Germline mutations in Dicer1 cause a pleiotropic susceptibility syndrome characterized by the development of pediatric or early-onset tumors including pleuropulmonary blastoma, Wilm's tumors, pineoblastomas, meningiomas, and neurofibromas (NF2). The variant, c.1552G>A;p.E518K in DGCR8, is also a hotspot somatic mutation in Wilms tumors. Using miRNA profiling, we found that tumors with the variant showed a specific miRNA profile different from wild type tumors. These findings reinforce that NF1 is one of the first in a cascade of mutations leading to GBM in these patients.

RARE-24. LARGE CONGENITAL NEOPLASMATIC NEVI AND NEOCUTANEOUS MELANOTYSIS: A RETROSPECTIVE CASE SERIES

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Neurocutaneous melanotysis (NCM) is a rare disease characterized by excessive proliferation and deposition of melanocytes in the leptomeninges and brain parenchyma, occurring in children with large congenital nevus (LCMN). Manifestations of NCM range from asymptomatic CNS melanocytic deposits to cranial neoplasms, meningiomas, and hydrocephalus. Patients with NCM are at risk for malignant melanoma. We conducted a retrospective, single-institution study of patients with LCMN evaluated at Memorial Sloan Kettering Cancer Center from June 2000 to January 2020. Of 55 patients studied, 15 had no radiographic NCM, and 40 had radiographic NCM at initial evaluation. MRI findings included: focal melanocytosis (33), diffuse leptomeningeal disease (4), solid melanoma (3). Malformations were identified in 13, including arachnoid cyst (4), congenital hydrocephalus (4), Dandy-Walker malformation (3), and tethered cord (3). Twenty-one patients completed imaging once and were followed clinically. Seventeen with serial imaging (10 with focal melanocytosis, 7 with normal MRI) remained stable over a median 24-month follow-up (range: 1–124).

INTRODUCTION: Everolimus is an inhibitor of mTORC1 (mammalian target of rapamycin complex 1), it is Health Canada and FDA approved for hydromyelia and renal angiomyxoma in the setting of tuberous sclerosis complex (TSC). There is little data available in regards to this treatment of TSC associated retinal astrocytoma (RA). Although the behaviour of RA is often indolent or slowly progressive, aggressive behaviour with retinal detachment and neovascular glaucoma requiring enucleation has been reported in several patients. Definite TSC diagnosis is established when either two major features or one major and two minor features are present. Probable TSC diagnosis is established when one major plus one minor feature is present. METHOIDS: We report a child with probable TSC, mosaicism from melanocytic nevi (LCMN). Manifestations of NCM range from asymptomatic CNS melanocytic deposits to cranial neoplasms, meningiomas, and hydrocephalus. Patients with NCM are at risk for malignant melanoma.

CONCLUSION: mTORC1 inhibition is effective therapy to preserve vision in the setting of retinal astrocytoma and tuberous sclerosis mosaicism.
Abstracts

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BACKGROUND: Choroid plexus tumors (CPT) include choroid plexus papilloma (CPP), atypical choroid plexus papilloma (aCPP), and choroid plexus carcinoma (CPC). Because of their rarity, limited data are available on the current status of treatment and outcomes for pediatric CPTs. METHODS: We retrospectively reviewed clinical information on patients with CPT aged between 0 and 30 years at diagnosis and were treated in 8 institutions in Japan. RESULTS: Of forty-two cases initially diagnosed as CPT, 18 cases were reviewed by central pathologists. As a result, the diagnosis of CPC or aCPP in five cases were changed to other tumors including AT/RT and astroblastoma. The remaining 37 cases were subjected to analysis. Median age at diagnosis was two years (0 to 25) and the duration of follow-up period was seven years. Among 26 patients with CPT (n=20) or aCPP (n=6) underwent gross-total resection without adjuvant therapy. Of them 24 patients are alive without recurrence. Four patients with patients of CPT (n=11) died of cancer. Five patients including three patients experienced local recurrence after resection as well as chemotherapy. All three patients with dissemination of CPC at diagnosis or relapse died of the disease. At least three patients were diagnosed with Li-Fraumeni syndrome: one died of medulloblastoma and one of a choroid plexus carcinoma. CONCLUSION: Because of the excellent prognostic of CPP, the survival rates for CPC, especially disseminated CPC are unsatisfactory. Our results also underline the importance of considering genetic testing of TP53 for patients with CPC.

