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| Reporting requirements for cancers or precursor biomarkers |
| **Reportable Biomarkers**  This document comes into effect on 18 September 2025.  This document identifies the biomarkers that are required to be reported to the Secretary of the Department of Health for the purposes of the *Improving Cancer Outcomes (Diagnosis Reporting) Regulations 2025* (the Regulations).  Schedule 1 to this document sets out the cancer or cancer precursor biomarkers that Victorian pathology laboratories are required to report to the Secretary of the Department of Health in accordance with clause 8 of Schedule 3 of the Regulations. |

# Schedule 1

## List of Biomarkers

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| **Unit Name (Biomarkers)** | **Description** |
| **A 20 deletion** | A20 deletion refers to the loss or inactivation of the TNFAIP3 gene, which encodes the A20 protein, a key negative regulator of the NF-κB signalling pathway. This deletion is frequently observed in various B-cell lymphomas and other cancers |
| **AE1/AE3** | AE1/AE3 is an antibody cocktail used in immunohistochemistry (IHC) to detect a broad range of cytokeratins, which are structural proteins found in epithelial cells. It helps to distinguish carcinomas from non-epithelial tumours. |
| **AFP** | AFP (Alpha-Fetoprotein) is a tumour marker primarily used to help diagnose and monitor liver cancer (especially hepatocellular carcinoma) and germ cell tumours of the testes or ovaries |
| **ALK** | ALK (Anaplastic Lymphoma Kinase) is a biomarker and oncogene that plays a key role in the development of certain cancers, especially non-small cell lung cancer (NSCLC) |
| **Androgen Receptor** | The androgen receptor (AR) is a nuclear hormone receptor that binds androgens (like testosterone), and functions as a biomarker and therapeutic target in cancers such as prostate cancer and certain subtypes of breast cancer |
| **APC** | APC (Adenomatous Polyposis Coli) is a tumour suppressor biomarker that plays a crucial role in regulating cell growth, adhesion, migration, and apoptosis, primarily by antagonizing the Wnt/β-catenin signalling pathway. It is used as biomarkers for early detection and prognosis in cancers like colorectal, prostate, and gastric cancer |
| **ATM** | ATM (Ataxia Telangiectasia Mutated) is a tumour suppressor biomarker and a key regulator of the DNA damage response. It encodes a protein kinase that activates repair pathways when DNA double-strand breaks occur. It is used to guide treatment decisions, especially in cancers like breast, ovarian, and pancreatic cancer, where targeted therapies may be effective. |
| **B72.3** | B72.3 is a monoclonal antibody that targets tumour-associated glycoprotein 72 (TAG-72), a mucin-like complex found on the surface of many cancer cells. It helps to distinguish lung adenocarcinoma from mesothelioma. |
| **BARD1** | ARD1 (BRCA1-Associated RING Domain 1) is a dual-function biomarker that can act as both a tumour suppressor and an oncogene, depending on its isoform and mutation status. |
| **B-cell Ig gene rearrangement** | Used to detect clonal B-cell malignancies, typically expressed as monoclonal immunoglobulin chains via molecular assays. |
| **BCL2** | BCL1, also known as Cyclin D1, is a biomarker and oncogene involved in cell cycle regulation, particularly the transition from the G1 to S phase, and is frequently used in the diagnosis of mantle cell lymphoma and other cancers. |
| **BCL-2** | Overexpressed in lymphomas, chronic lymphocytic leukemia, and some breast cancers, associated with resistance to apoptosis. It is reported via IHC as positive or negative. |
| **BCL6** | BCL6 (B-cell lymphoma 6) is a proto-oncogene and transcriptional repressor that plays a central role in germinal center B-cell development and is widely used as a biomarker in hematologic malignancies, especially diffuse large B-cell lymphoma (DLBCL). |
| **BCR-ABL** | BCR-ABL is a fusion gene and oncogenic biomarker formed by a reciprocal translocation between chromosomes 9 and 22, resulting in the Philadelphia chromosome, which is a hallmark of chronic myeloid leukemia (CML) and also found in some cases of acute lymphoblastic leukemia (ALL). |
| **BerEp4** | BerEp4 is a monoclonal antibody biomarker used in immunohistochemistry (IHC) to detect epithelial cell adhesion molecule (EpCAM), a protein found on the surface of most epithelial cells. It is used to distinguish carcinoma from mesothelioma. |
