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to better understand the biology and clinical phenotype. We summarize our institutional experience with spinal MPE including methylation-profiling. METHODS: A retrospective analysis of charts during the period of 2001 to 2019 of histologically proven MPE was done. We performed methylation profiling for 12 patients by Infinium MethylationEPIC Kit. RESULTS: 26 patients with spinal MPE were identified, median age of diagnosis was 34.2 years with a range of 11 to 59.9 years. Ten patients were between 1 and 5 years, lumbar spine location was common, 6 had leptomeningeal spread at diagnosis. All the patients underwent surgery and 11 received radiation following surgery. Eight patients below the age of 30 received radiation due to residual disease or metastases. Methylation profiling revealed 11,752 CpG sites differentially methylated between the younger and older patients (p < 0.05), however only one CpG cg22496254 associated with gene NAPC4/DCAF16 (role in promoting mitosis) was detectable with FDR < 0.25 that overly methylated in the younger age group. This is a new finding in MPE. CONCLUSIONS: Spinal MPE is a rare spinal tumor. Genetic alterations and limited reported cases characterize this tumor, making it a prime candidate for aggressive phenotyping, most requiring radiation. Methylation profiling reaffirmed this finding and trend in the younger patients. Prospective studies in a larger cohort of patients with methylation profiling are needed to identify prognostic variables and new targets for treatment.

EPEN-11. ONGOING RESPONSE IN A MULTIPLY RELAPSED METASTATIC POSTERIOR FOSSA EPENDYMOMA A AFTER VORINOSTAT AND CONCOMITANT IRRADIATION

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BACKGROUND: Among the nine molecular subgroups of ependymoma identified, posterior fossa ependymoma A (PF-EPN-A) confers the worst prognosis. These tumors often relapse despite aggressive resection and irradiation, resulting in limited therapeutic options. Although the genomic profile of PF-EPN-A does not typically show any recurrent alterations; they demonstrate distinct patterns of epigenetic changes which can be targeted with modulators such as histone deacetylase (HDAC) inhibitors. These inhibitors have shown efficacy in pre-clinical studies in both their anticancer and radiosensitizing properties. CASE: We describe a male diagnosed with a posterior fossa ependymoma at 3 years of age. After initial therapy with resection and focal irradiation, he went on to have a number of recurrences requiring multimodal therapy. Most recently, he developed diffuse intraventricular and leptomeningeal disease not amenable to surgical intervention. Genetic analysis demonstrated a BCOR mutation and methylation profile was consistent with PF-EPN-A. He received 23.4 Gray craniospinal irradiation with a 30.6 Gray boost to the nodular lesions. Vorinostat was given concomitantly for radio-sensitization in 2 week intervals for a total of 6 weeks. Serial imaging after irradiation revealed decreased tumor burden with progression post-vorinostat. MRI at 18 months exhibited mild interval growth of 2 lesions. CONCLUSIONS: To our knowledge, this is the first report of a clinical response in a pediatric patient with PF-EPN-A following irradiation administered concomitantly with vorinostat therapy. This response highlights the importance of further studies evaluating this combination therapy and its potential use in this population.

EPEN-13. PRIMARY EXTRADURAL SACROCOECCYGEAL SUBCUTANEOUS MALIGNANT EPENDYMOMA MISDIAGNOSED AS PILOLINAL CYST IN A 7 YEAR-OLD BOY: A CASE REPORT

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BACKGROUND: Ependymomas occur in the brain or spinal cord and rarely as an extradural variety at the sacrococcygeal region, separated from the spinal cord. This rare presentation is thought to originate from a group of heterotopic ependymal cells called the coccygeal medullary vestige. There are few reports of this occurrence in children. CLINICAL CASE: A 7-year-old male presented with a history of a soft mass arising in the sacrococcygeal area 3 years earlier, diagnosed as pilonidal cyst at primary level and treated with surgery twice, as this mass recurred the boy was sent to our hospital, a 3rd surgery was performed, all tumoral tissue was removed, no attachments. Pathology revealed a malignant ependymoma with positivity for PS100, EMA and Vimentin. After surgery a Follow up MRI of cranium and spine showed absence of disease, no radiotherapy neither chemotherapy was implemented. He has been on surveillance from 3 years now without recurrence. CONCLUSION: This report highlights the fact that pediatric ependymoma can have an extradural presentation and can be confounded with pilonidal cyst, total resection is needed to control the disease. Potential for recurrence or metastatic disease can continue 20 years from the time of primary tumor, so prolonged surveillance is important.

EPEN-14. GENERATION OF A CI10ORF95-RELA FUSION SPECIFIC ANTIBODY AS A DIAGNOSTIC TOOL FOR SUPRATENTORIAL EPENDYMOMA

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Ependymomas account for 10% of paediatric brain tumours and arise in the ventricular walls of the central nervous system. Ependymomas were previously classified as one tumour type and all patients received similar treatment. However, recent genomic studies have identified nine different molecular subgroups of the disease, including one supratentorial subtype characterized by a novel fusion gene CI10ORF95-RELA. When introduced into neural stem cells, this fusion is a potent driver of tumorigenesis and its presence in patient samples has previously been shown to negatively correlate with overall survival. Accurate diagnosis of this subgroup is currently required to adopt sophisticated approaches such asbreak-apart FISH or RNA sequencing. Here, we report the generation of a CI10ORF95-RELA Fusion-specific antibody that can be used for routine immunohistochemistry (IHC). Candidate antibodies were first selected using phage display and favourable leads were subjected to affinity maturation using ribosome display after a screening process involving immunoblotting and IHC. Further IHC-based screening of affinity-matured candidates using fusion-positive and -negative mouse tissue as well as human fusion-negative ependymoma tumour tissue revealed the antibody detects a nuclear staining pattern on fusion-positive tissues and does not react with fusion-negative tissues. This candidate antibody is currently being tested on human fusion-positive ependymoma tissue. This accurate diagnostic tool holds great promise to transform the management of patients with supratentorial ependymoma.

