LG-60. OBJECTIVE RESPONSE TO VEMURAFENIB IN A CHILD TREATED FOR METASTATIC DESMOPLASTIC INFANTILE ASTROCYTOMA
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Desmoplastic infantile astrocytoma/ganglioglioma (DIA/DIG) is a rare primary neuroepithelial brain tumour, which typically occurs in infants aged between 1 and 24 months. Knowledge of the genetic background of DIA/DIG is very limited, recently the presence of BRAFV600E mutation was described in some cases of DIA/DIG. We present a case of 11-month old girl with 5-month history of nystagmus. Magnetic resonance imaging of the brain and spine showed contrast enhancing mass in the suprasellar region with spreading into both hemispheres and additional lesions in cerebellum and within the spine. Biopsy of the tumour was performed and histopathology examination revealed desmoplastic infantile astrocytoma. BRAFV600E mutation in the tumour cells was detected by molecular genetic testing. The patient started treatment according to SIOP LGG 1997 protocol with vincristine and carboplatin. After 2 months of therapy, the girl was admitted to our clinic with clinical signs of intracranial hypertension. MRI showed progression of the tumour size, which lead to development of hydrocephalus. The installation of ventriculo-peritoneal shunt was performed. Due to progression on the first line therapy, we decided to start second line individualized therapy with weekly dose of vinblastine in combination with BRAF inhibitor vemurafenib. After 6 months of therapy with vinblastine + vemurafenib partial remission was documented on MRI together with clinical improvement. Conclusion: BRAF V600E mutation affects a subset of DIA/DIGs and offers new therapeutic options.