

I. Diagnosis Associated With Syndromes by Organ

Breast

Breast Carcinoma, Female

Breast Carcinoma, Male

Breast Table

Blood and Bone Marrow

Acute Lymphoblastic Leukemia and Non-Hodgkin Lymphoma

Acute Myeloid Leukemia and Myelodysplastic Syndrome

Blood and Bone Marrow Table

Bone and Soft Tissue

Chondrosarcoma

Chordoma

Malignant Peripheral Nerve Sheath Tumor

Osteosarcoma

Rhabdomyosarcoma

Schwannoma

Bone and Soft Tissue Table

Head and Neck

Endolymphatic Sac Tumor

Squamous Cell Carcinoma

Head and Neck Table

Salivary Glands Table

Endocrine

Adrenal Cortex

Adrenal Cortical Adenoma

Adrenal Cortical Carcinoma

Adrenal Cortical Neoplasms in Children

Primary Pigmented Nodular Adrenocortical Disease

Adrenal Cortex Table

Adrenal Medulla and Paraganglia

Adrenal Medullary Hyperplasia

Neuroblastoma

Pheochromocytoma and Paraganglioma

Adrenal Medulla and Paraganglia Table

Pancreas

Pancreatic Neuroendocrine Neoplasms

Pancreas Table

Parathyroid

Parathyroid Adenoma

Parathyroid Carcinoma

Parathyroid Hyperplasia

Parathyroid Table

Pituitary

Pituitary Adenoma

Pituitary Hyperplasia

Pituitary Table

Thyroid, Medullary

C-Cell Hyperplasia

Medullary Thyroid Carcinoma

Thyroid, Medullary Table

Thyroid, Nonmedullary

Familial Thyroid Carcinoma

Follicular Thyroid Carcinoma

Thyroid, Nonmedullary Table

Gastrointestinal

Tubular Gut

Colon Adenoma

Esophageal Adenocarcinoma

Esophageal Squamous Cell Carcinoma

Gastric Adenocarcinoma

Gastrointestinal Stromal Tumor

Hamartomatous Polyps of GI Tract

Small Bowel Adenocarcinoma

Colon/Rectum Table

Esophagus/Stomach/Small Bowel Table

Tubular Gut Table

Hepatobiliary and Pancreas

Biliary Tract Neoplasia

Hepatoblastoma

Hepatocellular Carcinoma

Pancreatic Adenocarcinoma

Biliary Tract/Liver/Pancreas Table

Hepatobiliary and Pancreas Table

Genitourinary

Bladder

Bladder Table

Kidney

Angiomyolipoma

Clear Cell Renal Cell Carcinoma

HLRCC Syndrome-Associated Renal Cell Carcinoma

Papillary Renal Cell Carcinoma

Renal Oncocytoma, Chromophobe, and Hybrid Tumors

Wilms Tumor

Kidney Table

Prostate

Prostate Table

Renal Pelvis and Ureter

Renal Pelvis and Ureter Table

Testicle

Germ Cell Tumor

Sertoli Cell Neoplasms

Testicle Table

Gynecology

Cervical Tumors

Fallopian Tube Tumors

Ovarian Tumors

Uterine Tumors

Cervix

Endometrium

Fallopian Tube

Ovary

Nervous System

Central Nervous System

Eye

Peripheral Nervous System

Pulmonary

Adenocarcinoma, Lung

Adenocarcinoma With Lepidic (Bronchioloalveolar) Predominant Pattern

Lymphangioliomyomatosis

Neuroendocrine Carcinoma, Lung

Pleuropulmonary Blastoma

Lung Table

Skin

BAP1-Inactivated Melanocytic Tumor

Basal Cell Carcinoma

Cutaneous Melanoma

Cutaneous Squamous Cell Carcinoma

Sebaceous Carcinoma

Skin Table

II. Overview of Syndromes

Introduction

Pathology of Familial Tumor Syndromes

Clinical Diagnosis and Management of Familial/Hereditary Tumor Syndromes

Molecular Aspects of Familial/Hereditary Tumor Syndromes

Syndromes

Ataxia Telangiectasia

BAP1 Tumor Predisposition Syndrome

Basal Cell Nevus Syndrome/Gorlin Syndrome

Beckwith-Wiedemann Syndrome

Birt-Hogg-Dubé Syndrome

Bloom Syndrome

Brooke-Spiegler Syndrome

Carney Complex

Costello Syndrome

Cystic Nephroma Syndrome

Denys-Drash Syndrome

Diamond-Blackfan Anemia

DICER1 Syndrome

Down Syndrome

Dyskeratosis Congenita

Familial Acute Myeloid Leukemia and Myelodysplastic Syndrome

Familial Adenomatous Polyposis

Familial Chordoma

Familial Gastrointestinal Stromal Tumor

Familial Infantile Myofibromatosis

Familial Isolated Hyperparathyroidism

Familial Medullary Thyroid Carcinoma

Familial Nonmedullary Thyroid Carcinoma

Familial Paraganglioma-Pheochromocytoma Syndrome

Familial Testicular Tumor

Familial Uveal Melanoma

Familial Wilms Tumor

Fanconi Anemia

GATA2 Spectrum Disorders

Glucagon Cell Hyperplasia and Neoplasia

Hereditary Breast/Ovarian Cancer Syndrome: BRCA1

Hereditary Breast/Ovarian Cancer Syndrome: BRCA2

Hereditary Diffuse Gastric Cancer

Hereditary Leiomyomatosis and Renal Cell Carcinoma Syndrome (HLRCC)

Hereditary Multiple Exostosis

Hereditary Neuroblastoma

Hereditary Pancreatic Cancer Syndrome

Hereditary Papillary Renal Cell Carcinoma

Hereditary Paraganglioma/Pheochromocytoma Syndromes

Hereditary Prostate Cancer

Hereditary Renal Epithelial Tumors, Others

Hereditary Retinoblastoma

Hereditary SWI/SNF Complex Deficiency Syndromes

Heritable Gastrointestinal Stromal Tumors Syndromes

Howel-Evans Syndrome/Keratosis Palmares and Plantares With Esophageal Cancer

Hyperparathyroidism-Jaw Tumor Syndrome

Juvenile Polyposis Syndrome

Li-Fraumeni Syndrome/Li-Fraumeni-Like Syndrome

Lynch Syndrome

McCune-Albright Syndrome

Melanoma/Pancreatic Carcinoma Syndrome

Multiple Endocrine Neoplasia Type 1 (MEN1)

Multiple Endocrine Neoplasia Type 2 (MEN2)

Multiple Endocrine Neoplasia Type 4 (MEN4)

MYH-Associated Polyposis

Neurofibromatosis Type 1

Neurofibromatosis Type 2

Nijmegen Breakage Syndrome

Pancreatic Neuroendocrine Tumor Syndromes

**PDGFRA*-Mutant Syndrome*

Peutz-Jeghers Syndrome

PTEN Hamartoma Tumor Syndromes

RASopathies: Noonan Syndrome

Rhabdoid Predisposition Syndrome

Schwannomatosis

Shwachman-Diamond Syndrome

Steatocystoma Multiplex

Succinate Dehydrogenase (SDH)-Deficient Renal Cell Carcinoma

Tuberous Sclerosis Complex

Tumor Syndromes Predisposing to Osteosarcoma

von Hippel-Lindau Syndrome

Werner Syndrome/Progeria

Wilms Tumor-Associated Syndrome

Wiskott-Aldrich Syndrome

Xeroderma Pigmentosum

Reference

Molecular Factors

Molecular Factors Index