Urology CHEAT SHEETS

SYNDROMES THAT IMPACT GENITOURINARY SYSTEM I

SYNDROME	GENETICS/CAUSE	MANIFESTATIONS
ALPORT	X linked DOMINANT Mutation collagen IV	**Glomerulonephritis, hematuria**, sensorineural hearing loss, lenticonus (vision problems)
BECKWITH-WIEDEMANN	Congenital WT2 (11p15) Overgrowth disorder	Macrosomia, macroglossia, hemihyperplasia, omphalocele, visceromegaly, hypoglycemia. **15%medullaty sponge kidney.** **Wilms tumors**, rhabdomyosarcoma, hepatoblastoma
BEHCET	Immune-mediated vasculitis	Aphthous ulcers, **genital ulcers**, uveitis Treatment: immunosuppressors and corticosteroids
BERDON	Autosomal recessive Bad prognosis	Megacystis-microcolon-intestinal hypoperistalsis **Dilated bladder**
BIRT-HOGG DUBE	Mutation folliculin gene 17p	**Renal cell carcinoma (chromophobe), oncocytoma** Cutaneous fibromas, pulmonary cysts, spont. pneumothorax
DENYS DRASH	Defect WT1 (11p13)	**Wilms tumor, gonadal dysgenesis,** **Nephropathy** (diffuse mesangial sclerosis)
EHLERS DANLOS	Congenital collagen disorder	Skin and joint laxity, hypotonia, hernias **Bladder diverticulum**
ELSBERG	Herpes Simplex Virus 2	**Acute urinary retention** Radiculomyelitis, motor dysfunction. Self-limited
FOWLER	Associated polycystic ovarian syndrome	Young woman. **Urinary retention**, inability to relax external sphincter. No neurologic problem
GOUVERNEUR		**Vesicoenteric fistula** (pain, urinary symptoms) Pneumaturia, fecaluria UTI)
HINMANS		**Non neurogenic neurogenic bladder** Learned, voluntary dyssynergia. **Obstruction symptoms** Treatment: Anticholinergics, biofeedback
KALMANN	Congenital	**Hypogonadotropic hypogonadism**, anosmia ↓FSH, LH, testosterone
KARTAGENER	Autosomal recessive	Situs Inversus, sinusitis, bronchiectasias **Immotile spermatozoids, infertility**---IVF.ICSI
KLINEFELTER	47 XXY	Primary testicular failure, tubules degeneration, **azoospermia** **Hypergonadotropic hypogonadism** (↑FSH/LH, ↓testostero) Tall, eunuchoid habitus, breast cancer
LERICHE	Atherosclerosis aorta and iliacs arteries	**Erectile dysfunction**, claudication, ↓femoral pulses Treatment: bypass
LESCH NYHAN	X recessive Enzyme HGPRT	Hyperuricemia, hyperuricosuria, **uric acid calculi**, self mutilation
LOIN PAIN	Unknown	Young women, **recurrent flank pain and hematuria**
MAYER ROKITANSKY-KUSTER-HAUSER	46XX defect WNT4 gene (1p). Absent Mullerian Ducts	Normal external female genitalia, absent uterus and fallopian tubes. Primary amenorrhea. **Associated renal anomalies**

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