| **b-hCG** | β-hCG (beta-human chorionic gonadotropin) is a tumour marker used in oncology to help diagnose, monitor, and assess treatment response in certain cancers, particularly germ cell tumours of the testes and ovaries, and sometimes trophoblastic tumours. |
| **BRAF** | BRAF is a proto-oncogene and biomarker that encodes a protein kinase involved in the MAPK/ERK signalling pathway, which regulates cell growth, proliferation, and survival. |
| **BRCA** | BRCA biomarkers refer to the BRCA1 and BRCA2 genes, which are tumour suppressor genes involved in DNA repair, particularly through the homologous recombination repair pathway. Identifies individuals at high risk for hereditary cancers such as breast and ovarian cancer. |
| **BRIP1** | BRIP1 (BRCA1-interacting protein C-terminal helicase 1) is a tumour suppressor biomarker involved in the DNA damage repair pathway, particularly in the repair of double-strand breaks, and is associated with increased risk for certain hereditary cancers, such as breast and ovarian cancer. |
| **CA-125** | A high-molecular-weight glycoprotein and tumour biomarker primarily used in the diagnosis, monitoring, and follow-up of ovarian cancer |
| **CA19.9** | Used to help diagnose, monitor, and assess treatment response in pancreatic cancer, and also in other gastrointestinal malignancies such as colorectal, gastric, and biliary tract cancers |
| **CA9** | CA9 (Carbonic Anhydrase IX) is a transmembrane enzyme and tumour biomarker that plays a key role in cellular pH regulation, especially under hypoxic (low oxygen) conditions commonly found in solid tumours. It is highly expressed in renal cell carcinoma. |
| **Calcitonin** | Calcitonin is a polypeptide hormone and tumour biomarker produced by the parafollicular C cells of the thyroid gland, primarily used in the diagnosis and monitoring of medullary thyroid carcinoma (MTC) |
| **CAM5.2** | CAM5.2 is a monoclonal antibody biomarker used in immunohistochemistry to detect low molecular weight cytokeratins, specifically cytokeratin 7 (KRT7) and cytokeratin 8 (KRT8), which are commonly expressed in simple and glandular epithelial tissues. |
| **CCND1** | CCND1 or Cyclin D1 is used to identify and characterize cancers with dysregulated cell cycle control—especially mantle cell lymphoma, breast, prostate, colon, and multiple myeloma—and is typically expressed as gene amplification or protein overexpression detected via IHC or FISH assays |
| **CD10** | CD10 is a cell surface enzyme and biomarker, also known as neprilysin or CALLA (Common Acute Lymphoblastic Leukemia Antigen), that is used in pathology to identify certain hematologic malignancies and solid tumours. It helps to classify lymphomas and leukaemias. |
| **CD103** | CD103 is a transmembrane surface receptor and immune biomarker, also known as integrin αEβ7, that plays a key role in immune cell adhesion, migration, and tissue retention, particularly in epithelial tissues. |
| **CD11c** | CD11c is a well-established biomarker primarily used to identify dendritic cells (DCs), which are key players in the immune system. It is used in cancer research. |
| **CD123** | CD123 is a key biomarker and therapeutic target in immunology and hematologic research. It is highly expressed in acute myeloid leukaemia, hairy cell leukaemia, acute lymphoblastic leukaemia. |
| **CD13** | CD13, also known as aminopeptidase N, is a multifunctional cell surface enzyme and biomarker involved in a wide range of physiological and pathological processes. It appears to be a cause of some types of leukaemias and lymphoma. |
| **CD138** | CD138, also known as syndecan-1, is a transmembrane heparan sulfate proteoglycan that serves as a key biomarker for plasma cells and has important roles in both normal immune function and cancer biology. It is highly expressed in plasma cell neoplasms. |
| **CD15** | CD15, also known as Lewis X (Le^x) or 3-fucosyl-N-acetyl-lactosamine, is a carbohydrate antigen and cell surface adhesion molecule used as a biomarker in immunology and oncology.it is a key marker in classical Hodgkin lymphoma. |
| **CD16** | CD16, also known as Fc gamma receptor III (FcγRIII), is a critical immune cell surface receptor involved in recognizing and responding to antibody-coated targets. It plays a central role in innate immunity, particularly in antibody-dependent cellular cytotoxicity (ADCC) and phagocytosis. It is targeted in cancer immunotherapy. |
