Liquid Biopsy for Detection of Histone H3 Mutations in Pediatric Diffuse Midline Gliomas

SAN DIEGO (April 17, 2019) — Winner of the Columbia Softball Pediatric Award, Amanda Muhs Saratsis, MD, presented her research, Liquid Biopsy for Detection of Histone H3 Mutations in Pediatric Diffuse Midline Gliomas, during the 2019 American Association of Neurological Surgeons (AANS) Annual Scientific Meeting.

Pediatric diffuse midline glioma (DMG) is a highly morbid tumor not amenable to surgical resection. Mutations in histone H3 encoding genes occur in 80 percent of cases and portend a worse clinical prognosis. Dr. Saratsis was the first to detect H3 mutations in circulating tumor DNA (ctDNA) in cerebrospinal fluid (CSF) derived from children with DMG and high grade glioma. This project describes a collaborative effort to develop a more sensitive and specific approach for H3 mutation detection and quantification in plasma and CSF with low starting [ctDNA], with validation performed across multiple centers.

The study demonstrates the utility of liquid biopsy for identifying H3K27M mutations in plasma and CSF with low starting [DNA], representing a rapid, minimally invasive method for diagnosis and therapeutic monitoring of pediatric brain tumors. Currently underway is a study of ddPCR of additional pediatric glioma liquid biopsy specimens.

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