

Contents

1	Introduction	1
1.1	Common Characteristics of Rare Diseases	1
1.2	Glossary of Selected Genetic Terms	2
1.3	Congenital Anomaly of the Genitalia	2
1.4	General Principles Used in This Book for Illustration of the Genitourinary Anomalies	3
	References	3

Part I Male Genital Anomalies

2	Penis	7
2.1	Embryology and Penile Development	7
2.2	Penile Agenesis (PA)	8
2.3	Microphallus	11
2.4	Megalopenis	13
2.5	Penile Duplication (PD)	14
2.6	Penile Deviation	16
2.7	Penile Chordee	17
2.8	Webbed Penis	18
2.9	Penoscrotal Transposition (PST)	20
2.10	Genital Median Raphe Anomalies (GMR)	22
2.11	Penile Lymphedema	24
2.12	Penile Cysts	25
2.13	Megameatus	26
2.14	Congenital Meatal Stenosis	27
2.15	Congenital Double Meatus	29
	References	30
3	Prepuce	33
3.1	Development and Function of the Prepuce	33
3.2	Preputial Function	34
3.3	Aposthia	34
3.4	Microposthia	35
3.5	Congenital Megaprepuce (CM)	36
3.6	Preputial Lymphedema	38
3.7	Preputial Cysts and Smegma	39
3.8	Balanitis Xerotica Obliterans (BXO)	39
	References	41

4	Scrotum	43
4.1	Normal Anatomy and Histology	43
4.2	Variant of Inguinal Hernia	43
4.2.1	Littre's Hernia	43
4.2.2	Amyand's Hernia	45
4.2.3	Richter's Hernia	46
4.3	Variant of Hydrocele	47
4.3.1	Hydrocele of the Canal of Nuck	47
4.3.2	Abdominoscrotal Hydrocele (ASH)	48
4.3.3	Meconium Hydrocele	49
4.4	Ectopic Scrotum (ES)	49
4.5	Accessory Scrotum	50
4.6	Bifid Scrotum (Scrotal Bifida)	51
4.7	Scrotal Hypoplasia	53
4.8	Scrotal Agenesis	54
4.9	Scrotochisis	55
4.10	Epidermal Cyst	55
4.11	Pigmented Epidermal Cysts	56
4.12	Scrotal Lithiasis and Scrotal Calcinosis	57
	References	58
5	Testis	61
5.1	Testicular Development	61
5.2	Variant of Undescended Testicle	61
5.2.1	Ascending Testis	61
5.2.2	Ectopic Testis	63
5.2.3	Crossed Testicular Ectopia	65
5.2.4	Anorchia and Monorchia	66
5.2.5	Microorchidism	68
5.3	Supernumerary Testicles	68
5.4	Splenogonadal Fusion	70
5.5	Persistent Müllerian Duct Syndrome (PMDS)	72
5.6	Vas Deferens Anomalies	75
5.6.1	Agenesis of the Vas Deferens	75
5.6.2	Duplication of the Vas Deferens	76
5.7	Epididymal Congenital Anomalies	77
5.7.1	Anomalies of Epididymal Attachment	77
5.7.2	Epididymal Cyst (EC)	77
5.7.3	Appendages of Morgagni and its Torsion	79
	References	80

Part II Urinary Bladder and Urethra

6	Urinary Bladder	85
6.1	Bladder Agenesis	85
6.2	Bladder Duplication	86
6.3	Bladder Diverticulum	88

6.4	Variant of Bladder Exstrophy	90
6.5	Variant of Cloacal Exstrophy.....	93
6.6	Congenital Megacystis.....	94
6.7	Urachal Abnormalities.....	95
6.7.1	Patent Urachus	96
6.7.2	Urachal Cyst.....	97
6.7.3	Urachal Diverticulum.....	98
6.7.4	Urachal Sinus.....	99
6.7.5	Malignancy of Urachus	99
6.8	Bladder Neck.....	100
6.8.1	Variant of Posterior Urethral Valve	100
6.8.2	Anterior Urethral Valves (AUV)	100
	References	102
7	Urethral Anomalies	105
7.1	Embryology and Anatomy	105
7.2	Variant of Hypospadias	106
7.2.1	Chordee Without Hypospadias (CWH)	106
7.2.2	Megameatus Intact Prepuce (MIP) Hypospadias Variant	108
7.3	Epispadias	108
7.3.1	Female Epispadias	111
7.4	Megalourethra	112
7.5	Urethral Duplication	114
7.6	Congenital Urethral Fistula	117
7.6.1	Congenital Anterior Penile Isolated Urethrocutaneous Fistula	118
7.6.2	Congenital Urethral Hypoplasia with Urethral Fistula Without Imperforate Anus.....	119
7.7	Urethral Diverticulum	120
7.8	Urethral Agenesis	122
7.9	Urethral Cyst	122
7.9.1	Parameatal Urethral Cyst	122
7.9.2	Prostatic Utricle Cyst.....	123
7.9.3	Skene's Duct Cysts	125
7.9.4	Periurethral Cyst	125
7.10	Urethral Polyp	125
7.10.1	Ectopic Prostatic Tissue and Prostatic Urethral Polyp	126
7.11	Congenital Urethral Stricture	127
7.12	Lacuna Magna (LM)	128
	References	130
8	Female Genital Anomalies, Clitoris	133
8.1	Clitoris.....	134
8.2	Clitoral Anomalies.....	135
8.2.1	Clitoromegaly.....	135
8.2.2	Pseudohermaphroditism.....	137

