sus observation in patients with colorectal cancer: a randomised study [J]. Lancet, 2007, 370(9604): 2020 - 2029.
[18] Morris M, Platell C, McCaul K, *et al*. Survival rates for stage Ⅱ colon cancer patients treated with or without chemotherapy in a population-based setting[J]. Int J Colorectal Dis,2007,22(8): 887 - 895.
[19] Figueredo A, Coombes M E, Mukherjee S. Adjuvant therapy for completely resected stage Ⅱ colon cancer[J]. Cochrane Database Syst Rev, 2008(3): CD005390.
[20] Sargent D, Sobrero A, Grothey A, *et al*. Evidence for cure by adjuvant therapy in colon cancer: observations based on individual patient data from 20,898 patients on 18 randomized trials[J]. J Clin Oncol, 2009, 27(6): 872 - 877.
[21] Benson A B, 3rd, Schrag D, Somerfield M R, *et al*. American Society of Clinical Oncology recommendations on adjuvant chemotherapy for stage Ⅱ colon cancer[J]. J Clin Oncol, 2004, 22(16): 3408 - 3419.
[22] Nordlinger B, Rougier P, Arnaud J P, *et al*. Adjuvant regional chemotherapy and systemic chemotherapy versus systemic chemotherapy alone in patients with stage Ⅱ-Ⅲ colorectal cancer: a multicentre randomised controlled phase III trial[J]. Lancet Oncol, 2005, 6(7): 459 - 468.
[23] Wolmark N, Yothers G, O'Connell M J, *et al*. A phase Ⅲ trial comparing mFOLFOX6 to mFOLFOX6 plus bevacizumab in stage Ⅱ or Ⅲ carcinoma of the colon: Results of NSABP Protocol C-08[J]. J Clin Oncol, 2009, 27:18s.
[24] Allegra C J, Yothers G A, O'Connell M J, *et al*. Overall survival (OS) and updated disease-free survival (DFS) results of the NSABP C-08 trial assessing bevacizumab (B) in stage Ⅱ and Ⅲ colon cancer [J]. J Clin Oncol, 2011, 29(Suppl): 3508.
[25] Andre T, Van Cutsem E, Schmoll H, *et al*. A multinational, randomized phase Ⅲ study of bevacizumab (Bev) with FOLFOX4 or XELOX versus FOLFOX4 alone as adjuvant treatment for colon cancer (CC): Subgroup analyses from the AVANT trial[J]. J Clin Oncol, 2011, 29(Suppl): 3509.
[26] Mahoney M R, Sloan J A, Hubbard J M, *et al*. Quality of life (QOL) for patients treated with FOLFOX with or without cetuximab (Cmab) following complete resection of colorectal cancer (CRC): Results from North Central Cancer Treatment Group (NCCTG) phase Ⅲ trial N0147[J]. J Clin Oncol, 2011, 29(Suppl): 3541.
[27] Des-Guetz G, Uzzan B, Morere J F, *et al*. Duration of adjuvant chemotherapy for patients with non-metastatic colorectal cancer[J]. Cochrane Database Syst Rev, 2010: CD007046.
[28] Paul J, Briggs A, Harkin A, *et al*. SCOT: Short Course Oncology Therapy--A comparison of 12 and 24 weeks of adjuvant chemotherapy in colorectal cancer[J]. J Clin Oncol, 2011, 29(Suppl): e14145.

(**收稿**:2011-08-23;**修回**:2011-10-09)
(编辑 汪勤俭)

个案与短篇

文章编号:1000-5404(2012)01-0004-01

Sturge-Weber 综合征伴脑皮质发育不良 1 例

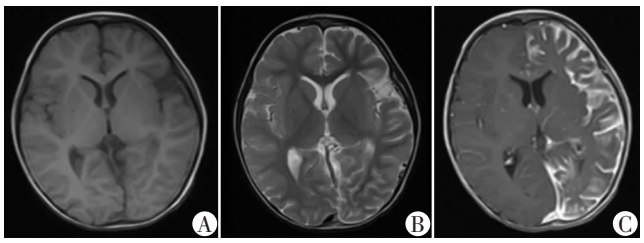
胡月华,史树贵,陈康宁,胡 俊 (400038 重庆,第三军医大学西南医院神经内科)

