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Pediatric Brain Tumours: Biological Mechanisms, Diagnostic Innovations, and Evolving Treatment Paradigms

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ABSTRACT:

Paediatric brain tumours represent the most common solid malignancies in children and remain a leading cause of cancer-related mortality in this population. Unlike adult brain tumours, paediatric neoplasms exhibit distinct molecular profiles, developmental origins, and therapeutic responses. This review provides a comprehensive overview of paediatric brain tumours, including epidemiology, molecular pathogenesis, clinical features, diagnostic approaches, and current treatment strategies. Emphasis is placed on recent advancements such as molecular classification, minimally invasive diagnostics, targeted therapies, and immunotherapeutic approaches. The article also highlights ongoing challenges, including treatment-related neurotoxicity and the need for precision-based paediatric oncology.

KEYWORDS: paediatric brain tumours, medulloblastoma, diffuse intrinsic pontine glioma, neuro-oncology, molecular diagnostics, targeted therapy.

INTRODUCTION

Brain tumours in children differ significantly from adult counterparts in terms of origin, biology, and clinical behaviour. These tumours arise during critical stages of neurodevelopment, often leading to profound neurological and developmental consequences. Paediatric brain tumours include a diverse group such as medulloblastomas, low- and high-grade gliomas, ependymomas, and brainstem gliomas.

Despite advances in surgical and medical therapies, managing paediatric brain tumours remains complex due to tumour location, treatment-related toxicity, and long-term sequelae affecting growth and cognition. Consequently, research has increasingly focused on understanding developmental neurobiology and tailoring therapies to paediatric-specific tumour subtypes.

Paediatric brain tumours account for approximately 20–25% of all childhood cancers worldwide.

Key Features

i. Age Distribution:

- Most common in children aged 0–14 years.
- Embryonal tumours such as medulloblastoma peak in early childhood.

ii. Tumour Types:

- Medulloblastoma: most common malignant tumour.
- Pilocytic astrocytoma: most common benign tumour.

iii. Survival Trends:

- Improved outcomes in low-grade tumours.

- Poor prognosis in high-grade tumours like diffuse intrinsic pontine glioma (DIPG).

iv. Gender Differences:

- Slight male predominance in most tumour types .

PATHOGENESIS AND MOLECULAR BASIS

Paediatric brain tumours originate from aberrations in neural progenitor cells during brain development.
Genetic and Molecular Alterations

- WNT and SHH Pathways: Critical in medulloblastoma subtypes
- H3K27M Mutation: Associated with aggressive midline gliomas
- BRAF Mutations: Common in low-grade gliomas
- MYC Amplification: Linked to high-risk disease

DEVELOPMENTAL ORIGIN

Unlike adult tumours, paediatric tumours are closely linked to disrupted neurodevelopmental signalling pathways.

Tumour Microenvironment

- Interaction between tumour cells and immature immune cells.
- Reduced immune surveillance compared to adults.

RISK FACTORS

i. Genetic Syndromes:

Neurofibromatosis type 1 and 2

- Tuberous sclerosis.
- Gorlin syndrome.

ii. Prenatal and Early-Life Exposures:

- Ionizing radiation.
- Environmental toxins (limited evidence).

iii. Family History:

- Rare but significant in hereditary syndromes.

CLINICAL PRESENTATION

Symptoms vary depending on tumour location and growth rate.

- Common Signs in Children.
- Persistent headaches.
- Morning vomiting.
- Visual disturbances.
- Ataxia and balance issues.
- Developmental regression.
- Seizures.

Infants may present with:

- Enlarged head circumference.
- Bulging fontanelle.

Diagnostic Approaches:

Neuroimaging

- MRI (Magnetic Resonance Imaging): Primary diagnostic tool.

Advanced Imaging Techniques:

- Diffusion-weighted imaging.
- Perfusion imaging.

Histopathology

- Essential for definitive diagnosis
- Integrated with molecular profiling

Molecular Diagnostics

- DNA methylation profiling.
- Identification of actionable mutations.

Liquid Biopsy

- Emerging tool using cerebrospinal fluid biomarkers.

Treatment Strategies

Surgical Management

- Aim: maximal safe resection.
- Use of neuro navigation and intraoperative imaging.

Radiation Therapy

- Limited in very young children due to neurotoxicity.
- Proton therapy reduces collateral damage.

Chemotherapy

- Multi-agent regimens tailored to tumor type.
- Often used to delay radiation in infants.

Targeted Therapy

- BRAF inhibitors for mutated gliomas.
- SHH pathway inhibitors.

Immunotherapy

- Vaccine-based approaches.
- CAR T-cell therapy under investigation.

Emerging Modalities

- Nanoparticle-based drug delivery.
- Epigenetic therapy.
- Tumour-specific viral therapies.
- Prognosis and Long-Term Outcomes.

Survival rates vary widely:

- Low-grade gliomas: >80% survival.
- Medulloblastoma: 60–70% survival.
- High-grade gliomas: poor prognosis.

Long-Term Challenges

- Cognitive impairment.
- Endocrine dysfunction.
- Secondary malignancies.
- Psychosocial effects.

Current Challenges

- Limited treatment options for aggressive tumours.
- Long-term toxicity in survivors.
- Blood-brain barrier limiting drug delivery.
- Tumour heterogeneity.

Future Directions

- Precision Medicine: Personalized therapy based on molecular profiling.
- Artificial Intelligence: Improved imaging and prediction models.
- Minimally Invasive Diagnostics: Liquid biopsy advancements.
- Combination Therapies: Integrating multiple modalities.

CONCLUSION

Paediatric brain tumours remain a critical challenge in modern medicine due to their complexity and impact on developing brains. Advances in molecular biology and targeted therapies are reshaping treatment approaches, offering hope for improved survival and quality of life. Continued interdisciplinary research and innovation are essential to overcome current limitations and develop safer, more effective treatments for children.

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