| **CD19** | CD19 is a well-characterized cell surface biomarker primarily associated with B cells, playing a central role in B cell development, signalling, and immune regulation. It is used to diagnose B cell lymphomas and leukaemias. |
| **CD1a** | CD1a is a transmembrane glycoprotein and a member of the CD1 family of antigen-presenting molecules. It plays a crucial role in the immune system, particularly in presenting lipid and glycolipid antigens to T cells. It is used to diagnose Langerhans cell histiocytosis. |
| **CD2** | CD2 is a cell surface adhesion molecule and immune biomarker primarily expressed on T cells and natural killer (NK) cells. It plays a key role in immune cell activation, adhesion, and signalling is used to diagnose T-cell lymphomas and leukaemias. |
| **CD20** | CD20 is a well-established cell surface biomarker found primarily on B cells, and it plays a crucial role in immune regulation, diagnostics, and targeted therapies, especially in hematologic malignancies. It is a diagnostic marker for diffuse large B-cell lymphoma, chronic lymphocytic leukaemia, hairy cell leukaemia. |
| **CD21** | CD21, also known as complement receptor 2 (CR2), is a cell surface glycoprotein that plays a key role in B cell activation, immune complex processing, and viral entry, particularly for Epstein-Barr virus (EBV).It is used to diagnose follicular dendritic cell sarcomas. |
| **CD22** | CD22, also known as cluster of differentiation 22, is a transmembrane glycoprotein and a member of the Siglec (sialic acid-binding immunoglobulin-like lectin) family. It plays a vital role in regulating B cell function, particularly by acting as an inhibitory receptor. It is a therapeutic target in B cell malignancies. |
| **CD23** | CD23, also known as Fc epsilon receptor II (FcεRII), is a low-affinity receptor for immunoglobulin E (IgE) and serves as an important biomarker in immunology and hematopathology. It helps to distinguish chronic lymphocytic leukaemia from mantle cell lymphoma. |
| **CD25** | CD25, also known as Interleukin-2 receptor alpha chain (IL-2Rα), is a cell surface glycoprotein that plays a central role in immune regulation, particularly in T cell activation and immune tolerance. It is a diagnostic marker for Adult T-cell leukaemia, hairy cell leukaemia, cutaneous T-cell lymphoma. |
| **CD279** | CD279, also known as Programmed Cell Death Protein 1 (PD-1), is a critical immune checkpoint receptor expressed on T cells, B cells, and other immune cells. It plays a central role in regulating immune responses, particularly by suppressing T cell activity to maintain self-tolerance and prevent autoimmunity. It is a biomarker for immunotherapy response. |
| **CD3** | CD3 is a protein complex and T cell co-receptor that plays a central role in T cell activation and immune signalling. It is a critical biomarker in immunology, clinical diagnostics, and therapeutic development. It helps to diagnose T-cell lymphomas. |
| **CD30** | CD30, also known as TNFRSF8 (Tumour Necrosis Factor Receptor Superfamily Member 8), is a transmembrane glycoprotein receptor that plays a significant role in immune regulation, cell signalling, and oncology, particularly in lymphoid malignancies. It is a diagnostic marker for Hodgkin lymphoma, anaplastic large cell lymphoma, embryonal carcinoma. |
| **CD31** | CD31, also known as Platelet Endothelial Cell Adhesion Molecule-1 (PECAM-1), is a transmembrane glycoprotein and a key biomarker involved in vascular biology, immune cell trafficking, and inflammation.it is used to identify vascular tumours. |
| **CD33** | CD33, also known as Siglec-3, is a transmembrane glycoprotein and a member of the sialic acid-binding immunoglobulin-like lectins (Siglecs) family. It serves as a key biomarker for cells of the myeloid lineage and plays an important role in immune regulation, cancer diagnostics, and therapeutic targeting. It is used to diagnose acute myeloid leukaemia. |
| **CD4** | CD4 is a well-known cell surface glycoprotein and a critical biomarker used to identify and characterize helper T cells in immunology, diagnostics, and clinical medicine. It is used to classify lymphomas, leukaemias. |
| **CD43** | CD43, also known as leukosialin or sialophorin, is a transmembrane sialoglycoprotein that functions as a pan-leukocyte marker and plays diverse roles in immune cell adhesion, migration, and signalling is used to diagnose T-cell lymphomas. |
