

Molecular Pathology

The molecular pathology service offers ancillary diagnostic tests for bone and soft tissue tumours on paraffin-embedded material and frozen (listed below). Tests are also provided for research and clinical trials.

List of tests provided:

ASPSCR1-TFE3 (RT-PCR)
BCOR-CCND3 (RT-PCR)
CIC gene rearrangement (FISH)
C-MYC gene amplification (FISH)
COL1A1-PDGFB fusion (FISH)
CTNNB1 (?? Catenin) - Specific mutation analysis: Thr41Ala, Ser45Pro, Ser45Phe
DDIT3 (CHOP) gene rearrangement (FISH)
EWSR1 gene rearrangement (FISH)
EWSR1-ATF1 type 1 EWSR1-ATF1 type 2, EWSR1-ATF1 type 3 (RT-PCR)
EWSR1-ATF1 (RT-PCR)
EWSR1-NR4A3 type 1, EWSR1-NR4A3 type 2, EWSR1-NR4A3 type 3 (RT-PCR)
EWSR1-CREB1 (RT-PCR)
EWSR1-ERG (RT-PCR)
EWSR1-FLI1 type1 / EWSR1-FLI1 type 2 (RT-PCR)
EWSR1-WT1 (RT-PCR)
FOXO1 gene rearrangement (FISH)
FUS gene rearrangement (FISH)
FUS-CREB3L2 (RT-PCR)
FUS-DDIT3 (RT-PCR)
GNAS1 - specific mutation analysis: Arg201Cys, Arg201His, Gln227Leu
HEY1-NCOA2 fusion gene (RT-PCR)
MDM2 gene amplification (FISH)
NR4A3 gene rearrangement (FISH)
PAX3-FOXO1 (FISH and RT-PCR)
PAX7-FOXO1 (FISH and RT-PCR)
SS18 gene rearrangement (FISH)
SS18-SSX1 and SS18-SSX2 (RT-PCR)
TAF2N-NR4A3 (RT-PCR)
USP6 gene rearrangement (FISH)

USP6-MYH9 (RT-PCR)
WT1 gene rearrangement (FISH)
WWTR1-CAMTA1 (RT-PCR)

List of tests provided by diagnosis:

Alveolar soft part sarcoma: ASPSCR1-TFE3 (RT-PCR)

Alveolar rhabdomyosarcoma: *FOXO1* gene rearrangement (FISH), PAX3-FOXO1, PAX7-FOXO1 (FISH and RT-PCR)

Aneurysmal bone cyst: *USP6* gene rearrangement (FISH)

Angiomatoid fibrous histiocytoma: *EWSR1* gene rearrangement (FISH), EWSR1-CREB1 and EWSR1-ATF1 (RT-PCR)

Angiosarcoma, radiation-induced: *C-MYC* gene amplification (FISH)

Atypical lipomatous tumour/well differentiated liposarcoma/dedifferentiated liposarcoma: *MDM2* gene amplification (FISH)

CIC-DUX4 fusion-positive round-cell sarcomas: *CIC* gene rearrangement (FISH)

Clear cell sarcoma: *EWSR1* gene rearrangement (FISH), EWSR1-ATF1 type 1 EWSR1-ATF1 type 2, EWSR1-ATF1 type 3, and EWSR1-CREB1 (RT-PCR)

Dermatofibrosarcoma protuberans: COL1A1-PDGFB fusion (FISH)

Desmoid-type fibromatosis: *CTNNB1* (?? *Catenin*) - substitutions, Specific mutation analysis: Thr41Ala, Ser45Pro, Ser45Phe.

Desmoplastic small round cell tumour: *EWSR1* gene rearrangement (FISH), EWSR1-WT1 (RT-PCR)

Epithelioid hemangioendothelioma: WWTR1-CAMTA1 (RT-PCR)

Extraskelatal myxoid chondrosarcomas: *EWSR1* gene rearrangement (FISH), *NR4A3* gene rearrangement (FISH), EWSR1-NR4A3 type 1, EWSR1-NR4A3 type 2, EWSR1-NR4A3 type 3, TAF2N-NR4A3 (RT-PCR)

Ewing sarcoma: *EWSR1* gene rearrangement (FISH), EWSR1-FLI1 type1, EWSR1-FLI1 type 2, EWSR1-ERG and rare variants (RT-PCR)

Fibrous dysplasia: *GNAS1* - substitutions. Specific mutation analysis: Arg201Cys, Arg201His, Gln227Leu

Intramuscular myxoma: *GNAS1* - substitutions. Specific mutation analysis: Arg201Cys, Arg201His, Gln227Leu

Low-grade fibromyxoid sarcoma: *FUS* gene rearrangement (FISH), FUS-CREB3L2 (RT-PCR)

Mesenchymal chondrosarcoma: HEY1-NCOA2 (RT-PCR)

Myxoid liposarcoma: *FUS* gene rearrangement (FISH), *DDIT3* (CHOP) gene rearrangement (FISH), FUS-DDIT3 (RT-PCR)

Nodular fasciitis: *USP6* gene rearrangement (FISH), USP6-MYH9 (RT-PCR)

Paraosteal osteosarcoma: *MDM2* gene amplification (FISH)

Small round cell sarcomas lacking *EWSR1* gene rearrangement: BCOR-CCND3 (RT-PCR)

Synovial sarcoma: *SS18* gene rearrangement (FISH), SS18-SSX1 SS18-SSX2 (RT-PCR)

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