

**ADDENDUM**Addendum #1

This addendum is issued to report the result of FISH.

Interphase FISH studies were performed using a probe set for the Ewing sarcoma locus in chromosome band 22q12. The results were abnormal, consistent with a translocation that interrupts this locus.

Previous studies, by FISH and by RT-PCR at an outside laboratory, of the chest mass from this patient demonstrated a EWS translocation. The results are therefore consistent with metastatic Ewing sarcoma/PNET.

DNA has been isolated from the frozen tumor if a SNP array would be helpful in evaluating this patient. Previous array studies were non-contributory.

Addendum Signed \_\_\_\_\_ (signature on file) \_\_\_\_\_

**FINAL DIAGNOSIS**

History of chest wall Ewing sarcoma/ primitive neuroectodermal tumor family  
with EWSR1/FLI1 fusion confirmed by RT-DNA amplification status  
post treatment.

Brain, site not specified, biopsy:

-Metastatic Ewing sarcoma/primitive neuroectodermal tumor (ES/PNET) family

[REDACTED]

## MEDICAL HISTORY

DOCTOR'S NAME: [REDACTED]  
PRE-OP DX: BRAIN TUMOR  
POST-OP DX: PENDING  
PROCEDURE: CRANIOTOMY, BRAIN TUMOR EXCISION  
HISTORY: FIVE YEAR OLD WITH HISTORY OF EWING'S SARCOMA

## TISSUES

A. BRAIN, NOS - METASTATIC TUMOR, B. BRAIN, NOS - METASTATIC TUMOR

## GROSS DESCRIPTION

A. The specimen is received fresh for frozen section in a container labeled with the patient's name, medical record number and designated "metastatic brain tumor". Specimen consists of multiple irregular bloody fragments of red soft tissue 2.3 x 1.6 x 0.5 cm. Touch preps are performed, a representative section is submitted for frozen section. The frozen section is submitted for cytogenetic studies. The remaining tissue is entirely submitted in cassette A.

B. The specimen is received fresh in a container labeled with the patient's name, medical record number and designated "metastatic brain tumor". The specimen consists of an irregular portion of red soft focally cauterized tissue 1 x 0.8 x 0.5 cm.

The specimen is entirely submitted according to "CBTTC" protocol.

## FROZEN SECTION DIAGNOSIS

HISTORY OF EWING'S SARCOMA; NOW WITH BRAIN LESIONS.  
ROUND BLUE CELL TUMOR CONSISTENT WITH METASTATIC EWING'S SARCOMA/PNET

[REDACTED]

## MICROSCOPIC DESCRIPTION

Microscopic examination was performed on H&E stained sections and additional sections stained with synaptophysin, Ki-67, GFAP, MIC2 and O13.

Microscopic sections demonstrate a round blue cell tumor with hemorrhage. The tumor is composed of cells with fairly high nuclear/cytoplasmic ratio, strongly and diffusely positive for MIC2 and O13. The tumor cells are negative for GFAP and synaptophysin. The proliferative activity is around 40%. These features are c/w metastatic tumor.

[REDACTED]

**TISSUES:**TUMOR - Two very flat bloody tissue in OCT media

**PROCEDURES:** CYTOGEN FISH/2, FISH 100-300 CE, CYTOGEN INTERP, GENOME WIDE ARR

**PATIENT HISTORY**

INDICATION FOR TEST: EWING'S SARCOMA

**FISH TESTING**

PROBE	CELLS COUNTED
22q12 EWS	100

**RESULTS**

loc ish(EWSx2) (5'EWS sep 3'EWS) [99/100]

The probes used in this study have been developed and/or validated for FISH analysis by the laboratory. The probes have not yet been approved by the FDA for clinical diagnostic testing.

**INTERPRETATION**

Interphase FISH studies were performed using a probe set for the Ewing sarcoma locus in chromosome band 22q12. The results were abnormal, consistent with a translocation that interrupts this locus.

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