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Pediatric tumor pathology: Challenges in diagnosis and classification.

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Introduction

Pediatric tumor pathology is a critical subspecialty within anatomical pathology, focusing on the identification and classification of neoplasms in children. Compared to adult tumors, pediatric tumors are biologically distinct, often arising from embryonic tissues and showing unique histological and molecular profiles. Accurate diagnosis and classification are essential for guiding therapy, yet several challenges complicate the diagnostic process in pediatric oncology. One of the primary challenges in pediatric tumor pathology is the rarity and diversity of neoplasms. Pediatric tumors account for less than 1% of all cancers, and many types are extremely rare. This rarity limits pathologists' exposure and experience, making it difficult to confidently identify unusual tumor entities. Additionally, some tumors exhibit overlapping histological features, such as Small Round Blue Cell Tumors (SRBCTs), which include Ewing sarcoma, rhabdomyosarcoma, neuroblastoma, and desmoplastic small round cell tumor. Distinguishing between these entities based on morphology alone is difficult. [1].

Molecular diagnostics have significantly improved the accuracy of tumor classification, but access and interpretation pose another challenge. Many pediatric tumors are now defined by specific genetic or epigenetic alterations, such as EWSR1 gene rearrangement in Ewing sarcoma or MYCN amplification in neuroblastoma. However, not all pathology laboratories are equipped with next-generation sequencing (NGS) or Fluorescence In Situ Hybridization (FISH) technologies. Furthermore, the interpretation of molecular data requires expertise in bioinformatics and oncogenic pathway analysis. Another obstacle lies in the evolving nature of tumor classification systems.These updates demand continual education for pathologists and may create inconsistencies between institutions [2].

Tissue sampling also poses significant challenges. Biopsies in children are often limited in size due to safety concerns, making comprehensive histological and molecular analyses difficult. Inadequate samples may result in misdiagnosis or non-diagnostic results, necessitating re-biopsies or reliance on ancillary testing such as imaging, which may be less definitive. [3]

Collaboration between pathologists, pediatric oncologists, radiologists, and surgeons is essential for accurate diagnosis and classification. Tumor boards and centralized pathology review panels can help mitigate diagnostic errors and harmonize treatment approaches across centers . Moreover, integrating artificial intelligence and digital pathology tools offers promise in enhancing diagnostic precision, especially in rare and ambiguous cases [4].

The World Health Organization (WHO) has updated its classification of pediatric tumors in recent years, incorporating molecular subtypes and grading schemes. For example, pediatric gliomas were previously classified based solely on histology but are now divided into low- and high-grade gliomas with defined molecular markers [5].

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Conclusion

Pediatric tumor pathology remains a complex and evolving field. The heterogeneity of tumors, limited tissue availability, lack of molecular resources, and frequent updates in classification systems all contribute to diagnostic challenges. Continued investment in training, technology, and collaborative frameworks is vital to improve outcomes for pediatric cancer patients.

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