allied academies

2nd International Conference on

Pathology and Surgical Pathology

July 05-06, 2019 | Paris, France

Recurrent EP300-BCOR fusions in pediatric gliomas with distinct clinicopathologic features

Sanda Alexandrescu

Boston Children's Hospital and Harvard Medical School, USA

BCOR is an epigenetic regulator and is genetically altered by mutation, deletion, or gene fusion in a range of cancers. "Central nervous system high-grade neuroepithelial tumor with BCOR alteration" is a recently described entity with characteristic internal tandem duplications within exon 15 of the BCOR gene (hereafter CNS HGNET-BCOR ex15 ITD). In this case series of three patients, we report the clinicopathologic, molecular (arrayCGH, RNA fusion analysis and targeted exome sequencing) and methylome features of gliomas with novel EP300-BCOR in-frame gene fusions, thus expanding the spectrum of BCOR alterations seen in CNS tumors. The gliomas in this series arise in children (age 10-18), involve the supratentorial compartment and have an infiltrative pattern of growth and a myxoid/microcystic background with frequent psammomatous calcifications and prominent chicken-wire vessels. All three cases had areas with low-grade morphology and two of them demonstrated histologic high-grade transformation. In contrast to CNS HGNET-BCOR ex15 ITD, they lack perivascular pseudorosettes. On methylation studies and a t-distributed stochastic neighbor embedding (tSNE) plot they cluster perfectly together, away from CNS HGNET-BCOR ex15ITD, consistent with a different entity. Gliomas with EP300-BCOR fusions and high-grade histology can demonstrate relatively rapid regrowth after debulking or subtotal resection. In conclusion, our study demonstrates that EP300-BCOR gliomas are a unique entity and calls for a more specific nomenclature for the existing HGNET-BCOR, as not all BCOR-altered gliomas are high-grade.

e: sanda.alexandrescu@childrens.harvard.edu