MEDGENOME | Prima Cancer Genetics

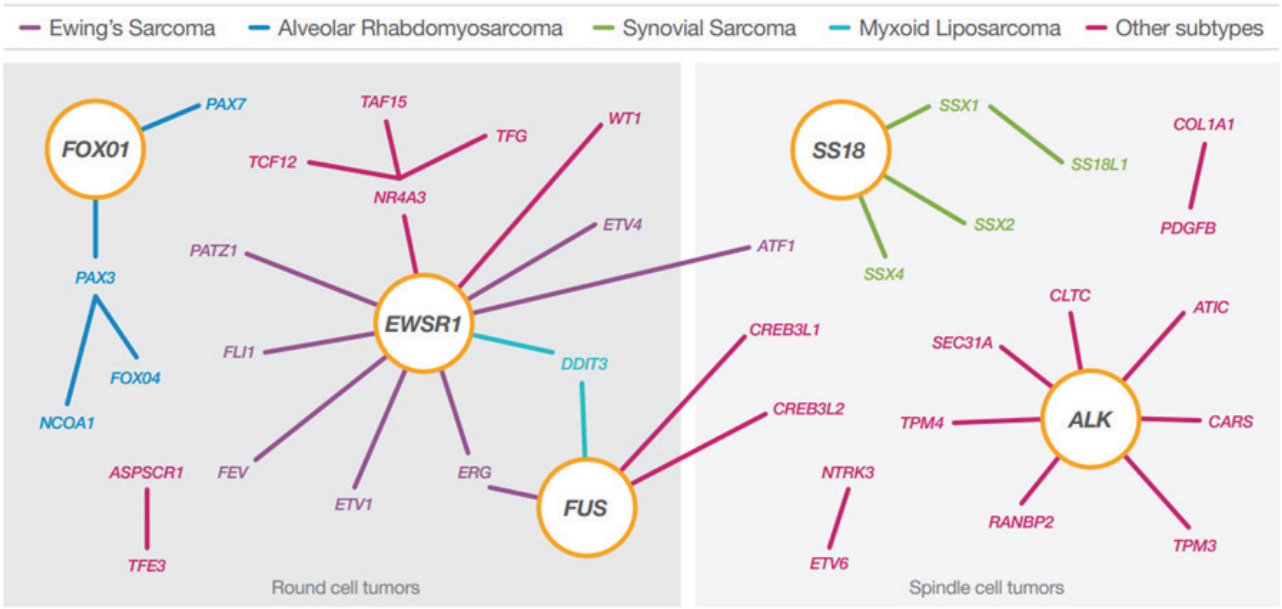
CAP ACCREDITED
COLLEGE of AMERICAN PATHOLOGISTS

# SOFT TISSUE SARCOMA PANEL

Next Generation Sequencing (NGS) based genomic profiling.

**Single test, Multiple fusions:** Reproducible and economical solution to identify fusions in Sarcomas using RNA sequencing.

## Common Fusion in Soft Tissue Sarcoma




## MedGenome Soft Tissue Sarcoma Panel

Genomic alterations (SNVs, InDels and Fusions) in all NCCN guideline recommended soft tissue sarcoma cancer related genes are screened using NGS. A total of 109 genes are screened out of which 83 are analysed for fusions through RNA sequencing.

- Provides accurate diagnosis, prognosis and therapy selection
- Known and Unknown fusion partners can be identified through this assay
- Identifies actionable/potentially actionable biomarkers
- Result interpretation and treatment recommendations are based on AMP-ASCO-CAP guidelines
- Comprehensive coverage of complete coding regions of all the genes and intron/exon boundaries
- Covers tumour agnostic biomarkers such *BRAF*, *RET*, *NTRK1*, *NTRK2* and *NTRK3* which have approved targeted therapies across all metastatic solid tumours.