EPEN-16. TRANSCRIPTIONAL REGULATORY CIRCUITRIES AS MOLECULAR TARGETS IN EPENDYMOMA

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Genomic sequencing has driven precision-based oncology therapy; however, even genetic drivers remain unknown or non-targetable for many malignancies, demanding alternative approaches to identify therapeutic leads. Ependymomas comprise histologically similar tumors in young patients that are genetically different. These tumors display distinct somatic alterations from younger to older patients (p < 0.05), however only one CpG cg22496254 associated with gene NAPC4/DCAF16 (role in promoting mitosis) was detectable with FDR < 0.25 that overly methylated in the younger age group. This is a new finding in MPE. CONCLUSIONS: Spinal MPE is a rare spinal tumor. Genetic alterations and limited reported cases characterize this tumor, making it a prime candidate for aggressive phenotyping, most requiring radiation. Methylation profiling reaffirmed this finding and trend in the younger patients. Prospective studies in a larger cohort of patients with methylation profiling are needed to identify prognostic variables and new targets for treatment.

EPEN-17. FAVORABLE OUTCOME TO INTENSIVE CHEMOTHERAPY WITHOUT IRRADIATION IN INFANTILE METASTATIC EPENDYMOMA WITH A NOVEL MOLECULAR PROFILE: A CASE REPORT

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METASTATIC DISEASE AT INITIAL PRESENTATION OF INTRACRANIAL EPIENDYMOMA IS AN UNCOMMON OCCURRENCE WITH ONLY RARE REPORTS OF SURVIVAL AND IS REPORTEDLY MORE PREVALENT IN THE YOUNGER CHILDREN. CLINICAL AND MOLECULAR CHARACTERISTICS ASSOCIATED WITH METASTATIC PRESENTATION, THEIR PROGNOSTIC IMPLICATIONS, AS WELL AS OPTIMAL TREATMENT OPTIONS FOR SUCH PATIENTS, HAVE NOT BEEN IDENTIFIED. CASE REPORT: A SEVEN MONTHS OLD CHILD PRESENTED WITH POSTERIOR FOSSA ANAPLASTIC EPIENDYMOMA FOLLOWING SUB-TOTAL RESECTION OF PRIMARY TUMOR, A SPINE MRI REVEALED LEPTOMENOCEL INVASION ALONG THE CEREBRAL SPINAL CORD AND NERVE ROOTS OF THE CAUDA EQUINA. THE PATIENT WAS SUCCESSFULLY TREATED WITH FIVE CYCLES OF INTENSIVE INDUCTION CHEMOTHERAPY (AS PER HEAD START WITH HIGH-DOSE MESTHOTREXATE) FOLLOWED BY THREE SEQUENTIAL CYCLES OF MARILOM-BASED CHEMOTHERAPY AND AUTOGOLOUS HEMATOPOIETIC PROGENITOR CELL RESCUE (ASHPISR) WITHOUT IRRADIATION; HE IS CURRENTLY WITHOUT EVIDENCE OF DISEASE FOLLOWING SEVEN MONTHS FOLLOWING INITIAL DIAGNOSIS. MOLECULAR GENOMIC RESULTS: THE PATIENT WAS ENROLLED ON A PATIENT-CENTRIC COMPREHENSIVE MOLECULAR PROFILING PROGRAM, WHICH INCLUDED PAIRED TUMOR-NORMAL EXOME SEQUENCING, RNA SEQUENCING OF THE DISEASE-INVOLVED TISSUE, AND DNA METHYLATION PHENOTYPIC CLASSIFICATION. THE GENOMIC PROFILE OF THE TUMOR WAS REMARKABLY RESEMBLING, REVEALING ONLY A TERMINAL GAIN OF CHROMOSOME 3P AND A TERMINAL DELETION OF CHROMOSOME 22Q, SUGGESTIVE OF AN UNBALANCED TRANSLLOCATION. USING RNA SEQUENCING, WE IDENTIFIED A NOVEL FUSION BETWEEN RELA AND YAP1, A TUMOR HUB GENE INVOLVED IN TRANSCRIPTOMIC AND DNA METHYLATION PROFILES, FAILING TO DISCRETELY CLASIFY WITH WELL-ESTABLISHED EPIENDYMOMA SUBGROUPS. CONCLUSION: USE OF GENOMIC PROFILING TECHNIQUES PROVIDES MEANINGFUL INFORMATION FOR DISEASE CHARACTERIZATION ALLOWS FOR FURTHER EXPANSION OF THE MOLECULAR SPECTRUM ASSOCIATED WITH MALIGNANT DISEASE.

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