[**关键词**] Sturge-Weber 综合征;脑皮质发育不良
[**中图法分类号**] R742 [**文献标志码**] B

Sturge-Weber 综合征,又称为脑颜面血管瘤病或软脑膜血管瘤病,是一种先天性神经皮肤的发育异常,在临床上少见报道[1]。我科于 2011 年 8 月 20 日收治 1 例患儿,伴有脑皮质发育不良,分析其临床表现及 MRI、脑电图表现,现报告如下。

1 临床资料

患儿,男性,2 岁,因"阵发性抽搐 1 年余"来我科就诊。患儿无明显诱因出现发作性抽搐,伴有双眼凝视,牙关紧闭,肢体抽搐,持续数分钟,反复发作,间隔时间不等。患儿足月顺产,系第 1 胎第 1 产,母亲孕期体健无病毒感染史,非近亲婚育。否认出生时宫内窒息史,否认家族中有遗传病、传染病及类似疾病史。出生后发现额、面、颈部皮肤散在火焰状鲜红斑痣,以左侧为主,我院皮肤科诊断为"面部鲜红斑痣"。1 年前患儿家长发现患儿左眼角膜较右眼大,不伴畏光、流泪、哭闹等,偶尔左眼红,后发现双眼角膜逐渐透明、增大。我院眼科 A 超示双眼眼压增高,诊断为"青光眼"。入院查体:神志清,言语清,对答切题,双眼瞳孔等大等圆,对光反射灵敏。心肺腹(-),四肢关节活动无障碍,各肌群肌力及肌张力无异常,双下肢无水肿。双侧肱二、肱三头肌反射、膝跳反射、腹壁反射等生理反射均正常引出。双侧克氏征(-)、布氏征(-),双侧巴宾斯基征、霍夫曼氏征等病理征均未引出。辅助检查:脑电图示双侧频率不对称,左侧为 2 ~ 3 Hz δ 波或 4 ~ 5 Hz θ 波,右侧为 6 ~ 7.5 Hz θ 波或 α 波,左侧额、中央、颞区波幅较右侧降低。头颅 MR 检查示:左侧脑沟加深,增强后脑回信号增高,诊断为左侧大脑萎缩(图 1)。根据患儿临床表现及影像学特征,诊断为 Sturge-Weber 综合征。治疗给予抗癫痫、降低眼压等对症治疗。



A:T_1WI;B:T_2WI;C:增强 T_1WI

图 1 Sturge-Weber 综合征患儿头颅 MRI 表现

[**通信作者**] 胡 俊,E-mail:hujuncq@163.com

(下转 12 页)

达,检测了在不同浓度5-FU作用该组细胞和对照细胞的半数抑制率,实验结果表明,PDK-1干扰的LS174T细胞的IC_{50}数值显著低于对照组细胞。说明抑制PDK-1后结肠癌LS174T对5-FU治疗更敏感,使结肠癌细胞在较低的药物浓度下达到较高的凋亡率。

在哺乳动物体内目前鉴定出PDK 1~4这4种同工酶,其在不同的组织中表达丰度各异。我们既往研究表明PDK特异性抑制剂二氯乙酸盐(DCA)可增强5-FU诱导的结肠癌细胞凋亡[13],但究竟通过对何种同工酶作用尚无定论,本研究结果证实PDK-1抑制对5-FU诱导结肠癌细胞凋亡的促进作用。

总之,本研究表明通过RNA干扰抑制PDK-1表达联合5-FU可促进结肠癌细胞化疗诱导凋亡,为基因治疗和化学药物联合治疗大肠癌提供新的策略。

参考文献:

[1] Jemal A, Siegel R, Xu J, *et al*. Cancer statistics, 2010[J]. CA Cancer J Clin, 2010, 60(5): 277-300.

[2] Wolpin B M, Mayer R J. Systemic treatment of colorectal cancer[J]. Gastroenterology, 2008, 134(5): 1296-1310.

[3] Holness M J, Sugden M C. Regulation of pyruvate dehydrogenase complex activity by reversible phosphorylation[J]. Biochem Soc Trans, 2003, 31(Pt 6): 1143-1151.

[4] Roche T E, Hiromasa Y. Pyruvate dehydrogenase kinase regulatory mechanisms and inhibition in treating diabetes, heart ischemia, and cancer[J]. Cell Mol Life Sci, 2007, 64(7/8): 830-849.

[5] Rustum Y M. Clinical implications of 5-FU modulation[J]. Oncology (Williston Park), 1999, 13(7 Suppl 3): 22-25.

[6] Catenacci D V, Kozloff M, Kindler H L, *et al*. Personalized colon cancer care in 2010[J]. Semin oncol, 2011, 38(2): 284-308.

