in 21 patients including 13 in MB survivors. Mutations were inherited in 58/66 (88%) of cases in which inheritance could be tested and de novo in 8. In 6/67 families (9%), >2 children were diagnosed with a MB. CONCLUSION: In this large cohort of germline *SUFU* mutation carriers, MB in infants is the most frequent tumor but the spectrum also includes typical Gorlin syndrome tumors (BCC, meningiomas, and ovarian stromal/fibrous tumors) either as first tumors or as second malignancies. This broad tumor spectrum and the high risk of second malignancies justify the implementation of specific cancer surveillance programs.

RARE-22. GERMLINE PATHOGENIC VARIANT C.1552G>A;P.E518K IN DGCR8 CONFERS SUSCEPTIBILITY FOR SCHWANNOMATOSIS AND THYROID TUMORS

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Germline mutations in *DICER1* cause a pleiotropic susceptibility syndrome characterized by the development of pediatric or early-onset tumors including pleuropulmonary blastoma, Wilms tumors, pineoblastomas, multinodular goiter (MNG) and thyroid cancers. Somatic mutations in the other two microprocessors *DROSHA* and *DGCR8* have been found in Wilms Tumors and pineoblastomas. We present here two families with peripheral schwannomatosis and thyroid tumors carrying a germline variant c.1552G>A;p.E518K in *DGCR8*. Family one had six affected members with early-onset MNG and five of them developed schwannomatosis. All five members were heterozygous for the variant. One of the carriers had also been diagnosed with a choroid plexus papilloma at 7 years old. The common second event in all tumors tested was the loss of chromosome 22 at the somatic level. In family two, a 35-year-old male was diagnosed with a peripheral schwannoma at the age of 12. Since then, he has developed seven extra peripheral schwannomas (one of which was an ancient schwannoma) and papillary thyroid cancer. *DGCR8* lies on chromosome 22q, adjacent to the three schwannoma genes: *LZTR1*, *SMARCB1* and *NF2*. The variant, c.1552G>A;p.E518K localizes to the first RNA-binding domain of *DGCR8* and structural modelling predicts that it abolishes proper binding of RNA. It is also a hotspot somatic mutation in Wilms tumors. Using miRNA profiling, we show that this variant disrupts global microRNA production and *DGCR8* mutated tumors show a specific miRNA profile different from *DGCR8* wild type tumors. These findings reinforce *DGCR8* as a novel susceptibility gene for schwannomatosis and thyroid tumors.

RARE-23. NOVEL NF1 MUTATIONS IN TWO OCCURRENCES OF GLIOBLASTOMA MULTIFORM IN A PATIENT WITH CONSTITUTIONAL MISMATCH REPAIR DEFICIENCY SYNDROME

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Constitutional mismatch repair deficiency (CMMRD) syndrome is a rare cancer predisposition syndrome in children. Its main associated tumor types include brain and CNS tumors, hematologic malignancies, intestinal polyps and colorectal tumors, and other malignancies. Tumor genesis within this population is highly complex and poorly understood. We describe a case of a patient with two occurrences of glioblastoma multiforme (GBM), each with unique NF1 mutations. The patient is a female with CMMRD who was first diagnosed with GBM of the right frontal lobe in 2015. She subsequently underwent gross total resection, radiation to the field and concomitant and maintenance therapy with Temozolomide and Everolimus, due to high suspicion for NF-1. Genetic studies didn't show NF-1, instead revealing a diagnosis of CMMRD. Molecular testing of the GBM showed a high mutational burden and an NF1 mutation. Later, screening revealed stage IV colon cancer, for which she underwent subtotal colectomy, partial liver resection and chemotherapy. Molecular testing from the colon cancer found a hypermutant malignancy without mutations in NF1. Surveillance imaging detected a mass at the original site of her GBM, for which she had a resection. Notably, the genetic profile of the second tumor substantially different from the original tumor and the colon cancer sample, but had new mutations in NF-1. These findings highlight the significant variability in the genetic profiles of tumors in the context of CMMRD. It is also worth considering that NF1 is one of the first in a cascade of mutations leading to GBM in these patients.

RARE-24. LARGE CONGENITAL MELANOCYTIC NEVI AND NEUROCUTANEOUS MELANOCYTOSIS: A RETROSPECTIVE CASE SERIES

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Neurocutaneous melanocytosis (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 melanocytic nevi (LCMN). Manifestations of NCM range from asymptomatic CNS melanin deposition to cranial neuropathies, seizures, 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 (1). 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). Six had suspected radiographic progression of NCM without melanoma. Malignant melanoma developed in 11 patients, 5 with focal melanocytosis on initial imaging. Median time from focal melanocytosis identification to melanoma diagnosis was 80 months (range: 18–200). Median age at melanoma diagnosis was 9.9 years (range: 1.1–25.3). Median survival from melanoma diagnosis was 9.1 months (range: 1–60.4). Focal NCM on neuroaxis imaging does not predict time to transformation to malignant melanoma. Serial imaging is not indicated in absence of disease-modifying treatment. Clinical follow up of at-risk individuals is essential in early identification of complications.

