PathGroup
Physician Centered. Patient Focused.

SmartGenomics™ Sarcoma Profile

Oncology Services

Clinical to Genomic

Advanced Standard of Care

PathGroup SmartGenomics: Sarcoma is designed for ***use at diagnosis*** of primary sarcomas to uncover therapeutic options and aid in treatment planning to improve patient outcomes.

- **Facilitates selection of appropriate clinical trials as recommended by NCCN guidelines**
- Clinically actionable genomic information for 43 gene mutations, 4 cytogenomic abnormalities (FISH), and whole genome copy number changes (CMA) from a single biopsy
- Fully integrated testing on every case for a complete patient picture

Tailored Genomic Sarcoma Profile

Next Generation Sequencing (NGS)	
AKT1	Promotes cell growth and survival, target of inhibition
APC	Mutations may promote cell proliferation, documented in synovial sarcoma
ARAF	Clinically acquired resistance to RAF therapy
ATM	Loss of function mutations lead to genomic instability
BRAF	Therapeutic responses reported in langerhans cell sarcoma under treatment with dabrafenib
BRCA1/2	Potential therapeutic target
CDKN2A	Recurrent alterations seen in follicular dendritic cell sarcoma
CTNNB1	Potentially targetable mutation seen in synovial sarcoma
EGFR	Associated with acquired resistance to BRAF inhibitors, rare in primary sarcomas
ERBB2/4	Promotes cell growth and survival, targetable
FBXW7	A critical tumor suppressor often mutated and inactivated in soft-tissue sarcoma
FGFR1/2/3/4	Mutation occurs in many sarcoma subtypes, targetable
IDH1/2	Seen in more than 50% of patients with chondrosarcomas, potential target
JAK1/2/3	Driver of myeloid sarcoma, targetable
KDR	Activating mutations in angiosarcomas are sensitive to specific kinase inhibitors
KIT	Responses seen in various sarcomas with TKI therapy
KRAS	Implicated in soft tissue sarcoma formation
MET	Potentially targetable mutation
NF1/2	Mutations are present in many types of sarcoma, prognostic value
NOTCH1	Mutation can lead to uncontrolled cell growth, potentially targetable
NRAS	Implicated in soft tissue sarcoma formation
NTRK1/2/3	Potential therapeutic target
PDGFRA	Implicated in sarcoma formation, targetable
PIK3CA	Targetable mutation seen in 18% of sarcomas
PTEN	Rare in sarcoma, prognostic value in combination with TP53
SMAD4	Mutation can lead to uncontrolled cell growth
SMO	A proto-oncogene thought to be an early genetic factor in its tumourigenesis
SRC	Potentially targetable mutation
STK11	Reported in ewing sarcoma, may have prognostic value
TERT	Reported in soft-tissue sarcomas, associated with younger age at presentation and high histological grade
TP53	Predictive of response to VEGF inhibition in soft-tissue sarcoma
VHL	Known tumor suppressor gene

(profile components listing continued on back)

Fluorescence In Situ Hybridization (FISH)	
DDIT3 (CHOP)	Aides in subclassification of various sarcoma types
EWSR1	Diagnostic of Ewing sarcoma
FOX01	Diagnostic of aveolar rhabdomyosarcoma
SS18	Diagnostic of synovial sarcoma

Cytogenomic Microarray (CMA)
Whole genome copy number changes in >22,000 genes, 500 of which are implicated in cancer