Patient ID: PO_3043

MI-ONCOSEQ INTEGRATIVE SEQUENCING EXECUTIVE REPORT*

PATIENT INFORMATION

Name:	[redacted]	**Age/Gender**	4 y/o	**DOB:**	[redacted]
Ethnic background:		**Treating physician:**	Mody	**MI-ONCOSEQ ID:**	PO_3043
Medical Record #:		**Client:**		**Client #:**	
Submitted Diagnosis:	Infantile Fibrosarcoma-diagnosed at birth	**Submitted primary tumor:**	Soft tissue, Left hand and wrist	**Submitting Pathologist:**	
Stage at time of collection:		**Submitted Specimen Site:**		**Specimen collection date:**	09-10-2013
Specimen Type(s):		**Date received:**		**Collection Method:**	Surgical resection
Precision Tumor Board Mtg Date:	11-01-2013	**Report Date:**			
Relevant history:	Patient is adopted; there is limited to no information available about biological relatives				

POTENTIALLY ACTIONABLE/INFORMATIVE RESULTS

Mutation class	Gene/Aberration	Potential Therapies/Clinical Trials
Copy number variation	Chr3q copy loss Chr16 copy gain *CDKN2A/2B homozygous deletion	*CDK inhibitors
Somatic point mutations (6 total)	Detected, unknown significance	
Insertions/deletions (indels)	N/A	
Gene fusions	LMNA-NTRK1	
Outlier expression	Both LMNA and NTRK1 are expressed	
Germline variants	N/A	
Pathogens	N/A	

PRECISION TUMOR BOARD DISCUSSION/INTERPRETATION

- *Aberrations that may relate to standard of care:* N/A
- *Aberrations that may make patient eligible for an open clinical trial or other therapies:*
  In the context of CDKN2A/B homozygous deletion, a CDK inhibitor could be considered.
- *Germline mutations/family history- implications for disclosure:* N/A
- *Other informative results:*
  - LMNA and NTRK1 are 720 kb apart on chr1, and the fusion could be produced by a deletion of less than 1 Mb. This would not be detectable cytogenetically. The LMNA-NTRK1 fusion protein encodes a coiled-coil dimerization domain of LMNA fused to the tyrosine kinase domain of NTRK1.
  - Other NTRK family fusions have been identified in other cancers as well as congenital fibrosarcoma (*Nat Genet*. 1998 Feb;18(2):184-7).
  - There are several TRK family tyrosine kinase inhibitors are in development:
    - Lastaurtanib, CEP-170 (Cephalon)- also targets FLT3 (*Clin Cancer Res*. 2010 Mar 1;16(5):1478-85);
    - AZ-64, AZ-23 (Astra Zeneca) (*Cancer Chemother Pharmacol*. 2012 Sep;70(3):477-86 and *Mol Cancer Ther.* 2009 Jul;8(7):1818-27);
    - AR523 (Array Biopharma).

MI-ONCOSEQ (Michigan Oncology Sequencing) project is a clinical tumor sequencing study that focuses on patients with cancers to 1) provide clinically significant genomic sequencing data to patients and their doctors, and 2) expand the molecular taxonomy of cancers. Integrative sequencing results are derived from whole exome sequencing of tumor and normal samples and paired-end transcriptome sequencing of tumor samples.

***A detailed research report on all of the molecular alterations identified in a patient's tumor is available upon request**

Note: Clinical sequencing is carried out in a CLIA-certified lab for patients enrolled on IRB approved protocols (HUM00046018; HUM00067928). This test is not FDA approved.

Michigan Center for Translational Pathology

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