mutations might increase sensitivity of cancer cells to some chemotherapy through modulating gene expression and/or interfering with DNA repair mechanisms, therefore, affecting treatment outcome.

RARE-44. CLINICAL CHARACTERIZATION AND OUTCOME; OUR EXPERIENCE OF CHORDOMAS IN PEDIATRIC AND YOUNG ADULTS
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Pediatric chordomas are exceedingly rare and there are limited data to guide treatment decisions. We report a retrospective analysis of 19 patients with chordomas who received treatment at our institution from 2001–2020. Of the 19 patients, 15 had clival (79%), 3 cervical and 1 sacral chordoma. There were 9 males (47%). Median age at diagnosis of 10.6 years. Eight patients (42%) had gross total and 11 (58%) had subtotal resection. As front line therapy 15 patients (79%) underwent surgery followed by radiation (1 photon and 14 proton), 3 patients (16%) received surgery and chemotherapy (anaplastic histology) and 1 patient received only surgery. For patients treated with radiation therapy the average prescribed dose was 70 Gy (range: 32–74). Post-surgery and radiation, 14 of 15 patients remained in remission. Five (26%) patients had progressive disease (PD) with median time to progression of 13 months of whom 3 died of disease at median of 18 months. Three of 6 patients of PD consisted of chemotherapy and radiotherapy for 3 re-resection with radiation for 1 and chemotherapy alone for 1 patient. The patients with metastatic and anaplastic disease mean survival is 21 month versus 45 for the rest of the cohort. In summary, post-operative adjuvant radiation provided an overall good outcome in majority of patients. Patients with anaplastic pathology and metastasis at diagnosis had worse outcome. Those who relapsed, subsequent treatment was palliative at best with short survival. Molecular analysis is warranted in future for better disease stratification.

RARE-45. SARCOMAS INVOLVING THE CENTRAL NERVOUS SYSTEM AT INITIAL PRESENTATION IN CHILDREN AND YOUNG ADULTS: A CASE SERIES
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Sung-Hui Tseng1, Kevin L.C. Huey7, Chiay-Yau Chang1, Jinn-Li Wang1,2, Jia-Rong Mei1, and Tai-Tong Wang1,2,4, National Taiwan University Medical Center, Taipei, Taiwan, 2Taipei Medical University Hospital, Taipei, Taiwan, 3Taipei Cancer Center, Taipei, Taiwan, 4Shuang Ho Hospital, New Taipei, Taiwan, 5Taipei Municipal Wan Fang Hospital, Taipei, Taiwan, 6City of Hope, Duarte, CA, USA
Sarcomas of bone, soft tissue, or neural origin may occasionally invade the central nervous system (CNS), causing diagnostic and therapeutic challenges. We aim to investigate the clinical features of sarcomas involving the CNS at initial presentation. During 2015/01–2019/12, nine consecutive patients (4 Males and 5 Females) younger than 30 years of age treated at a University Healthcare System in Northern Taiwan were included. The median age was 27 years (range, 2–24 years); diagnoses were Ewing Sarcoma with EWSR1 rearrangements (n=4), CIC-NUT1 Sarcoma (n=1), Osteosarcoma (n=2), Malignant Peripheral Nerve Sheath Tumor (MPNST; n=1), and extramedullary myeloid sarcoma (n=1). The tumors originated from the skull (n=4), vertebra (n=4), spinal canal (n=1), or extra-CNS sites (n=4). Four patients had metastases (1 Ewing sarcoma, 2 osteosarcoma, and 1 extramedullary myeloid sarcoma). The main symptom at diagnosis was facial/eye pain (n=2), back pain (n=3), arm weakness (n=1), or gait disturbance (n=3). Frontline neurosurgical decompression (n=7) or urgent radiotherapy (n=1) was performed in most patients. At a median follow-up duration of 20.1 months, the overall survival rate was 70%. All patients with Ewing sarcoma (n=4) and CIC-NUT1 sarcoma (n=1) achieved Complete Response after surgery, interval-compressed chemotherapy, radiotherapy, and adjuvant chemotherapy. Patients with stage IV osteosarcoma (n=2) had Partial Response; the patients with MPNST and extraskull myeloid sarcoma died of Progressive Disease at 18 and 3 months after diagnosis, respectively. We conclude that surgery, interval-compressed chemotherapy, radiotherapy, and adjuvant chemotherapy offers the best chance of cure but is not an option in infants with giant lesions; as in our case. We inform on alternative targeted treatment strategies and review the literature on these rare lesions. [i] Kambogiorgas D, St George EJ, Chapman S, English M, Solanki G: Infantile Chvost chondroma without clivus involvement: Case report and review of the literature, Childs Nerv System (2006) 22:1369-1374