| **CD45** | CD45, also known as protein tyrosine phosphatase receptor type C (PTPRC) or leukocyte common antigen, is a transmembrane glycoprotein and a key biomarker for immune cells, especially in hematology and immunopathology. It is used to diagnose lymphomas, leukaemias, myeloid sarcoma. |
| **CD5** | CD5 is a transmembrane glycoprotein and a key immune biomarker primarily expressed on T cells and a subset of B cells, with important roles in immune regulation, tolerance, and disease diagnostics. It is used to diagnose T cell lymphomas and leukaemias. |
| **CD52** | CD52, also known as the CAMPATH-1 antigen, is a small, heavily glycosylated cell surface glycoprotein that serves as a biomarker for mature immune cells and a therapeutic target in certain hematologic malignancies and autoimmune conditions. It is used to diagnose chronic lymphocytic leukaemia. B-cell acute lymphoblastic leukaemia, and some T-cell leukaemias. |
| **CD56** | CD56, also known as neural cell adhesion molecule 1 (NCAM1), is a transmembrane glycoprotein and a widely used biomarker in both immunology and oncology. It is a prognostic marker for multiple myeloma. |
| **CD68** | CD68 is a transmembrane glycoprotein and a widely used biomarker for cells of the monocyte/macrophage lineage, including tissue-resident macrophages, circulating monocytes, and specialized phagocytes like microglia and Kupffer cells. It helps to distinguish lymphomas from leukaemias. |
| **CD7** | CD67, also known as CD66b or CEACAM8, is a glycosylphosphatidylinositol (GPI)-anchored glycoprotein and a member of the carcinoembryonic antigen (CEA) family, which belongs to the immunoglobulin superfamily. It is primarily recognized as a granulocyte activation marker. It is a diagnostic marker for T-cell lymphoblastic leukaemia and other T-cell neoplasms. |
| **CD79a** | CD79a is a transmembrane protein encoded by the CD79A gene, located on chromosome 19 in humans. It plays a critical role in the immune system, particularly in B cell development and function. It is used to identify B-cell lineage in lymphomas and leukaemias. |
| **CD8** | CD8 is a transmembrane glycoprotein and a key biomarker for cytotoxic T cells, also known as CD8+ T cells. It plays a central role in the immune system by helping these cells recognize and eliminate infected or malignant cells. It is used in cancer immunotherapy. |
| **CDK4** | CDK4 (Cyclin-Dependent Kinase 4) is a serine/threonine kinase that plays a pivotal role in cell cycle regulation, particularly in the transition from the G1 phase to the S phase of the cell cycle. It is used in diagnosis of glioblastoma multiforme, cutaneous melanoma, liposarcoma |
| **CDKN2A** | CDKN2A (Cyclin-Dependent Kinase Inhibitor 2A) is a tumour suppressor gene that encodes two distinct proteins—p16^INK4A and p14^ARF—through alternative splicing and use of different first exons. It is a diagnostic and prognostic marker for melanoma, pancreatic cancer, glioblastoma multiforme, head and neck squamous cell carcinoma. |
| **CDX2** | CDX2 (Caudal Type Homeobox 2) is a homeobox transcription factor and a highly specific biomarker for intestinal epithelial cells, especially in the context of gastrointestinal pathology. It is used to diagnose tumours of intestinal tract. |
| **CEA** | CEA (Carcinoembryonic Antigen) is a glycoprotein biomarker primarily used in oncology to monitor and manage certain types of cancer. Elevated levels of CEA may be seen in colorectal cancer, pancreatic, gastric, lung, breast, and medullary thyroid carcinoma. |
| **CHEK2** | CHEK2 is a tumour suppressor gene involved in DNA damage repair. Mutations may increase susceptibility to breast, prostate, and colorectal cancers. Reported as positive (mutation detected) or negative |
| **Chromogranin A** | A protein secreted by neuroendocrine cells, used as a marker for neuroendocrine tumours including carcinoid, pancreatic, and prostate cancers. Reported as positive or negative. |
| **Chromosome 19q:loss of heterozygosity (LOH)** | Loss of heterozygosity (LOH) at chromosome 19q is a molecular alteration associated with oligodendrogliomas and other CNS tumours. Reported as present or absent. |
| **Chromosome 1p:loss of heterozygosity (LOH)** | Chromosome 1p: Loss of Heterozygosity (LOH) refers to the loss of genetic material from the short arm (p) of chromosome 1, specifically in one of the two copies normally present in a diploid cell. This genetic alteration is a form of chromosomal damage that can impair tumour suppressor functions and contribute to cancer development. It is used for diagnosis of certain brain tumours. |