8.3	Clitoral Agenesis	138
8.4	Bifid Clitoris	139
8.5	Clitoral Duplication	139
8.6	Clitoral Cyst.	139
8.7	Accessory Phallic Urethra	140
8.8	Clitoral Hood Anomalies	142
	References	143

Part III Female Anomalies

9	Labial Anomalies	147
9.1	Anatomical and Embryological Background	147
9.2	Labial Hypertrophy	148
9.3	Labial Hypoplasia	149
9.4	Labial Fusion	150
9.5	Ectopic Labium	151
9.6	Labial Cyst.	151
9.6.1	Mucous Cysts.	152
9.6.2	Epidermal Inclusion Cyst (EIC).	152
9.6.3	Bartholin's Cyst	153
9.6.4	Gartner's Duct Cysts	154
9.6.5	Hydrocele of the Canal of Nuck	155
9.7	Vascular Malformations of the Labia	155
	References	156
10	Hymen	159
10.1	Embryological Background.	159
10.2	Variation in Hymenal Development	160
10.3	Imperforate Hymen (IH)	160
10.3.1	Microperforate Hymen.	162
10.3.2	Hydrocolpos	162
10.3.3	Pyocolpos	163
10.3.4	Hydrometrocolpos	163
10.3.5	Hematocolpos	164
10.4	Vaginal Septa	166
10.4.1	Transverse Vaginal Septa (TVS)	166
10.4.2	Longitudinal Vaginal Septum	168
10.5	Hymenal Polyp and Tags	168
	References	169
11	Female Urethra	171
11.1	Urethral Prolapse	171
11.2	Urethral Caruncle	173
11.3	Urethral Polyps	174
11.4	Prolapsed Ectopic Ureterocele	174
11.4.1	Clinical Picture.	174
11.5	Paraurethral Cyst	175
11.5.1	Skene's Duct Cyst	175

11.6	Urethral Diverticulum (UD)	177
11.7	Persistent Urogenital Sinus (UGS)	178
	References	181
12	Vagina	183
12.1	Vaginal Atresia (VA)	183
12.2	Vaginal Polyps	185
12.3	Vaginal Sarcoma Botryoides	186
12.4	Vaginal Fistula	187
12.4.1	Rectovaginal Fistula	188
12.4.2	Vesicovaginal Fistula (VVF)	188
12.4.3	Vaginourethral Fistula	189
12.5	Ectopic Ureter in the Vagina	189
	References	191
13	Uterus	193
13.1	Embryological Background	193
13.2	Müllerian Duct Anomalies (MDA)	195
13.2.1	Müllerian Duct Agenesis (Müllerian Aplasia)	198
13.2.2	Classification	198
13.2.3	Incomplete Müllerian Fusion	199
13.2.4	Anomalies of Lateral Fusion of Müllerian Ducts	199
13.2.5	Partial Fusion Anomalies	200
13.2.6	Duplication of Müllerian System	201
13.3	Neonatal Uterine Prolapse	201
13.4	Cervical Anomalies	203
13.4.1	Atresia of the Cervix	203
13.5	Fallopian Tube Anomalies	204
13.5.1	Tubal Anomalies	204
13.5.2	Paratubal Cysts	206
13.5.3	Benign Tubal Tumors	206
	References	207
14	Ovary	209
14.1	Ovarian Agenesis	209
14.2	Ovarian Dysgenesis	211
14.2.1	Diagnosis	212
14.3	Ovarian Cyst	213
14.3.1	Fetal Ovarian Cyst (FOC)	213
14.3.2	Neonatal Ovarian Cysts	215
14.3.3	Ovarian Dermoid Cyst (DC)	218
14.4	Congenital Ovarian Tumors	219
14.4.1	Ovarian Germ Cell Tumors (OGCTs)	219
14.4.2	Overview of Ovarian Dysgerminomas	220
14.4.3	Malignant Ovarian Masses	220
14.4.4	Secondary Ovarian Neoplasms	221
14.5	Undescended and Maldescent Ovary	221
14.6	Ectopic Ovary	223

14.7	Supernumerary Ovary	223
14.8	Ovotestes Anomalies	224
14.8.1	Persisting Mesonephric Duct.	225
	References	225

Part IV Caudal Regression Syndrome

15	Caudal Regression Syndrome and Sacral Agenesis	231
	References	235