

Table of Contents

Chapter 1

History of Cancer Genetics in Man. HENRY T. LYNCH, M.D.	1
Man as a problem for genetic studies	1
Cancer genetics in animals	1
Early studies in man	1
Cancer of specific anatomic sites	2
Twin studies	2
Cancer families	3
Mendelian inherited cancer disorders	4
Cytogenetics and carcinoma	6
Blood group antigens and carcinoma	7
Cancer resistance	8
References	10

Chapter 2

Classical Examples of Heredity and Malignancy. HENRY T. LYNCH, M.D.	18
Introduction	18
Familial polyposis coli	19
Gardner's syndrome	20
Multiple nevoid basal cell carcinoma syndrome	21
Xeroderma pigmentosum	23
Thyroid carcinoma (medullary thyroid carcinoma with amyloid production associated with pheochromocytoma)	23
Hereditary multiple exostoses	25
Hereditary polyendocrine adenomatoses and the Zollinger-Ellison syndrome	26
Tylosis and esophageal cancer (keratosis palmaris et plantaris)	27
Bloom's syndrome (congenital telangiectatic erythema and stunted growth)	28
Fanconi's aplastic anemia (constitutional aplastic anemia)	28
Retinoblastoma	29
Von Recklinghausen's neurofibromatosis	29
References	30

Chapter 3

Heredity and the More Common Malignancies of Man: Carcinoma of the Breast, Stomach, Colon, Lung, Prostate, and Endometrium. HENRY T. LYNCH, M.D.	35
Breast Cancer	35
Stomach Cancer	36
Colon carcinoma	37
Solitary polyps of the rectum and colon	38
Turcot's syndrome	39
Ulcerative colitis and carcinoma of the colon	39
Juvenile polyposis coli	40
Colon carcinoma in the absence of polyps	41
Lung carcinoma	41
Prostate carcinoma	43

Endometrial carcinoma	45
References	47

Chapter 4

Hereditary Aspects of Some Less Frequently Occurring Malignancies Exclusive of the Reticuloendothelial System. HENRY T. LYNCH, M.D.	53
Introduction	53
Kaposi's sarcoma	53
Carcinoma of the nasopharynx	54
Carotid body tumors	54
Generalized keratoacanthoma	55
Hepatocellular carcinoma	56
Familial pheochromocytoma	56
Malignant melanoma	58
Carcinoid tumor	60
Carcinoma of the duodenum	60
Hypernephroma	61
Testicular tumors	61
References	63

Chapter 5

Hereditary Disorders with Variable Malignant Neoplastic Predisposition. HENRY T. LYNCH, M.D.	67
Introduction	67
Werner's syndrome	67
Paget's disease (osteitis deformans) and osteogenic sarcoma	68
Ataxia telangiectasia (Louis-Bar syndrome)	68
Congenital sex-linked agammaglobulinemia (Bruton's type)	69
Chediak-Higashi syndrome	69
Wiskott-Aldrich syndrome	70
Dermatomyositis	70
Sjogren's syndrome	71
Scleroderma (progressive systemic sclerosis)	72
Albinism	72
Dysgenetic gonads and disorders of somatosexual disturbance	72
Peutz-Jeghers syndrome	74
Porphyria cutanea tarda	75
Systemic lupus erythematosus	76
References	78

Chapter 6

Genetic Aspects of Tumors of the Nervous System. JOHN A. AITA, Ph.D., M.D.	86
Introduction	86
Familial isolated tumors of the nervous system	86
Established genetic diseases with significant incidence of tumors of nervous system	87
Von Recklinghausen's neurofibromatosis	87
Neurofibromas	88
Sarcomatous neurofibromas	88
Central and peripheral forms of neurofibromatosis	88
Intracranial tumors	88
Intracranial neurofibromas	88
Meningiomas	89
Glioma of optic nerve	89
Other intracerebral tumors	89
Disseminated gliosis	90
Intraspinal tumors	90

Multiple primary intracranial and intraspinal neoplasms	90
Pheochromocytoma	91
Von Hippel Lindau's hemangioblastoma	91
Anatomic sites of hemangioblastomas	93
Tuberous sclerosis	93
Hereditary multiple endocrine adenomatosis	94
Multiple nevoid basal cell carcinoma syndrome	94
Retinoblastoma	95
Pheochromocytoma	95
Neuroblastoma	95
Summary	96
Bibliography	97

Chapter 7

Hereditary and Malignancies of the Reticuloendothelial System. HENRY T. LYNCH, M.D.	111
Introduction	111
Hodgkin's disease	111
Multiple myeloma	112
Leukemia	113
Early genetic studies	113
Chronic lymphocytic leukemia and Ch ¹ chromosome	114
Chronic myelogenous leukemia and Ph ¹ chromosome	114
Waldenström's macroglobulinemia	115
References	117

Chapter 8

Congenital Cancer and Congenital Abnormalities Associated with Cancer.	
HENRY T. LYNCH, M.D.	120
Introductory survey	120
Neuroblastoma	120
Wilm's tumor	121
Aniridia	121
Hemihypertrophy	121
Adrenal cortical carcinoma	121
Hepatic carcinoma	121
Wilm's tumor	121
Neuroblastoma	121
Klinefelter's and Turner's syndromes	122
Congenital epithelial parotid gland tumors	122
References	123

Chapter 9

"Cancer Families": Adenocarcinomas (Endometrial and Colon Carcinoma) and Multiple Primary Malignant Neoplasms. HENRY T. LYNCH, M.D.	
Introduction and historical review	125
Definition of a "cancer family"	126
Importance of histologic diagnosis	126
Study of six "cancer families"	127
Hereditary and multiple primary malignant neoplasms	128
Cancer families in the literature from Warthin (Family G) to present	130
The hereditary multiple cancer syndrome	131
Implications for this cancer syndrome from the standpoint of early cancer detection and cancer control	132
References	134

Chapter 10

Heredity, Emotions, and Carcinoma: Delay in Cancer Detection. ANNE J. KRUSH, M.S., A.C.S.W.	143
Introduction	143
Tumor service Patient series	144
Two "cancer families"	145
College Alumnae series	146
Analysis of findings	146
Implications for cancer control	147
References	149

Chapter 11

Cancer and Genetic Counseling. HENRY T. LYNCH, M.D.	154
History of genetic counseling	154
Genetic counseling and diagnosis	154
Genetic counseling, physician responsibility, a patient-physician relationship	156
Genetic counseling as "family problems"	156
Clinical examples, emotional and psychiatric factors	157
Protocol for genetic counseling	157
Genetic terminology and disease	158
Phenocopies	158
Penetrance	158
Variable expressivity	158
Genetic counseling and cancer patients	159
Family with xeroderma pigmentosum	162
Cancer family	162
Familial polyposis coli	163
Summary	163
References	167

Chapter 12

Epilogue. HENRY T. LYNCH, M.D.	169
Figures	173