在DFSP的诊断中, COL 1A1/PDGFB融合基因是DFSP较为特异性、敏感性的标记, 而CD34是DFSP相对理想的标记。

关键词: 隆突性皮肤纤维肉瘤, 免疫组织化学, FISH, COL 1A1/PDGFB 融合基因

Abstract:

Objective: To investigate the values of immunophenotype and the Collagen type 1 alpha1/Proto-oncogene Proteins c-sis (COL1A1/PDGFB) fusion gene in the diagnosis of dermatofibrosarcoma protuberans (DFSP). Methods: IHC markers and the COL 1A1/PDGFB fusion gene were detected by IHC staining and interphase fluorescence in situ hybridization (FISH) in 73 cases previously diagnosed as DFSP. A total of 85 and 10 non-DFSP cases were also included as controls for IHC staining and FISH, respectively. Results: In the 73 DFSP cases, the positive detection rates for immunohistochemical marker vimentin, CD34, CD 99, S100, desmin and SMA were 100%, 91.78%, 61.64%, 0, 0, and 6.85%, correspondingly. Protein expression levels in these cases varied from the control group, and CD34 expression was significantly different among the differential diagnoses. The positive detection rate for the COL1A1/PDGFB fusion gene was 86.96% (60/69), whereas the gene expression in the control group was negative. Conclusion: The COL 1A1/PDGFB fusion gene is a highly specific and sensitive marker in the diagnosis of DFSP. CD 34 is a suitable marker for DFSP.

Key words: dermatofibrosarcoma protuberans immunohistochemistry FISH COL1A1/PDGFB fusion gene

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通讯作者: 叶洪涛 E-mail: dr.hongtaoye@goolemail.com

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