| **CK20** | Cytokeratin 20 is an intermediate filament protein used to identify colorectal, bladder, and Merkel cell carcinomas. Reported as positive or negative. |
| **CK5/6** | Cytokeratins 5 and 6 are markers for squamous cell carcinomas and mesotheliomas. Reported as positive or negative. |
| **CK7** | Cytokeratin 7 is used to differentiate between carcinomas of lung, breast, and ovary (CK7+) versus gastrointestinal origin (CK7−). Reported as positive or negative. |
| **c-KIT** | c\_kit or CD117 is a receptor tyrosine kinase expressed in gastrointestinal stromal tumours (GISTs), melanomas, and some leukemias. Reported as positive or negative. |
| **cMET** | MET is a proto-oncogene encoding a receptor tyrosine kinase involved in cell growth and metastasis. Overexpression or mutation is seen in various cancers. Reported as positive or negative. |
| **CTNNB** | CTNNB1 (Beta-Catenin) is a protein involved in cell adhesion and Wnt signalling. Mutations are associated with hepatocellular carcinoma and colorectal cancer. Reported as positive or negative. |
| **D2-40** | A monoclonal antibody that binds to podoplanin, used to identify lymphatic endothelium and differentiate mesothelioma from adenocarcinoma. Reported as positive or negative. |
| **Desmin** | An intermediate filament protein used to identify muscle-origin tumours and assess invasion in urothelial carcinoma. Reported as positive or negative. |
| **dMMR - mismatch repair** | Deficiency in mismatch repair proteins is associated with microsatellite instability and colorectal cancer. Reported as present or absent. |
| **DOG1** | A marker for gastrointestinal stromal tumours (GISTs), expressed in interstitial cells of Cajal. Reported as positive or negative. |
| **E-Cadherin** | E-Cadherin is a transmembrane glycoprotein that plays a crucial role in cell-cell adhesion, particularly in epithelial tissues. It is encoded by the CDH1 gene located on chromosome 16q22.1. |
| **EGFR** | Epidermal Growth Factor Receptor is involved in cell proliferation and survival. Mutations or amplifications are common in lung, anal, and head and neck cancers. Reported as amplified, deleted, insertion, mutation, or negative |
| **EMA** | EMA (MUC1) or epithelial membrane antigen, a glycoprotein expressed in most epithelial cells and tumours. Used to identify epithelial origin |
| **Exosomal RNA** | Derived from tumour exosomes and reflects tumour activity in cancers such as glioblastoma, breast, and pancreatic. It is reported as expression levels via PCR or sequencing. |
| **FGFR2 and FGFR3 gene mutations** | FGFR2 and FGFR3 mutations are used to identify targetable oncogenic drivers in cancers such as urothelial carcinoma, endometrial, breast, gastric, and glioblastoma, typically expressed as activating mutations, amplifications, or gene fusions that lead to aberrant FGFR signaling, detectable via next-generation sequencing or FISH |
| **FIP1L1-PDGFR alpha** | A fusion gene found in hematologic malignancies, especially eosinophilic disorders. Reported as detected or not detected. |
| **GFAP** | The GFAP marker refers to Glial Fibrillary Acidic Protein, a type III intermediate filament protein primarily expressed in astrocytes, which are a type of glial cell in the central nervous system (CNS). It is used to diagnose tumours of glial origin. |
| **Granzyme B** | A serine protease involved in apoptosis, expressed in cytotoxic T cells and NK cells. Used in cancer and infection diagnostics. Reported as positive or negative. |
| **HBME-1** | An immunohistochemical marker used to identify mesothelial cells and is frequently expressed in papillary and follicular thyroid carcinomas. It assists in distinguishing malignant from benign thyroid lesions and is typically reported as positive or negative based on staining patterns. |
| **HER 2** | Human Epidermal Growth Factor Receptor 2 testing determines protein overexpression or gene amplification, primarily in breast and gastric cancers. It guides eligibility for HER2-targeted therapies such as trastuzumab. Results are typically reported as positive, negative, equivocal, or pending, depending on the testing method (e.g., IHC, FISH). |
| **HIV Status Codes** | Indicates whether a patient is HIV-positive or negative. HIV status is relevant in cancer registries due to its association with immunosuppression and increased risk of cancers such as Kaposi sarcoma and non-Hodgkin lymphoma. Reported as positive or negative. |
