Urology
CHEAT SHEETS

SYNDROMES THAT IMPACT GENITOURINARY SYSTEM II

SYNDROME	GENETICS/CAUSE	MANIFESTATIONS
MULTIPLE ENDOCRINE NEOPLASIA (MEN)	-MEN1 (chromosome 11, type 1) -RET (chromosome 10, type 2)	**MEN 1 (Werner):** pituitary adenoma (prolactin, **ED**), parathyroid hyperplasia **(calcium kidney stones)**, pancreatic tumor **MEN 2a (Sipple): Pheochromocytoma**, medullary thyroid carcinoma, parathyroid hyperplasia/adenoma **(calcium stones)** **MEN 2b: Pheochromocytoma**, medullary thyroid carcinoma, gastrointestinal and mucosal neuroma, marfanoid features
NELSON	Pituitary adenoma after bilateral adrenalectomy	↑ACTH (no negative feedback) Hiperpigmentation, headaches, visual changes
NOONAN "male Turner sd"	Somatic chromosome defect. Autosomal dominant	Sort stature, shied chest,webbed neck,cognitive problems,heart disease,hypertelorism, bleeding problems, skeletal,**cryptorchidism** **Normal FSH/LF, testosterone and strogen**
OVARIC VEIN		Ureter compression by a dilated ovarian vein (**hydroureter**) TX: Vein embolitation/ligation
PRUNE BELLY (Eagle-Barrett)	Unknown Can occur with trisomy 18 and 21	**Cryptorchidism,** abdominal wall musculature defect**, urinary tract anomalies** (renal dysplasia, hydroureter, dilated prostatic urethra)
REITER	Autoimmune. Non-gonococcal urethritis Associated HLA-B27	**Balanitis circinata. Conjunctivitis, arthritis.** **TX: Doxycycline, NSAID**
SHEEHAN		Post-partum hypopituitarism (necrosis / hypoperfusion)
SHY DRAGER	Unknown	Parkinsonism, autonomic dysfunction, orthostatic hypotension **Bladder overactivity, poor compliance, sphincter denervation**
STAUFFER		**RCC**.Paraneoplastic.Abnormal liver fx test without metastasis
SWYER	Mutation SRY 46XY	**Phenotypically female. Complete gonadal dysgenesis. No testosterone** (no Wolffian struct. , no male exter.genital. develop.) **No AMH** (Mullerian develop.) **High risk gonadoblastoma: bilateral gonadectomy**
TUBEROUS SCLEROSIS	Autosomal dominant 9q34(TSC1), 16p13(TSC2)	Sebaceous adenoma, seizures, mental retardation, **angiomyolipoma. High risk renal carcinoma**
TURNER	45X0	Premature ovarian failure. Primary amenorrhea, webbed neck, aortic coarctation, **horse-shoe kidney** **Hypergonadotropic hypogonadism** (↑FSH, LH, ↓estrogen)
VACTERL	Mesoderm abnormalities	Vertebral defects, Anal atresia, Cardiac defect, Tracheo-Esophageal fistula, **Renal defects** (horse-shoe kidney, agenesis, hypoplasia), Limb defects
VON HIPPEL LINDAU	Gene VHL 3p 25-26	**RCC (clear cell), pheochromocytoma**, retinal angiomas, hemangioblastoma
WARG	Defect WT1 (11p13)	Wilms, Aniridia, Genitourinary (hypospadias), Retardation (mental)
WATERHOUSE FRIDERICHSEN	N. meningitidis	**Haemorrhagic adrenalitis**
WUNDERLICH	Angiomyolipoma	**Massive retroperitoneal hemorrhage**
YOUNG	Unknown (mercury/ genetic factors)	**Obstructive azoospermia**, bronchiectasis, sinusitis **Thick epididymal secretions**. Tx: IVF/ICSI
ZINNER		**Wolfian anomalies (unilateral renal agenesis, ipsilateral seminal vesicle cyst, and ejaculatory duct obstruction)**

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