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Clinical impact of molecular profiling in rare brain tumors

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Abstract

Purpose of review: The purpose of this review is to describe the commonly used molecular diagnostics and illustrate the prognostic importance to the more accurate diagnosis that also may uncover therapeutic targets.

Recent findings: The most recent WHO Classification of Central Nervous System Tumours (2021) lists over 100 distinct tumor types. While traditional histology continues to be an important component, molecular testing is increasingly being incorporated as requisite diagnostic criteria. Specific molecular findings such as co-deletion of the short arm of chromosome 1 (1p) and long arm of chromosome 19 (19q) now define IDH-mutant gliomas as oligodendroglioma. In recent years, DNA methylation profiling has emerged as a dynamic tool with high diagnostic accuracy. The integration of specific genetic (mutations, fusions) and epigenetic (CpG methylation) alterations has led to diagnostic refinement and the discovery of rare brain tumor types with distinct clinical outcomes. Molecular profiling is anticipated to play an increasing role in routine surgical neuropathology, although costs, access, and logistical concerns remain challenging.

Summary: This review summarizes the current state of molecular testing in neuro-oncology highlighting commonly used and developing technologies, while also providing examples of new tumor types/subtypes that have emerged as a result of improved diagnostic precision.