| **HPV** | Human Papilloma Virus testing identifies viral DNA or protein expression, commonly associated with cervical, anal, and oropharyngeal cancers. It is reported as positive or negative. |
| **HRAS** | HRAS mutation testing is used in thyroid, salivary gland, and kidney cancers to identify oncogenic drivers. It supports molecular classification and is reported as mutation detected or not detected. |
| **IDH1** | IDH1 mutations are common in gliomas and other brain tumours and are associated with prognosis and treatment response. Results are reported as positive or negative for mutation. |
| **IDH1/2** | Combined testing for IDH1 and IDH2 mutations is used in gliomas and hematologic malignancies to guide diagnosis and therapy. Reported as positive or wild type. |
| **IDH2** | IDH2 mutation testing is relevant in gliomas and acute myeloid leukemia. It informs prognosis and treatment decisions and is reported as positive or negative. |
| **IRF4** | IRF4 is a transcription factor used as a diagnostic and prognostic biomarker in hematologic malignancies such as multiple myeloma, diffuse large B-cell lymphoma, and other mature lymphoid neoplasms. It is typically overexpressed in malignant lymphoid cells, where it drives oncogenic transcription programs and immune regulation, detectable via immunohistochemistry or gene expression profiling |
| **JAK2** | JAK2 mutation analysis is used to diagnose myeloproliferative neoplasms such as polycythaemia vera and essential thrombocythemia. Reported as positive or negative. |
| **Ki67** | Ki67 is a nuclear protein expressed in proliferating cells and is used to assess tumour aggressiveness, especially in breast and neuroendocrine tumours. Reported as positive, negative, or as a percentage indicating proliferation index. |
| **KIT gene mutation** | The KIT gene mutation marker refers to changes in the KIT gene, which encodes a receptor tyrosine kinase known as CD117 or c-KIT. This protein plays a key role in cell signalling, particularly in hematopoietic stem cells, melanocytes, and gastrointestinal cells. It is associated with Gastrointestinal stromal tumours, acute myeloid leukaemia, melanoma, seminoma, mastocytosis. |
| **KRAS** | KRAS mutation testing is performed in colorectal, pancreatic, and lung cancers to guide prognosis and targeted therapy decisions. Reported as mutation detected or not detected. |
| **LDH** | Lactate dehydrogenase is a serum marker of tissue damage and tumour burden, particularly in lymphomas and germ cell tumours. Reported as elevated or normal. |
| **MCPyV-pve** | The MCPyV-pve marker refers to the presence of Merkel cell polyomavirus (MCPyV) in tumour tissue, particularly in Merkel cell carcinoma (MCC), a rare but aggressive form of skin cancer. |
| **MDM2** | MDM2 expression is associated with sarcomas and other tumours, influencing cell cycle regulation and p53 pathway activity. Reported as positive or negative. |
| **Melan-A** | Melan-A is a melanocytic marker used to confirm melanoma and distinguish it from other poorly differentiated tumours. Reported as positive or negative. |
| **MEN1** | MEN1 mutation testing is used to diagnose multiple endocrine neoplasia type 1, a hereditary cancer syndrome. Reported as positive or negative. |
| **Methylation of MGMT** | MGMT promoter methylation is assessed in glioblastoma to predict response to alkylating agents like temozolomide. Reported as methylated or unmethylated. |
| **MOC-31** | MOC-31 is an epithelial marker used to differentiate adenocarcinoma from mesothelial cells in effusion cytology. Reported as positive or negative. |
| **MUM1** | MUM1 is expressed in plasma cells and certain lymphomas, aiding in subclassification of lymphoid malignancies. Reported as positive or negative. |
| **MUTYH** | Involved in DNA base excision repair, mutations in this gene are linked to colorectal cancer syndromes such as MUTYH-associated polyposis. Typically reported as positive or negative for mutation. |
| **MYC gene expression** | MYC is a transcription factor used as a diagnostic, prognostic, and therapeutic biomarker in a wide range of cancers—including Burkitt lymphoma, leukemia, breast, colorectal, lung, liver, ovarian, and prostate cancers—and is typically expressed as gene amplification, chromosomal translocation, or protein overexpression, driving uncontrolled cell growth, metabolism, and proliferation |