Laboratory developed assay as per CAP guidelines

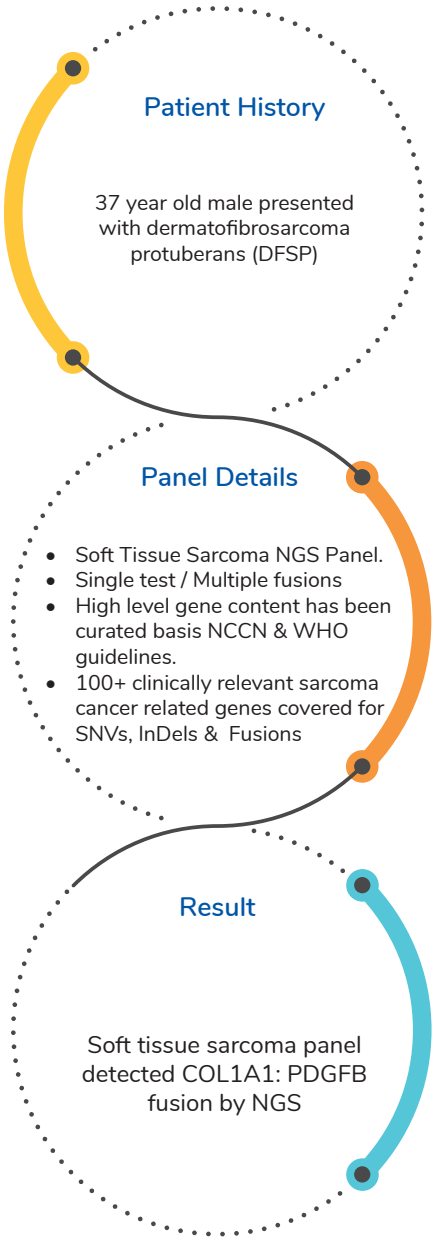
Sensitivity >98% and Specificity with 100%

Limit of detection: 5% VAF for SNV and InDels, >10 Spanning Reads for Fusions.

## Test Details

| Test Code | Test Name | Specimen Type | Methodology | TAT |
|---|---|---|---|---|
| MGM2515 | Soft Tissue Sarcoma Panel | FFPE Tissue Block | NGS | 14 Working days |

## Case Study




DFSP patients with COL1A1: PDGFB fusion can be benefitted using FDA approved targeted therapy (Imatinib mesylate)

https://www.fda.gov/ | PMID: 30085879 & 30601909

## Gene Coverage

| SNV's and InDels | | | | |
|---|---|---|---|---|
| APC | BCOR | BRAF | CDKN2A | CTNNB1 |
| EGFR | ERBB2 | ESR1 | FBXW7 | FGFR1 |
| FGFR2 | FGFR3 | FGFR4 | FLT4 | GNA11 |
| GNA14 | GNAQ | HRAS | IDH1 | IDH2 |
| KDR | KIT | KRAS | MDM2 | MET |
| MYCN | MYOD1 | NF1 | NRAS | PDGFRA |
| PIK3CA | PLCG1 | PTPBR | RET | SDHA |
| SDHB | SDHC | SMARCB1 | TP53 | |

| Fusions | | | | |
|---|---|---|---|---|
| ALK | ASPL | BCOR | BRAF | CDK4 |
| CIC | COL1A1 | CSF1 | DNAJB1 | EGFR |
| EPC1 | ERBB2 | ERG | ESR1 | ETV1 |
| ETV4 | ETV5 | ETV6 | EWSR1 | FGFR1 |
| FGFR2 | FGFR3 | FGFR4 | FLT4 | FOS |
| FOSB | FOXO1 | FUS | GLI | GRB7 |
| HEY1 | HMGA2 | INI1 | JAK3 | JAZF1 |
| MAML2 | MEAF6 | MET | MGEA5 | MKL2 |
| NAB2 | NCOA1 | NCOA2 | NCOA3 | NTRK1 |
| NTRK2 | NTRK3 | NUTM1 | OGA | PAX3 |
| PAX7 | PDGFB | PHF1 | PIK3CA | PLAG1 |
| PRKACA | PRKCA | PRKCB | PRKCD | RAF1 |
| RET | ROS1 | RSPO2 | RSPO3 | SS18 |
| SSX1 | SSX2 | STAT6 | SUZ12 | TAF15 |
| TAF2N | TCF12 | TFE3 | TFEB | TFG |
| TPM3 | USP6 | VGLL2 | VGLL3 | WWTR1 |
| YAP1 | YWHAE | ZC3H7B | | |


**Talk to the Experts:**

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