

磁共振成像

月刊
总第149期
2010年1月创刊

2024年第15卷第11期
2024年11月20日出版

刊名题写：时任第十一届全国人大常委会副委员长韩启德

主管单位 中华人民共和国国家卫生健康委员会

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首都医科大学附属北京天坛医院

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出版单位 《磁共振成像》
杂志社有限公司

发行范围 公开
发行单位 本刊发行部

国内发行 中国邮政集团有限公司
北京市报刊发行局
邮发代号 2-855
国外总发行 中国国际图书贸易集团有限公司
国外发行代号 M 8958
印刷单位 北京科信印刷有限公司

电话 010-67113815
E-mail editor@cjmri.cn
网址 www.chinesemri.com
定价 每册30元

国内统一连续出版物号 CN 11-5902/R
国际标准连续出版物号 ISSN 1674-8034
广告发布登记证号 京西市监广登字20170242号
本刊刊出的所有论文不代表本刊编委会的观点，除非特别声明

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脑胶质瘤是最常见的原发恶性脑肿瘤,发病率约为每十万人5~6例,其中异柠檬酸脱氢酶(isocitrate dehydrogenase, IDH)野生型胶质母细胞瘤占比最高,预后较差,5年生存率仅7%。2021年版世界卫生组织中枢神经系统肿瘤分类将异柠檬酸脱氢酶状态作为成人弥漫性胶质瘤的重要分子诊断依据, IDH表型对于患者的预后风险分层和治疗反应评估至关重要,与IDH野生型胶质瘤相比, IDH突变型与总生存期和治疗反应的改善相关。精准的分子分型诊断能协助临床制订个体化治疗方案,延长患者生存期。目前对于胶质瘤异柠檬酸脱氢酶状态的判断有赖于手术病理组织DNA测序技术,但操作有创,仍需要精准的术前无创评估方法。

MRI作为胶质瘤患者术前评估的重要组成部分,包括了常规序列和功能序列,两种序列可以从不同方面提供胶质瘤特征信息。常规MRI特征与胶质瘤IDH状态相关, IDH野生型胶质母细胞瘤更易出现环形强化、坏死以及瘤周水肿等影像学特征。扩散张量成像(diffusion tensor imaging, DTI)不仅可以评估组织内水分子扩散受限情况,还可以评估水分子扩散各向异性程度,从而反映肿瘤组织微观病理特征,采用直方图特征分析方法可以对肿瘤异质性进行定量分析,从而进一步评估肿瘤的生物学特征。然而,如何整合常规MRI视觉特征与DTI定量参数特征共同预测IDH基因状态有待进一步探索。

本研究分析了成人弥漫性胶质瘤患者术前常规MRI图像的视觉特征以及DTI定量参数特征,通过递归特征消除和Boruta算法进行特征筛选,首先采用高斯朴素贝叶斯模型分别建立常规MRI特征、常规DTI临床参数特征和DTI直方图特征的初级预测模型,并进一步基于堆叠泛化法集成各初级分类器与支持向量机,建立预测成人弥漫性胶质瘤IDH表型的集成式机器学习模型。结果显示模型具有良好的预测效能,能有效预测成人弥漫性胶质瘤的IDH表型,对于辅助临床制订个性化的治疗方案具有一定意义。详见内文第51页。

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CHINESE JOURNAL OF MAGNETIC RESONANCE IMAGING

磁共振成像
www.chinesemri.com

CHINESE JOURNAL OF MAGNETIC RESONANCE IMAGING

ISSN 1674-8034, CN 11-5902/R, CODEN CCIHBW, Established in 2010 Monthly Vol. 15, No. 11, Nov 20, 2024

Responsible Institution

National Health Commission of the People's Republic of China

Sponsor

Chinese Hospital Association
Beijing Tiantan Hospital of Capital Medical University

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Publishing

Publishing House of Chinese Journal of Magnetic Resonance Imaging

General Distributor

Domestic: Beijing Newspaper and Periodical Distribution Bureau of China Post Group Co., Ltd.
Postal Code: 2-855
Overseas: China International Book Trade Group Co., Ltd., P.O. Box 399, Beijing, China
Code No.: M 8958

Mail Order

Third Floor, Building 4, No. 358, Yudaihe East Street, Tongzhou District, Beijing 101100, China

Tel & Fax 8610-67113815

E-mail editor@cjmrj.cn

Website www.chinesemri.com

Price: USD 30.00

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About the cover

Glioma is the most common primary malignant brain tumor, with an incidence rate of about 5–6 cases per 100,000 people. Among them, isocitrate dehydrogenase (IDH) wild-type glioblastoma accounts for the highest proportion, with a poor prognosis and a 5-year survival rate of only 7%. The 2021 edition of the World Health Organization Classification of Tumors of the Central Nervous System uses IDH status as an essential molecular diagnostic basis for adult-type diffuse gliomas. IDH genotypes are crucial for the risk stratification of patients' prognosis and the assessment of treatment response. Compared with IDH wildtype gliomas, IDH mutation gliomas are associated with improved overall survival and treatment response. Accurate molecular typing diagnosis can assist in formulating individualized treatment plans in the clinic and prolong patient survival. Currently, the assessment of IDH status in gliomas predominantly depends on DNA sequencing of surgically obtained pathological tissue. However, this approach is invasive. Moreover, there remains a need for precise, non-invasive preoperative evaluation methods.

MRI is an essential part of the preoperative evaluation of glioma patients and includes conventional sequences and functional sequences. The two sequences can provide information on glioma characteristics from different aspects. The characteristics of conventional MRI are related to the IDH status of glioma. IDH wild-type glioblastoma is more likely to have imaging characteristics such as ring enhancement, necrosis, and peritumoral edema. Diffusion tensor imaging (DTI) can not only assess restriction of water molecule diffusion in tissues but also the degree of anisotropy of water molecule diffusion, thereby reflecting the microscopic pathological characteristics of tumor tissue. Histogram feature analysis can be used to quantitatively analyze tumor heterogeneity quantitatively, thereby further assessing the biological characteristics of the tumor. However, how to integrate the visual features of conventional MRI with the quantitative parameters of DTI to jointly predict the IDH gene status remains to be further explored.

This study analyzed the visual characteristics of conventional preoperative MRI images and the quantitative DTI parameter characteristics of adult-type diffuse glioma patients. We performed feature selection using recursive feature elimination and the Boruta algorithm. We first established a primary prediction model for the conventional MRI features, conventional DTI clinical parameter features, and DTI histogram features using a Gaussian Naïve Bayes model. We then ensembled each primary classifier with a support vector machine using the stacking method to create an ensemble machine learning model for predicting the IDH phenotype of adult-type diffuse gliomas. The results demonstrated the model's predictive solid efficacy and ability to accurately predict the IDH phenotype of adult-type diffuse gliomas, which is significant in assisting the clinical development of personalized treatment plans. Please see text page 51.

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