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Syndrome Gene(s) Inheritance
implicated Familial adenomatous
APC gene D polyposis (FAP) D DNA
mismatch Hereditary non-
polyposis repair genes colorectal
cancer (MLH1; MSH2; (HNPCC1),

Lynch MSH6) syndrome 1 D
HNPCC2 DNA mismatch repair
genes (MLH1; MSH2; MSH6)
Peutz-Jeghers syndrome STK11 D
a Cowden syndrome PTEN D
Retinoblastoma RB D Multiple
endocrine Menin D neoplasia
(MEN) type 1 MEN type 2A RET D
MEN type 2B RET D Li-Fraumeni
P53 D Familial breast cancer
BRCA1; BRCA2 D Familial
cutaneous CDNK2A; CDK4 D
malignant melanoma Associated
tumours and abnormalities
Strategies for prevention/ early
diagnosis Pr ophylactic Colorectal

cancer under the age of 25
panproctocolectomy Papillary
carcinoma of the thyroid Cancer of
the ampulla of Vater
Hepatoblastomas Primary brain
tumours (Turcot syndrome)
Osteomas of the jaw CHRPE
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retinal pigment epithelium) Color
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oidal anti

in /f_l ammatary drugs HNPCC associated with other cancers of the gastrointestinal or reproductive
system Bowel cancer; br east cancer; freckles round Surveillance colonoscopy; the mouth
mammography Active surveillance Multiple hamartomas of skin, breast and mucous membranes
Breast cancer Neuroendocrine tumours Endometrial cancer Thyroid cancer Surveillance of
uninvolved Retinoblastoma eye Pinealoma Osteosarcoma Awareness of associations Parathyroid
tumours and paying attention to Islet cell tumours relevant symptoms Pituitary tumours Regular
screening of blood Medullary carcinoma of the thyroid pressure, serum calcitonin
Pheochromocytoma and urinary catecholamines Parathyroid tumours Prophylactic thyroidectomy

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 Pheochromocytoma and urinary catecholamines Mucosal neuromas Prophylactic thyroidectomy
 Ganglioneuromas of the gut Very difficult to culture, since pattern of Sarcomas tumours is so
 heterogeneous Leukaemia and varies between patients Osteosarcomas Brain tumours
 Adrenocortical carcinomas Screening mammography; Breast cancer pelvic ultrasound Ovarian
 cancer PSA (in males) Papillary serous carcinoma of the peritoneum Prophylactic mastectomy;
 Prostate cancer prophylactic oophorectomy Cutaneous malignant melanoma Avoid exposure to
 sunlight, careful surveillance Continued

Robert Gorlin , 1923–2006, Professor of Dentistry , The University of Eugen von Hippel , 1867–1939,
 Professor of Ophthalmology , Göttingen, Germany . Arvid Lindau , 1892–1958, Professor of
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 New York University , New York, NY , USA, described the syndrome in 1954. Minnesota,
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Syndrome Gene(s) Inheritance implicated Basal cell naevus PTCH D syndrome (Gorlin) von
 Hippel-Lindau VHL D disease Neurofibromatosis type 1 NF1 D Neurofibromatosis type 2 NF2
 D R Xeroderma pigmentosum De deficient nucleotide excision repair (XPA,B,C)
 Ataxia-telangiectasia AT R Bloom syndrome BLM helicase R D, dominant; PSA, prostate-specific
 antigen; R, recessive. a One of the few clinical syndromes named for the patient rather than the
 clinician. Rachel Cowden was, in 1963, the first patient described with the syndrome. She died
 from breast cancer at the age of 20. TABLE 12.2 Environmental causes of cancer (and suggested
 measures for reducing their impact). Environmental/behavioural factor Tobacco Alcohol Ultraviolet
 exposure Ionising radiation Viral infections Human papillomavirus Associated tumours and
 abnormalities Strategies for prevention/ early diagnosis Careful surveillance, Basal cell carcinomas
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 Primitive neuroectodermal tumours concerning any rapid Optic gliomas changes in growth or
 Multiple neurofibromas character of any nodule Acoustic neuromas Spinal tumours Meningiomas
 Multiple neurofibromas Skin sensitive to sunlight. Early onset of Avoidance of sun exposure
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 Lung cancer Ban smoking in public places Head and neck cancer Punitive taxes on tobacco Bladder
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 radiation Lymphoma workers Thyroid cancer Avoid unprotected sex Cervical cancer Vaccination
 Penile cancer Head and neck cancer Continued

A more comprehensive view considers the management of cancer as taking place along two axes:
 one is an axis of scale, from the individual to the world population; the other is an axis based on
 the development of the disease, from prevention through to rehabilitation or palliative care (Figure

12.3).

Environmental/behavioural factor Viral infections – Human immunodeficiency virus continued
Hepatitis B Other infections Schistosomiasis Helicobacter pylori Inhaled particles Asbestos Wood
dust Chemicals Environmental pollutants/chemicals used in industry Medical Alkylating agents used
in cytotoxic chemotherapy Immunosuppressive treatment Tamoxifen Fungal and plant Anticancer
toxins Obesity/lack of physical exercise

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