UPDATES ON RETINOBLASTOMA MANAGEMENT: ADVANCES AND CHALLENGES

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Retinoblastoma is a rare pediatric cancer that affects the retina of the eye. Advances in diagnostic and treatment modalities have led to improved outcomes for children with this disease. However, the management of retinoblastoma is complex and requires a multidisciplinary approach. This review provides an overview of recent updates in retinoblastoma management, including advances in diagnosis, treatment, and genetic testing.

The diagnosis of retinoblastoma is based on clinical examination, imaging, and pathology. Advances in imaging techniques, such as magnetic resonance imaging (MRI), have improved the accuracy of tumor staging and identification of high-risk features. The International Classification of Retinoblastoma (ICRB) system is used to stage retinoblastoma based on the extent of tumor involvement and helps guide treatment decisions.[1]

The treatment of retinoblastoma depends on the extent and location of the tumor, as well as the patient's age and overall health. Intra-arterial chemotherapy (IAC) has emerged as a promising treatment option for advanced and recurrent retinoblastoma. IAC delivers chemotherapy drugs directly into the ophthalmic artery, resulting in higher drug concentrations in the tumor and lower systemic toxicity.[2] Other treatment options include external beam radiation therapy, plaque brachytherapy, and enucleation. Retinoblastoma is associated with germline mutations in the RB1 gene, which is a tumor suppressor gene. Genetic testing for RB1 mutations is essential for accurate diagnosis and management of retinoblastoma. Advances in genetic testing, such as next-generation sequencing (NGS), have improved the detection of RB1 mutations and enabled the identification of new genes and pathways involved in the development of retinoblastoma.[3]

Advances in genetic testing and targeted therapies have led to the development of personalized medicine for retinoblastoma. Targeted therapies aim to inhibit specific molecular pathways involved in tumor growth and survival. For example, drugs targeting the mTOR pathway have shown promising

results in preclinical and clinical studies.[4] Immunotherapy, such as Chimeric antigen receptor (CAR)-based T cell therapy and adoptive T-cell therapy, is also being investigated as a potential treatment option for retinoblastoma.[5] Survivorship and long-term follow-up are essential components of retinoblastoma management. Children with retinoblastoma are at increased risk of developing second primary tumors, such as osteosarcoma, soft tissue sarcoma, and melanoma. Late effects of treatment, such as vision loss, hearing loss, and endocrine dysfunction, can also impact the quality of life of survivors. Comprehensive survivorship care, including regular surveillance for second primary tumors and management of late effects, is essential for optimal outcomes.[6]

Despite recent advances in retinoblastoma management, challenges remain in the diagnosis, treatment, and survivorship of this disease. These challenges include the need for early detection, the development of resistance to chemotherapy, and the risk of second primary tumors. Future directions in retinoblastoma management include the investigation of novel treatment strategies, such as combination therapy and gene editing, and the development of risk stratification algorithms to guide personalized treatment.[7]

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