| **MYD88 gene mutation** | MYD88 mutations—especially the L265P variant—are used to diagnose and guide treatment in B-cell malignancies such as Waldenström macroglobulinemia (WM), lymphoplasmacytic lymphoma (LPL), and diffuse large B-cell lymphoma (DLBCL). These mutations are typically expressed as gain-of-function alterations that activate NF-κB and JAK/STAT signaling, promoting tumor cell survival and proliferation |
| **Myoglobin** | An iron- and oxygen-binding protein released from damaged muscle tissue; elevated levels may indicate rhabdomyolysis and potential kidney injury. Recognised as a marker for rhabdomyosarcoma, a cancer of skeletal muscle origin. Its presence was historically associated with muscle differentiation in tumours. Reported as positive or negative. |
| **Napsin-A** | A diagnostic marker for lung adenocarcinoma, used to differentiate primary lung tumours from metastases. Reported as positive or negative |
| **NBN** | Encodes nibrin, a protein involved in DNA damage response; mutations are associated with Nijmegen breakage syndrome and increased cancer risk. Reported as positive or negative. |
| **NRAS** | Mutations are implicated in colorectal, thyroid, and hematologic malignancies; testing aids in molecular classification and therapy selection. Reported as positive or negative. |
| **NTRK gene fusion** | NTRK gene fusions are used to identify targetable oncogenic drivers in a wide range of solid tumors—including thyroid, lung, colon, breast, salivary gland, sarcoma, and pediatric cancers—and are typically expressed as TRK fusion proteins resulting from chromosomal rearrangements, detectable via NGS, IHC, FISH, or PCR. These fusions drive tumor growth and are actionable with tumor-agnostic TRK inhibitors like larotrectinib and entrectinib |
| **Oestrogen (estrogen) receptor** | The estrogen receptor (ER) marker is a protein found inside or on the surface of certain cells, particularly in breast tissue, that binds to the hormone estrogen. It plays a key role in the growth and development of hormone-sensitive tissues and is a critical marker in breast cancer diagnosis and treatment planning. |
| **OLIG2** | A transcription factor used as a diagnostic marker in gliomas and other brain tumours. Reported as positive or negative. |
| **P16** | A tumour suppressor protein: loss or deletion is common in oesophageal and gastric cancers. Reported as positive, negative, or equivocal. |
| **P40** | A marker used to identify squamous cell carcinoma and distinguish it from other epithelial tumours. Reported as positive or negative. |
| **P53** | A key tumour suppressor protein involved in cell cycle regulation; mutations are common across many cancer types. Reported as positive or negative. |
| **P63** | Used to distinguish squamous cell carcinoma from other poorly differentiated tumours. Reported as positive or negative |
| **PALB2** | Mutations in this gene increase the risk of breast and pancreatic cancers; testing supports hereditary cancer risk assessment. Reported as positive or negative. |
| **PAX5** | A transcription factor involved in B-cell development; alterations are seen in lymphoid malignancies. Reported as positive or negative. |
| **PAX8** | Expressed in thyroid and renal epithelial tumours; used to confirm tumour origin. Reported as positive or negative. |
| **PDGFRA** | Mutations are associated with gastrointestinal stromal tumours and hematologic malignancies; testing informs targeted therapy. Reported as positive or negative. |
| **PD-L1** | An immune checkpoint protein: expression is used to predict response to immunotherapy in cancers such as melanoma and lung cancer. Reported as positive or negative. |
| **PI3K** | Mutations are common in breast, ovarian, and colorectal cancers, influencing cell survival and drug resistance. They are reported as mutation detected or not detected via NGS or PCR. |
| **PIK3CA** | Mutations are common in breast and colorectal cancers and guide use of PI3K inhibitors. They are reported as mutation detected or not detected. |
| **PML-RARalpha** | Fusion gene associated with acute promyelocytic leukemia; testing confirms diagnosis and guides treatment. Reported as positive or negative. |
| **PR Status** | Indicates presence of progesterone receptors in tumour cells, relevant for hormone therapy decisions. Reported as positive or negative. |
| **PSA** | Prostate-specific antigen level used to screen for and monitor prostate cancer. Reported as elevated or normal. |
| **PTEN** | Tumour suppressor gene involved in cell cycle regulation; loss is associated with various cancers. Reported as positive or negative. |
