Short Communication

iMedPub Journals
http://www.imedpub.com

Research Journal of Nervous System

2022
Vol 6. No. 3

DNA methylation based molecular profiling to improve diagnostic accuracy for central nervous system

Nicholas G. Gottardo

Department of Haematology and Oncology, Princess Margaret Hospital for Children, Perth, Western Australia, Australia

Abstract

Central nervous system primitive neuro-ectodermal tumors (CNS-PNETs), have recently been re-classified in the most recent 2016 WHO Classification into a standby catch all category, "CNS Embryonal Tumor, not otherwise specified" (CNS embryonal tumor, NOS) based on epigenetic, biologic and histopathologic criteria. CNS embryonal tumors (NOS) are a rare, histologically and molecularly heterogeneous group of tumors that predominantly affect children, and occasionally adults. Diagnosis of this entity continues to be challenging and the ramifications of misdiagnosis of this aggressive class of brain tumors are significant. We report the case of a 45-year-old woman who was diagnosed with a central nervous system embryonal tumor (NOS) based on immunohistochemical analysis of the patient's tumor at diagnosis. However, later genome-wide methylation profiling of the diagnostic tumor undertaken to guide treatment, revealed characteristics most consistent with *IDH*-mutant astrocytoma. DNA sequencing and immunohistochemistry confirmed the presence of *IDH1* and *ATRX* mutations resulting in a revised diagnosis of high-grade small cell astrocytoma, and the implementation of a less aggressive treatment regime tailored more appropriately to the patient's tumor type. This case highlights the inadequacy of histology alone for the diagnosis of brain tumours and the utility of methylation profiling and integrated genomic analysis for the diagnostic verification of adults with suspected CNS embryonal tumor (NOS), and is consistent with the increasing realization in the field that a combined diagnostic approach based on clinical, histopathological and molecular data is required to more accurately distinguish brain tumor subtypes and inform more effective therapy.

Received: March 10, 2022; **Accepted:** March 15, 2022; **Published:** March 20, 2022

Biography

Nicholas G. Gottardo Department of Haematology and Oncology, Princess Margaret Hospital for Children, Perth, Western Australia, Austra published every year in the month of March and September. Submission of an article for publication in Purva Mimaansa implies that the work described has not been published earlier or under consideration for publication elsewhere. The College has zero-tolerance plagiarism policy. We check every issue using plagiarism prevention tool. The College reserves the right to publish or reject any contribution submitted for publication without assigning any reason. No Publication fee is charged from the contributors.