[7] Chen Y, Cairns R, Papandreou I, *et al*. Oxygen consumption can regulate the growth of tumors, a new perspective on the Warburg effect [J]. PLoS One, 2009, 4(9): e7033.

[8] Sheng H, Niu B, Sun H. Metabolic targeting of cancers: from molecular mechanisms to therapeutic strategies[J]. Curr Med Chem, 2009, 16(13): 1561-1587.

[9] 童晶涛, 梁后杰, 陈建芳, 等. 丙酮酸脱氢酶激酶-1在人结肠癌中的表达及意义[J]. 第三军医大学学报, 2010, 32(3): 257-260.

[10] Michelakis E D, Webster L, Mackey J R. Dichloroacetate (DCA) as a potential metabolic-targeting therapy for cancer[J]. Br J Cancer, 2008, 99(7): 989-994.

[10] Wong J Y, Huggins G S, Debidda M, *et al*. Dichloroacetate induces apoptosis in endometrial cancer cells[J]. Gynecol Oncol, 2008, 109(3): 394-402.

[12] Cao W, Yacoub S, Shiverick K T, *et al*. Dichloroacetate (DCA) sensitizes both wild-type and over expressing Bcl-2 prostate cancer cells *in vitro* to radiation[J]. Prostate, 2008, 68(11): 1223-1231.

[13] Tong J, Xie G, He J, *et al*. Synergistic antitumor effect of dichloroacetate in combination with 5-fluorouracil in colorectal cancer[J]. J Biomed Biotechnol, 2011, 2011: e740564.

(**收稿**:2011-08-31;**修回**:2011-11-29)

(编辑 汪勤俭)

(上接4页)

2 讨论

Sturge-Weber综合征主要是由于胚胎早期血管系统发育异常所致,可累及软脑膜及三叉神经分布区域的面部血管。Sturge于1879年最先提出患者癫痫发作可能是脑内有与面部血管瘤相似的血管瘤有关,Weber于1922年首次使用X线检查,发现此病患者的颅内有大片钙化灶,故又称之为Sturge-Weber综合征。

Sturge-Weber综合征的典型临床表现包括:偏侧性葡萄酒色或鲜红色血管痣、癫痫发作、智能减退,少数患者可合并青光眼与眼球突出,还可合并隐睾、脊柱裂等先天畸形[2-4]。CT和磁共振作为确诊该病的重要手段,可以表现为:脑叶萎缩,脑回状皮质钙化,脉络膜丛扩大,病侧颅骨增厚和血管畸形等。CT对钙化较敏感,可以清晰地显示颅内的病理改变,MRI能清楚地显示脑萎缩所引起的颅内解剖结构的细微改变和程度,还可以清楚地显示软脑膜血管瘤及脉络膜丛的扩大,以及深静脉的异常[5]。MRI增强扫描可精确显示软脑膜血管瘤及其伴发的血管异常(表现为特征性的脑回样强化),对定性诊断具有重要作用。本例患儿具有其典型的表现:颜面血管瘤、癫痫发作,同时患儿合并青光眼。脑电图提示左侧脑电波频率较右侧减慢,左侧脑电波的波幅较右侧降低,头颅MRI可见左侧大脑较右侧明显萎缩,增强后有脑回强化的典型表现。目前对该病无特殊的治疗方法,主要是对症处理,关于Sturge-Weber综合征继发青光眼,目前治疗以手术为主,同时予以控制癫痫发作,但患者一般预后差。

参考文献:

[1] 陈祖培, 杨小庆. 临床综合征影像学[M]. 北京: 科学技术文献出版社, 2000: 238.

[2] Thomas-sohl K A, Vaslow D F, Maria B L. Sturge-Weber syndrome: a review [J]. Pediatr Neurol, 2004, 30(5): 303-310.

[3] Ouigg M, Rust R S, Miller J Q. Clinical findings of the phakomatoses: Sturge-Weber syndrome [J]. Neurology, 2006, 66 (4): E17-E18.

[4] Comi A M. Update on Sturge-Weber syndrome: diagnosis, treatment, quantitative measures, and controversies [J]. Lymphat Res Biol, 2007, 5(4): 257-264.

[5] 陈星荣, 沈天真, 段承祥, 等. 全身CT和MRI[M]. 上海: 上海医科大学出版社, 1999: 104.

(**收稿**:2011-08-31;**修回**:2011-09-23)

(编辑 王小寒)