| **RAD 51** | Involved in homologous recombination and DNA repair; mutations may contribute to cancer susceptibility. Reported as positive or negative. |
| **RAD50** | The RAD50 marker refers to the RAD50 gene and its protein product, which plays a critical role in DNA repair, genomic stability, and cancer susceptibility. It is used in genetic testing for hereditary cancer syndromes, tumour profiling, and clinical trials. |
| **RAD51C** | Plays a role in DNA repair; mutations are associated with hereditary breast and ovarian cancer. Reported as positive or negative. |
| **RAD51D** | Associated with increased risk of breast and ovarian cancers; testing supports genetic risk assessment. Reported as positive or negative. |
| **RET protooncogene** | Mutations or rearrangements in this gene are associated with medullary thyroid carcinoma and other endocrine tumours. Testing helps confirm diagnosis and guide targeted therapy. Reported as positive or negative. |
| **ROS1** | Gene fusions involving ROS1 are found in lung, ovarian, and other cancers. Detection is critical for identifying patients eligible for ROS1-targeted therapies. Reported as positive or negative. |
| **S-100** | A protein expressed in neural crest-derived cells, used to identify melanomas, schwannomas, and other neural tumours. Reported as positive or negative. |
| **SMA** | Smooth muscle actin is used to identify smooth muscle differentiation in tumours and diagnose autoimmune hepatitis. Reported as positive or negative. |
| **Somastin receptor** | Cell surface proteins used in the diagnosis and treatment of neuroendocrine tumours, including those of the gastrointestinal tract, pancreas, pituitary, and lung; results are expressed through immunohistochemistry or nuclear imaging, indicating receptor presence and density |
| **STK11** | A tumour suppressor gene involved in Peutz-Jeghers syndrome and various cancers. Mutations may influence prognosis and treatment. Reported as positive or negative. |
| **Synaptophysin** | A marker of neuroendocrine differentiation, used to identify tumours such as neuroblastoma, small-cell lung carcinoma, and carcinoid tumours. Reported as positive or negative. |
| **T-cell receptor gene rearrangement (TCR)** | T-cell receptor (TCR) gene rearrangement is a molecular biomarker used to detect clonal T-cell populations by analyzing the unique DNA sequences of rearranged T-cell receptors, primarily in T-cell lymphomas and T-cell leukemias. Results are typically expressed through PCR or sequencing, indicating whether the T-cell population is monoclonal, which suggests malignancy, or polyclonal, which suggests a reactive or benign process. |
| **TdT** | Terminal deoxynucleotidyl transferase is expressed in immature lymphoid cells and used to diagnose acute lymphoblastic leukemia. Reported as positive or negative. |
| **Thyroglobulin** | A protein produced by thyroid follicular cells, used as a tumour marker in differentiated thyroid cancers. Reported as positive or negative. |
| **TIA-1** | An immune marker expressed in cytotoxic lymphocytes, used in the classification of lymphomas and leukemias. Reported as positive or negative. |
| **TMPT** | Thiopurine S-methyltransferase activity influences metabolism of thiopurine drugs used in cancer and autoimmune diseases. Reported as a numeric concentration level. |
| **TP53** | A key tumour suppressor gene frequently mutated in many cancers; testing informs prognosis and potential treatment strategies. Reported as positive or negative. |
| **TTF-1** | Thyroid transcription factor 1 is expressed in lung and thyroid tumours, used to confirm tumour origin. Reported as positive or negative. |
| **UGT1A1** | Mutations affect bilirubin metabolism and drug clearance; relevant in pharmacogenomics and cancer treatment planning. Reported as positive or negative. |
| **VHL** | Mutations in this gene are linked to von Hippel-Lindau syndrome and renal cell carcinoma. Testing supports diagnosis and surveillance. Reported as positive or negative. |
| **Vimentin** | An intermediate filament protein expressed in mesenchymal cells, used to identify sarcomas and assess epithelial-mesenchymal transition. Reported as positive or negative. |
| **WT-1** | A transcription factor expressed in mesotheliomas and ovarian serous carcinomas, used to distinguish these from other tumours. Reported as positive or negative. |

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