RARE-27. DOUBLE MUTATIONS: DIFFERENT GERMLINE AND TUMOR MUTATIONS LEAD TO POOR OUTCOMES

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BACKGROUND: As genetic testing for both germline and tumor mutations has increased in completeness, complexity, and availability, more mutations and their impact on patient outcomes have been identified. METHODS: A retrospective review of pediatric patients who have identified germline mutations and a different tumor mutation was conducted. In follow-up period included documented initial diagnosis and outcome status, tumor mutation status, tumor mutation status, relapse status, and patient outcome. RESULTS: Six patients aged 8-13 years (median age 10 years) were identified for analysis. Four patients had pilocytic astrocytoma and two had high-grade gliomas. One of the patients with pilocytic astrocytoma also had MPNST diagnosed very early at age 9. The combination of germline/tumor mutations is as follows: Neurofibromatous Type I (NF1)/BRAF V600e, NF1, CHEK2/ MYB-QL1, NF1, Klfinefelter, ATM, MUTHY, CPC/BRF-KIAA fusions, NF1/BRF-KIAA (2 patients), and Marfan’s disease. The number of relapses per patient following initial diagnosis range from 3–7 with an average of 3.3. Four of the patients are alive and on therapy and two are deceased. The two deceased patients both had NF1/BRF-KIAA fusions and pilocytic astrocytomas. CONCLUSIONS: Patients with differing and compounded germline and tumor molecular genetic mutations have worse outcomes. These patients have more relapses and death when compared to those patients with one mutation, either germline or tumor. Broad molecular testing and germline testing for mutations is crucial in determining patient risk for poor outcomes.

RARE-29. PRIMARY CENTRAL NERVOUS SYSTEM NON-HODGKIN LYMPHOMA IN AN 11-YEAR-OLD BOY: A CASE REPORT

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RARE-30. A RARE CASE OF PRIMARY EWING’S SARCOMA OF THE CERVICAL SPINE

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Ewing sarcoma family of tumors predominantly affect the pediatric population in the long bones of the extremities or the pelvis, and only 8% of cases involve the spine. Primary Ewing’s sarcoma of the cervical spine is extremely rare and less than 30 cases have been reported in the literature thus far. Here we present a case of primary Ewing’s sarcoma of the cervical spine in a 28-year-old female who presented with a three-month history of neck pain and right arm radiculopathy. MRI revealed a homogeneously contrast enhancing, eccentric mass with dural tail at C2-C7. After undergoing a hemilaminectomy, histopathology confirmed extramedullary Ewing’s sarcoma with CD99 positivity. A comprehensive systemic and neuraxis work-up did not demonstrate any evidence of metastasis. Both ploidy and isochromosome 17q analysis was done. The patient was started on chemotherapy protocol as described below. To date, patient is in remission with no evidence of any residual disease in the cervical spine. In conclusion, although Primary Ewing’s sarcoma of the cervical spine is extremely rare it should be considered a differential diagnosis in patients with neck pain and a spinal mass under the age of thirty. Less than 25% of EFT’s present with overt metastasis and almost all have subclinical metastatic disease at the time of diagnosis, therefore, a comprehensive evaluation and systemic chemotherapy are recommended. With a multi-disciplinary approach of surgical decompression to preserve neurological functions, followed by comprehensive chemoradiotherapy regimens, reevaluation for local treatment, and adjuvant chemotherapy.

RARE-31. RECURRENT CHOROID PLEXUS CARCINOMA IN THE SETTING OF LI-FRAUMENI SYNDROME: REPORT OF TWO CHILDREN MANAGED WITH INTRAVENTRICAL RE-INDUCTION AND MARROW-ABLATIVE CONSOLIDATION CHEMOTHERAPY WITHOUT IRRADIATION FOLLOWED BY MOLECULARLY-TARGETED BIOLOGICAL THERAPY

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BACKGROUND: The optimal management for children with recurrent choroid plexus carcinoma (CPC) is not established. We report two children with germline TP53 mutations, whose CPC relapses were managed with marrow-ablative chemotherapy and oral biologically-targeted therapies. Pa-