RARE-47. DIFFUSE LEPTOMENINGEAL DISSEMINATED GLIONEURONAL TUMOR: CASE SERIES
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Diffuse leptomeningeal disseminated glioneuronal tumor (DL-GNT) is a rare brain tumor that presents as a plaque-like subarachnoid tumor, commonly involving the basal cisterns and interhemispheric fissure of children but rarely in adults. Here we report three cases of diffuse leptomeningeal dissemination diagnosed during childhood and adolescence. All patients presented with hydrocephalus. Imaging showed disseminated leptomeningeal involvement with minimal superficial periventricular abnormalities. The broadcast of the knowledge about this type of disease is important to increase awareness on this subject.

RARE-48. CHARACTERISTICS AND OUTCOME OF DIFFUSE LEPTOMENINGEAL DISSEMINATED GLIONEURONAL TUMOR (DLGNT): A SINGLE INSTITUTION EXPERIENCE
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Diffuse leptomeningeal glioneuronal tumors (DLGNT) are rare with an unknown etiology and unestablished incidence. Most frequently reported genetic alteration is KIAA1549-BRAF fusion. We present four DLGNT cases diagnosed between 2005–2018. Patient 1 is a female who presented with a 2-year history of back pain subsequently diagnosed with pilocytic astrocytoma. Re-imaging 3 months post-resection revealed a low grade glioma with BRAF duplication. Patient 2 is a female who presented with recurrent vomiting, dizziness, and hydrocephalus. The patient underwent biopsy which was consistent with oligodendrogliomatosic; no genetic analysis was done. Patient 3 is a male who presented with worsening headaches and intermittent vomiting. Approximately 5 months after resec- tion, imaging showed leptomeningeal disease and further testing revealed KIAA1549-BRAF fusion and 1p deletion. Patient 4 is a male who presented with hydrocephalus. Imaging showed disseminated leptomeningeal enhancement without a dominant mass lesion; biopsy and clinical history confirmed the diagnosis. All four patients received chemotherapy, Patients 1 and 3 underwent radiation therapy, and Patient 3 received a MEK-inhibitor to which he had a great response. However, the patient was non-compliant and had PD which continued despite re-starting therapy. Patients 1, 2, and 3 have died of progressive disease; survival was Patient 1, 276 days; Patient 2, approximately 7 years and 8 months, and Patient 3, 2 years and 11 months. Patient 4 remains alive with disease 4.5 years from diagnosis. There is much to be learned about this rare, poorly understood disease but hope for improvement through therapeutic targeting of the MAPK pathway.
RARE-50. TREATMENT RESPONSE OF CNS HIGH-GRADE NEOEPITHELIAL TUMORS WITH MN1 ALTERATION

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BACKGROUND: CNS high-grade neuroepithelial tumor with MN1 alteration (CNS HGNET-MN1) are a rare entity recently described as a highly aggressive tumor containing a mixture of brain tumor patterns with MN1 rearrangement. METHODS: CNS HGNET-MN1 patients were identified using genome wide methylation arrays across 5 institutions (the Hospital JP Garrahan, Hospital for Sick Children, the University Hospital Motol, Royal Children’s Hospital and Christchurch Hospital) and was correlated with treatment and outcome. Central imaging review with radiological features analysis was performed. RESULTS: We identified 9 patients harboring CNS HGNET-MN1 tumors through application of the Hendelberg brain tumor classifier. Seven tumors were supratentorial and two in the infratentorial. Median age was 5 (range 3.6–14.6). All patients had surgery (6 GTR and 3 STR) as initial management followed by radiotherapy (focal radiotherapy. Further multicenter, international prospective studies are required to determine the optimal treatment strategy for this group of tumors.