RARE-25. RETINAL ASTROCYTOMA MTOR INHIBITOR THERAPY IN TUBEROUS SCLEROSIS MOSAICISM

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INTRODUCTION: Everolimus is an inhibitor of mTORC1 (mammalian target of rapamycin complex 1), it is Health Canada and FDA approved for SEGA and renal angiomyolipoma 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. METHODS: We report a child with probable TSC mosaicism, with negative serum NGS for TSC but RA and retinal achromic patch on the left. A left retinal peripapillary astrocytoma around optic nerve and very close to fovea was noted. There was concern that if it grew or there were to be any leakage it would cause visual impairment. This lead to therapy with everolimus 4.5 mg/m2/d aiming for level between 5 and 10 mcg/L. RESULTS: This boy has had a gradual reduction of the RA over the last 29 months, with healthy retina in the region no longer occupied by the lesion and preserved vision. He has tolerated therapy well with occasional mouth ulcers. CONCLUSION: mTORC1 inhibition is effective therapy to preserve vision in the setting of retinal astrocytoma and tuberous sclerosis mosaicism.

RARE-26. RETROSPECTIVE ANALYSIS OF PEDIATRIC CHOROID PLEXUS TUMORS

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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. METHOD: We retrospectively reviewed clinical information on patients with CPT patients 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 mean follow-up period was seven years. All 26 patients with CPP (n=20) or aCPP (n=6) underwent gross-total resection without adjuvant therapy. Of them 24 patients are alive without recurrence. Four patients of patients with CPC (n=11) died of cancer. Five patients including three patients experienced local relapse, achieved complete remission after resection of tumor plus chemoradiotherapy. 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 patient developed osteosarcoma. CONCLUSION: Compared with the excellent prognosis 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. Data collected included demographics, tumor type, germline mutation status, tumor mutation status, relapse status, and patient outcome. RESULTS: Six patients aged 8–13 years old (median age 10 years) were identified for analysis. Four patients had pilocytic astrocytoma and two had pilomyxoid astrocytoma. 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: Neurofibromatosis Type I (NF1)/BRAF v600e, NF1, CHEK2/MYB-QKI, NF1, Klinefelter, ATM, MUTYH, GPC3/BRAF-KIAA fusion, NF1/BRAF-KIAA (2 patients), and Marfan's/BRAF-KIAA. 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, which two are deceased. The two deceased patients both had NF1/BRAF-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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BACKGROUND: Primary central nervous system lymphoma (PCNSL) are very rare in children. CLINICAL CASE: An 11-year-old male presented with a 2 months history with myoclonic movements in the upper right limb, and a sudden frontal headache, gait disturbance due to right hemiparesis and an ipsilateral convulsive episode. Upon admission he had critical condition, with hypertensive skull syndrome, Glasgow of 12, Karnofsky 40%, right hemiparesis, swallowing disorder, facial paralysis, and loss of photo motor reflex and unilateral amaurosis. A CT and MRI showed a huge tumor mass in the left tempo-parietal region, infiltrating the white matter and shifting the midline. A Tumor biopsy was done, and reported diffuse small cell non-Hodgkin lymphoma of high-grade, Burkitt type. Systemic lymphoma workup was negative. He received six cycles of chemotherapy based on high dose methotrexate, rituximab and triple intrathecal.After the second cycle an ophthalmologic evaluation was done, and found infiltration to the right retina, for which 6 cycles of intra vitreous chemotherapy with methotrexate were applied, he showed an excellent response, and recovered all his neurological functions except that right hemianopia persist. Control MRI showed partial response at 2nd cycle and complete response after the 4th cycle. No Radiation was performed. CONCLUSION: This report highlights the fact that pediatric PCNSL may be effectively treated by a combination of HDMTX and rituximab-based chemoimmunotherapy without irradiation. Lack of awareness of this rare entity may lead to extense resections of brain, and potential permanent secuelae that were avoided in this illustrative case.

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 arise within 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 extraosseous Ewing's sarcoma with CD99 positivity. A comprehensive systemic and neuraxis work-up ruled out overt metastasis. We extrapolated data from children's cooperative group studies and IESS-II clinical trial to formulate a three phase treatment 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 is recommended. We recommend a multidisciplinary approach of surgical decompression to preserve neurological functions, followed by compressed chemotherapy 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 INTENSIVE 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-