7316-2141 [redacted]

Results

Pathology Surgical [redacted]

Name	MRN	DOB	Sex
[redacted]			

Lab Collection Information

Collected: [redacted]

Component Results

Component

Pathology Surgical (Corrected)

[redacted]

ATRT
Temporal lobe
Parietal lobe

ADDENDUM

Addendum #2 [redacted]

The INI1/SMARCB1 MLPA assay was performed on DNA isolated from the patient peripheral blood [redacted]. The blood sample demonstrated a normal pattern. No deletion was detected in the blood sample, which suggests the deletion in the tumor sample is a somatic change. This test cannot rule out a germline mosaicism.

[redacted]

[redacted]

Addendum #1 [redacted]

The INI1/SMARCB1 MLPA assay was performed on DNA isolated from the patient tumor tissue. The tumor sample demonstrated a homozygous deletion of exons 1-9 of the INI1/SMARCB1 gene.
GNAZ, which is proximal to the INI1/SMARCB1 gene, was also homozygously deleted. PPIL2, proximal to the gene, and SNRPD3 and SEZ6L, distal to the gene, were heterozygously deleted.

MLPA analysis of the tumor tissue from this patient demonstrated a homozygous

deletion in chromosome band 22q11.2 that resulted in homozygous deletion of the SMARCB1 gene.
The identification of the homozygous deletion in the tumor sample is consistent with the clinical diagnosis of atypical teratoid rhabdoid tumor.
One of the two deleted alleles could be a germline deletion, which predisposes the patient to atypical teratoid rhabdoid tumor.
Analysis of a peripheral blood specimen is recommended to rule out a germline deletion.

[redacted]

[redacted]

FINAL DIAGNOSIS

A: Brain, left parietal lobe, tumor resection:
-Atypical teratoid rhabdoid tumor (AT/RT)

B: Brain, left temporal lobe, tumor resection:
-Atypical teratoid rhabdoid tumor (AT/RT)

C: Brain, left temporal lobe, tumor resection:
-Atypical teratoid rhabdoid tumor (AT/RT)

SYNOPTIC REPORT

History of Previous Tumor/Familial Syndrome:
None

Specimen Type/Procedure:
Tumor resection

Specimen Handling:
Frozen section
Formalin fixed tissue for routine permanent paraffin sections
CBTTC

Specimen Size:
See gross description

Laterality:

Left

Tumor Site:
Cerebrum

Histologic Type and Grade:
Atypical teratoid/rhabdoid tumor AT/RT

Histologic Grade (WHO histologic grade):
WHO grade IV

[redacted]

SYNOPTIC REPORT (Continued)

Ancillary Studies:
Genetic studies

MEDICAL HISTORY

[redacted]

PRE-OP DX: BRAIN MASS
POST-OP DX: SAA
PROCEDURE: CRANIOTOMY FOR BRAIN MASS RESECTION
HISTORY: 12 MONTH OLD MALE WITH RIGHT TWITCHING FACIAL AND DROOLING. CT SHOWING PREDOMINANTLY CYSTIC MASS WITH CALCIFICATION LEFT PARIETAL.

TISSUES

A. PARIETAL LOBE, NOS - LEFT PARIETAL LESION, B. TEMPORAL POLE - LEFT TEMPORAL BIOPSY,
C. TEMPORAL POLE - TEMPORAL LESION - LEFT

GROSS DESCRIPTION

A. The specimen is received fresh for frozen section in a container labeled with the patient's name, medical record [redacted] number and designated "left parietal lesion". It consists of pink-white, irregular soft tissue fragments measuring 3 x 2 x 0.5 cm in aggregate. Two vials of blood are received and submitted per the CBTTC protocol. A representative section is submitted for frozen section and then sent for cytogenetics. Rep. sections are submitted per the CBTTC protocol. The remaining tissue is submitted entirely for permanent section in one cassette (A).

B. The specimen is received fresh for frozen section in a container labeled with the patient's name, medical record [redacted] number and designated "left temporal biopsy". It consists of multiple red-tan, pink-white irregular soft tissue fragments measuring 4 x 3 x 0.8 cm in aggregate. A representative section is submitted for frozen section and then sent for cytogenetics. Rep. sections are submitted per the CBTTC protocol. The remaining tissue is submitted entirely for permanent section in two cassettes (B1 and B2).

C. The specimen is received fresh in a container labeled with the patient's name, medical record number [redacted] and designated "left temporal lesion". It consists of a 1 x 0.9 x 0.5 cm in aggregate of pale tan to red tan gelatinous tissue. The specimen is entirely submitted in one cassette (C).

[redacted]

FROZEN SECTION DIAGNOSIS

A. Possible desmoplastic infantile ganglioglioma/astrocytoma (DIG), deferred to permanent.

B. Favor high grade tumor, AT/RT

[redacted]

MICROSCOPIC DESCRIPTION

Microscopic examination was performed on H&E stained sections and additional sections stained with immunoperoxidase methods for INI-1, BRG-1, EMA, NFP, SMACT, MIB-1/Ki-67, and vimentin.

Microscopic sections demonstrate a patternless, highly cellular tumor composed of medium to large size cells with an epitheliod appearance and prominent nucleoli. In specimen A the tumor cells are spindled with focal fascicular pattern and a certain degree of desmoplasia.
All specimens demonstrate necrosis, numerous mitoses and apoptosis.

The proliferative index, measured with Ki-67, is high, approximately 50%-60%. INI-1 expression is lost in the tumor cells, but retained in vessel endothelial cells and in the reactive areas. BRG-1 is retained. SMA highlights only the vessels and, EMA, NFP and vimentin are all strong but patchy.

Please note that the frozen section of specimen A demonstrated a desmoplastic pattern suggestive of DIG (parietal lesion).
The frozen section of specimen B (temporal lesion) revealed a malignant, high grade tumor suggestive of AT/RT. The latter diagnosis was comunicated to the neurosurgeon and the possibility of DIG was excluded.

DISCLAIMER

"These tests were developed and their performance characteristics determined by the Pathology Department at [redacted] They have not been cleared or approved by the U.S. Food and Drug Administration.