RARE-51. MOLECULAR INSIGHTS INTO MALIGNANT PROGRESSION OF CHORDOID PLEXUS PAPILLOMA (CPP)

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Malignant transformation of CPP is rare and the mechanisms remain elusive. We report a case of progression of papilloma into carcinoma where we performed molecular sequencing of both samples. A boy was found to have a brain mass soon after birth. The gross total resection (GTR) was diagnostic of CPP. Six years later he developed a recurrent mass that demonstrated progression to a chordoid plexus carcinoma (CPC). The patient received chemotherapy according to the “HeadStart” protocol. He is 3.5 years off therapy and disease-free. A sequencing study consisting of 1700 genes and tumor transcriptome was done. The analysis of both samples revealed a germline variant of TP53(R248W) with LOH and an allele frequency of ≥3% in the carcinoma that was not detected in the initial tumor. Two somatic substitutions were analyzed by Sanger sequencing. RESULTS: Pathogenic germline TP53 variants were present in 4 cases confirming Li-Fraumeni syndrome (LFS). Two patients have somatic TP53 substitutions. Only one patient with LFS harbored somatic TP53 deletion. In 7 patients, heterozygous substitutions of RB1 involved single exons 1 and pseudogenes were detected by MLPA. All these findings were validated by sCounter CNV assay. Additionally, four patients have WTI deletions, two patients – BRCA2, and in one case - NFI, concomitant with RB1 deletions in 3 cases. Interestingly, in one patient who faced a progression of CPP to CPC germline RB1 deletion was detected, and in both subsequent tumors, the length of the deleted region progressively increased. Notably, that RB1 deletions are mostly mutually exclusive to TP53 substitutions. 3 of 4 patients with RB1 deletions have follow-up period >1 year faced with tumor-related adverse events. CONCLUSION: Somatic or uncommon germline RB1 heterozygous deletions have been unraveled as a novel mechanism of aggressive CPP and could be implemented in prognosis definition schemes.

RARE-52. RB1 GENE DELETIONS ARE THE NOVEL MECHANISM OF CHORDOID PLEXUS TUMORS (CPT) ONCOGENESIS

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BACKGROUND: CPTs are known to be rare TP53-dependent neoplasms, while major molecular alterations underlying tumor progression, especially in TP53-null type cases, are still unclear. METHODS: 18 primary CPTs, including 16 chordoid plexus carcinomas (CPC) and two anaplastic chordoid plexus papillomas (CPP), were evaluated for copy number status of 87 major oncogenes and tumor suppressor genes by sCounter Cancer CNV assay by Nanostring and TP53 and RB1 by MLPA. Germline TP53 nucleotide substitutions were analyzed by Sanger sequencing. RESULTS: Pathogenic germline TP53 variants were present in 4 cases confirming Li-Fraumeni syndrome (LFS). Two patients have somatic TP53 substitutions. Only one patient with LFS harbored somatic TP53 deletion. In 7 patients, heterozygous substitutions of RB1 involved single exons 1 and pseudogenes were detected by MLPA. All these findings were validated by sCounter CNV assay. Additionally, four patients have WTI deletions, two patients – BRCA2, and in one case - NFI, concomitant with RB1 deletions in 3 cases. Interestingly, in one patient who faced a progression of CPP to CPC germline RB1 deletion was detected, and in both subsequent tumors, the length of the deleted region progressively increased. Notably, that RB1 deletions are mostly mutually exclusive to TP53 substitutions. 3 of 4 patients with RB1 deletions have follow-up period >1 year faced with tumor-related adverse events. CONCLUSION: Somatic or uncommon germline RB1 heterozygous deletions have been unraveled as a novel mechanism of aggressive CPP and could be implemented in prognosis definition schemes.

RARE-53. PINEAL PARENCHYMAL TUMOR OF INTERMEDIATE DIFFERENTIATION (PPTID) AND DICER1 SYNDROME: A CASE REPORT

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BACKGROUND: DICER1 syndrome is a rare inherited tumor predisposition syndrome linked to an increased risk of several malignancies. Affected individuals most commonly develop pleuropulmonary blastoma (PPB) and ovarian sex cord-stromal tumors. Brain tumors in these patients are rare, however; the increased frequency of pineoblastoma in this population has been established. Traditionally, pineal parenchymal tumors of intermediate differentiation (PPTIDs) have been considered associated with PPB and with DICER1 syndrome with research suggesting alternative mutations driving tumorigenesis. These tumors are pathologically and clinically diverse, with long-term surveillance based on therapeutic interventions. Here we describe a case of a germline DICER1 mutation in a patient with a PPTID, suggesting that this mutation is not limited to pineoblastoma as previously reported. CASE: We describe a 19-year-old female with a WHO grade III PPTID treated with multimodal therapy including surgery, craniospinal irradiation (CSI) and chemotherapy. She was noted to have a third mass at diagnosis and was subsequently diagnosed with a benign thyroid nodule, followed most recently by a cataract with pathology concerning for medulloepithelioma of the ciliary body. Due to the known association between medulloepithelioma and DICER1 syndrome, targeted germline sequencing was obtained and confirmed a pathogenic heterozygous mutation. CONCLUSION: To our knowledge this